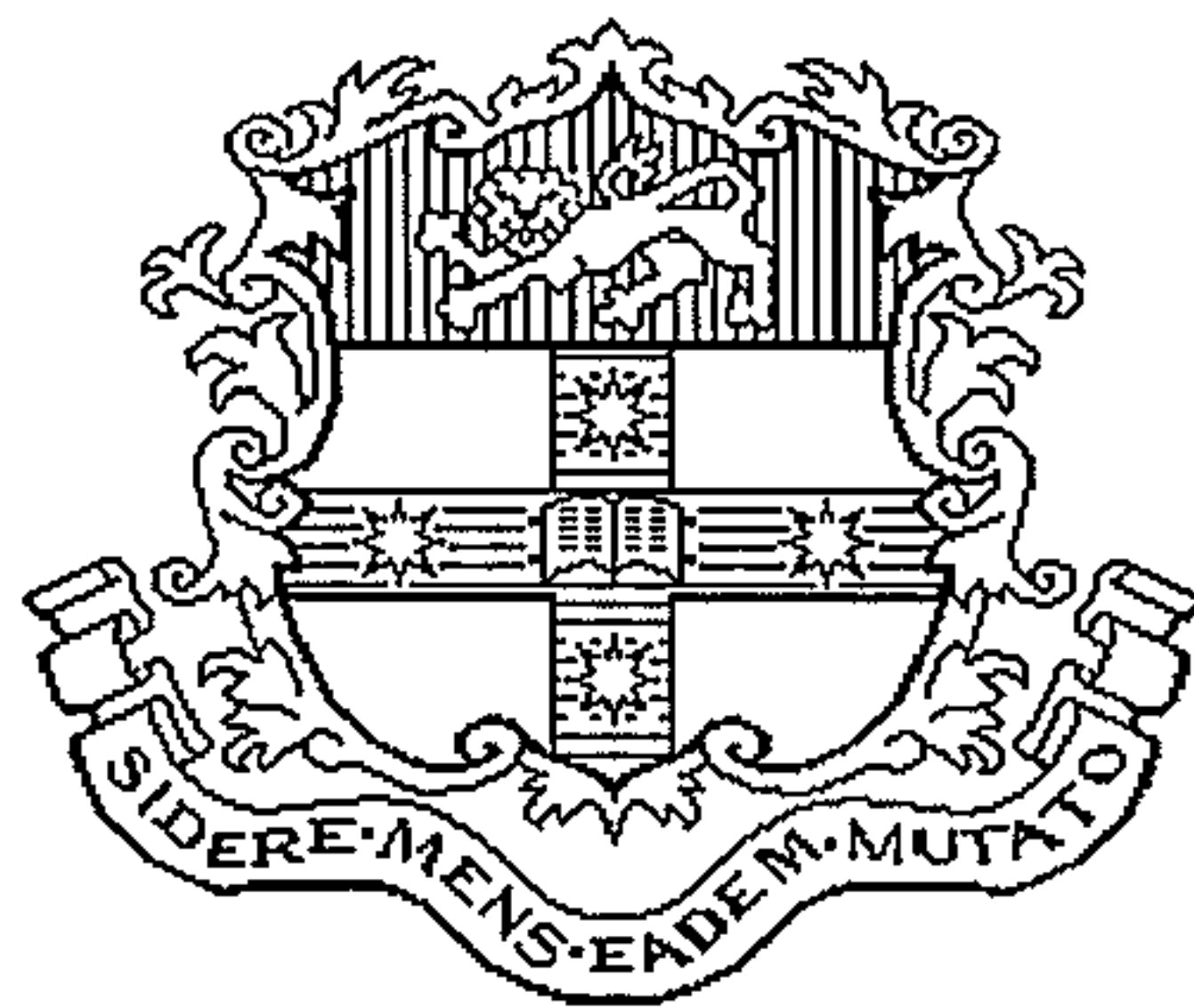




**CRANIOFACIAL FEATURES
OF
PATIENTS WITH
BINDER'S SYNDROME
AND
CHONDRODYSPLASIA PUNCTATA**

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Abstract

Binder's Syndrome and Chondrodysplasia Punctata are two rare and comparable syndromes, producing similar facies in man. However, current literature presents no consensus regarding the nomenclature of these syndromes. The diagnoses currently differ on the presence or absence of stippled immature bone, which may only be present if the patient presents in infancy. These syndromes affect midfacial growth and the aetiology appears to be linked to arrested development of the nasal septum. The majority of studies reported in the dental literature have focussed on hard tissue cephalometric analyses of Binder's Syndrome or Chondrodysplasia Punctata. Few studies have quantitatively analysed soft tissue profile characteristics of these patients, or compared relatives with affected individuals.

In Binder's Syndrome, the nasomaxillary complex is affected, resulting in a short nose, a hypoplastic maxilla, an absent anterior nasal spine in 91 percent of cases, a convex upper lip, a short columella, and an acute nasolabial angle (Horswell *et al.* 1988). The facial appearance of Chondrodysplasia Punctata is similar, however the unique punctate calcification (stippling) in the cartilaginous ends of immature bones, confirms its diagnosis. Chondrodysplasia Punctata has been associated with maternal vitamin K deficiency from exposure during pregnancy to agents such as Warfarin, Phenytoin and alcohol (Sheffield, Halliday and Jensen, 1991; Howe *et al.* 1992a). Genetic forms of the disorder vary in expression, but the majority remain idiopathic.

The punctate calcification in Chondrodysplasia Punctata disappears during the first few years of life as bones mature. Thus, if neonatal radiographs are not available, the older patient may be diagnosed with Binder's Syndrome. Hence, Binder's Syndrome and Chondrodysplasia Punctata may in fact be different names for the same entity (Sheffield *et al.* 1991; Howe *et al.* 1992a).

The aim of the present cephalometric and photographic study was to compare the craniofacial features of patients with a positive diagnosis of Chondrodysplasia Punctata or Binder's Syndrome and healthy individuals. The photographic study also compared the midfacial profile proportions of these patients with their relatives.

Cephalometric radiographs of 11 patients with Chondrodysplasia Punctata or Binder's Syndrome were evaluated using hard and soft tissue analyses. All subjects were matched for age and gender to published norms. Student t-tests were used for statistical analysis of the cephalometric study with the probability level set at $p < 0.05$.

The cephalometric study showed statistically significant findings for the following 12 of a total of 29 hard and soft tissue variables: a reduced anterior cranial base length (S-N), a recessive orbitale position (SNO), a reduced total maxillary

length (PNS-A), a reduced skeletal angle of facial convexity (N-A-Pog), a reduced effective maxillary length (Co-A), an increased mandibular to maxillary differential (Co-Gn to Co-A), an increased lower anterior face height (ANS-Men), an increased upper to lower anterior face height ratio, and from the Holdaway (1983a) analysis: a reduced nose prominence, an increased upper lip thickness and upper lip “strain”, and a prominent lower lip to ‘H’-line.

Life-size photographs of the facial profiles of 40 subjects (13 male patients, 10 female patients, six fathers and 11 mothers) were assessed using three midfacial soft tissue ratio measurements derived from published anthropometric norms (Farkas, 1981). Student t-tests were used for statistical analysis between the sample groups and the norms, with the probability level set at $p < 0.05$. There were statistically significant differences between the patient groups and normal values in the third ratio (Sn-C to Sn-Prn), which may indicate either that the columella length (Sn-C) in the patient group was shorter, or that there was a shorter nasal tip protrusion (Sn-Prn), or various combinations of these factors. The parent groups (six fathers and 11 mothers) had ratio values that were in the normal range.

The photographic study also evaluated the presence of measurable similarity of facial profile between 21 pairs of patients and their parents for these three ratios. Pearson’s Correlation Coefficients showed no correlation between parents and patients for any of the three ratio measurements evaluated.

The hard tissue cephalometric observations support findings in the current literature (Olow-Nordenram, Sjöberg and Thilander 1986; Horswell *et al.* 1988), that the nasomaxillary complex is affected in Binder’s Syndrome and Chondrodysplasia Punctata.

The soft tissue cephalometric analysis confirmed the presence of nasal retrusion and upper lip thickness due to the unique convex lip morphology. The lower lip prominence reflected an underlying Class III skeletal relationship, which was most often due to maxillary retrusion in the present sample. Contrary to findings by Horswell *et al.* (1988), there was a normal nasolabial angle in 55 percent of the sample.

The photographic study showed no similarity between parents and their children or between siblings, for the three ratios measured. This finding does not support studies which stated that the unusual facies was genetically inherited (Fraser and Scriver, 1954), and suggests that the individuals studied had significantly different midfacial morphology from normal subjects and their relatives.

Future research should aim at coordinated multi-centre studies to overcome deficiencies in sample size.

Declaration

This is to certify that the work presented in this thesis was carried out by the candidate in the Discipline of Orthodontics, University of Sydney, and has not been submitted to any other university or institution for a higher degree.

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Dedication

To Him who is called: “...Wonderful, Counsellor,
Mighty God, Everlasting Father,
Prince of Peace.” (Isaiah 9:6)

and

To my husband, and my parents
for their love.

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INTRODUCTION

Children with a short, flat-bridged nose, a short columella, an acute nasolabial angle, perialar flatness, a convex upper lip and a tendency to an Angle Class III malocclusion have been recognised for a long time as having a syndrome often requiring maxillofacial surgery and orthodontics. There is little overall agreement in the literature as to the classification of these patients, due to the variation in severity of individual cases, the particular history, the age of the patient at initial presentation, and the special area of interest of the research group.

As authors concentrated on different aspects of the Syndrome, a number of different names have been used, contributing to the unfortunate confusion of nomenclature (Becker *et al.* 1975). Some maintain the use of different names: either Binder's Syndrome (BS) or Chondrodysplasia Punctata (CDP), depending on the findings, whilst others prefer the blanket term of Chondrodysplasia Punctata and then proceed to subdivide them. As more information came to hand of this disorder, researchers have moved from descriptions of the unusual features to actually attempting to find out its complex aetiology and the location of the genetic mutation. To date, no published frequency or incidence data on either Binder's Syndrome or Chondrodysplasia Punctata is available for any given population.

Historically in 1962, Binder, as cited by Olow-Nordenram and Rådberg (1984), described three patients with the Syndrome as being the result of arhinencephalic dysplasia and called it "maxillonasal dysostosis". However, it became known as "Binder's Syndrome" and has also been variously described as "maxillonasal dysplasia", "hypoplasia of the middle third of the face", "nasomaxillary dysostosis", "midfacial retrusion", "congenital absence of the anterior nasal spine", "dish face", and "scaphoid face."

The term CDP is, in fact, a name given to a characteristic bone dysplasia with a number of subcategories. According to Becker *et al.* (1975), CDP was first described in 1914 by Conradi, who, considered it a form of chondrodystrophia fetalis hypoplastica. Since then, other names, such as "Conradi disease", "Conradi-Hunermann disease", "chondrodystrophia calcificans congenita", "dysplasia epiphysialis punctata", "chondrodystrophia fetalis calcarea", "chondroangiopathia calcerea seu punctata", and "congenital stippled epiphyses" have been used.

Individuals from all these groups may present as children or adolescents seeking orthodontic correction for their Class III malocclusion and/or plastic surgeons to correct their facial dysmorphism. It is important for these professional groups to obtain careful medical histories, including that of maternal pregnancy, in order to provide further information on the aetiology of these conditions.

Despite the presence of malocclusions among patients with Chondrodysplasia Punctata, very little is found in the orthodontic literature. Generally, plastic surgeons have paid much more attention to Binder's Syndrome or Chondrodysplasia Punctata, due to their interest in presenting different methods of surgically correcting the nasal deformity or the orthognathic surgical correction of the midfacial retrusion.

Geneticists, orthodontists, maxillofacial surgeons and plastic surgeons are often faced with the question: "How badly affected are these patients in their facial appearance in comparison to the 'norm' and can this be quantified?" In view of the lack of comprehensive knowledge of this condition, research which provide definitive data will gradually lead to a more scientific basis for understanding BS and CDP.

Clearly, multidisciplinary research and collaborative data-sharing are urgently required for a better understanding of these conditions, so that a management approach addressing preventive as well as corrective measures can be adopted.

**SECTION I:
LITERATURE REVIEW ON BINDER'S
SYNDROME AND CHONDRODYSPLASIA
PUNCTATA**

CHAPTER 1: BINDER'S SYNDROME AND CHONDRODYSPLASIA PUNCTATA

1.1 Classification

Binder's Syndrome primarily affects the nasomaxillary complex, resulting in a short nose, a hypoplastic maxilla, an absent anterior nasal spine in 91 percent of cases, a convex upper lip, a short columella and an acute nasolabial angle. There are associated cervicospinal malformations, hypoplastic frontal sinuses, and retarded development (Delaire *et al.* 1980; Jackson, Moos and Sharpe, 1981; Munro, Sinclair and Rudd, 1979; Horswell *et al.* 1988). In the literature, the varying nomenclature of this Syndrome, such as "hypoplasia of the middle third of the face", "nasomaxillary dysostosis", "midfacial retrusion", "congenital absence of the anterior nasal spine" and "dish face", adds to much of the confusion over its aetiology.

Chondrodysplasia Punctata (CDP) is a heterogeneous group of bone dysplasias that presents with similar facies to Binder's Syndrome (BS). The common characteristic of subgroups of CDP is the radiological finding of punctate calcifications (stippling) in the cartilaginous ends of some of the immature bones, which confirms the diagnosis.

1.2 Differential Diagnosis and Clinical Presentation

Chondrodysplasia Punctata has several distinct genetic forms. In 1971, Spranger, Opitz and Bidder reviewed cases from the Kiel registry of bone dysplasias, placing CDP into two major types, the lethal autosomal-recessive rhizomelic type (RCDP) and the mild Conradi-Hunermann type with autosomal-dominant inheritance (Table 1.1).

According to Becker *et al.* (1975) the diagnosis of CDP should be suspected in patients with a flattened nose, hypertelorism, frontal bossing, high-arched palate, short neck, and short stature. Less common signs mentioned by Tasker *et al.* (1970) were rhizomelia, micromelia, or both; flexion contracture, congenital dislocation of the hip, cataracts, optic atrophy, varying degrees of psychomotor retardation, skin rash, renal anomalies and congenital heart disease.

The differential diagnosis of CDP, according to Becker *et al.* (1975) includes : multiple epiphyseal dysplasia, trisomy 18, infection, cretinism, Zellweger cerebrohepatorenal syndrome, anencephaly, Down's syndrome and G_{M1} gangliosidosis. Interestingly, Becker *et al.* (1975) did not mention BS even as a differential diagnosis for CDP.

Holmström (1986a) gave a brief differential diagnosis for BS, which again is in essence a list of other forms of underdevelopment of the middle third of the face: general maxillary hypoplasia, Crouzon’s syndrome, Apert’s syndrome, sequelae of cleft lip and palate and post-traumatic midfacial retrusion.

Horswell *et al.* (1987) gave another list of syndromes with nasomaxillary hypoplasia as an associated finding: acrodysostosis, Bimler microrhine-dysplasia, oro-facial-digital syndrome, Robinow syndrome, Marshall syndrome, Kniest syndrome, foetal Warfarin syndrome, syphilitic nose and foetal alcohol syndrome.

Table 1.1: Differential diagnosis of Chondrodysplasia Punctata types [after Spranger *et al.* 1971].

	<i>Conradi-Hunermann Type</i>	<i>Rhizomelic Type</i>
<i>No. of Patients</i>	64	36
<i>Clinical findings</i>		
<i>Cataracts, %</i>	17.2	72.3
<i>Head circumference</i>	Normal for age	Small for age
<i>Psychomotor retardation</i>	Rare	Frequent
<i>Peculiar facial appearance, %</i>	53.1	44.5
<i>Contractures, %</i>	26.5	61.2
<i>Foot deformities, %</i>	18.7	11.1
<i>Radiological findings</i>		
<i>Severe bilateral shortening of the femora and/or humeri</i>	Absent	Present
<i>Severe metaphyseal changes of femora and/or humeri</i>	Absent	Present
<i>Distribution of lesions</i>	Frequently asymmetric	Mostly symmetric
<i>Prognosis</i>	Good after neonatal period	Death usually in 1st year of life
<i>Histological findings</i>	Normal or mildly affected endochondral bone formation	Severely disturbed endochondral bone formation

In 1984, Heymans *et al.* reported several biochemical abnormalities in Rhizomelic Chondrodysplasia Punctata (RCDP) and assigned it to the newly recognised group of peroxisomal disorders.

Sheffield *et al.* (1989) presented a 20-year report of 103 cases of CDP. Of these, eight cases of rhizomelic CDP were seen and biochemical abnormalities were found in two. There were 21 cases of Conradi-Hunermann CDP, which was a genetically defined subcategory. The most common type (57 cases) was the mild type of CDP, of which nine had Phenytoin exposure during pregnancy and three had Warfarin embryopathy. Many of these mild types were being diagnosed as BS when they presented to oral surgeons and orthodontists as adolescents. Thirteen cases were not classifiable, highlighting the need for further classification techniques/criteria.

When Sheffield *et al.* (1989) investigated 103 cases of CDP biochemically, only the rhizomelic type had abnormalities of peroxisomal function. The Conradi-Hunermann type and some mild types were found to have abnormalities related to serum osteocalcin levels. Sheffield *et al.* (1989) suggested that the serum osteocalcin may be a way of separating these two types.

The complexity of classifying CDP is further seen by the genetic and biochemical heterogeneity in patients with the rhizomelic form of CDP alone. A study of 10 cases of RCDP showed mutations in at least two different genes can lead to the clinical phenotype of RCDP (Heikoop *et al.* 1992).

There has been a strong association with CDP and maternal exposure during pregnancy to Warfarin, Phenytoin, and alcohol abuse. The stippling of the cartilaginous ends of immature bones has been associated with maternal vitamin K deficiency (Sheffield *et al.* 1991; Howe *et al.* 1992a). The punctate calcification in CDP, present at birth, disappears as it develops into normal bone in the first few years of life. Therefore, if neonatal radiographs are not available, older patients may be diagnosed as having BS. If the unusual facial features are not investigated, no diagnosis will be made. In many cases, BS and mild forms of CDP may actually represent different names for the same entity (Sheffield *et al.* 1991; Howe *et al.* 1992a).

1.2.1 Case Reports

The earliest case report of maxillonasal dysplasia, now termed BS, described the unusual facial features and ascribed the anomaly to a defective forward development of the midface after birth trauma from forceps delivery (Noyes, 1939). Noyes also noted the absence of the anterior nasal spine but did not comment on it. McLaughlin (1949) ascribed the malformation to the absence of the cartilaginous portion of the nasal septum.

According to Olow-Nordenram and Thilander (1987), the first to give a more detailed description of the defect was Binder (1962). He reported on three cases and recorded six special characteristics: (1) arhinoid face, (2) abnormal position of the nasal bones, (3) maxillonasal hypoplasia with consecutive malocclusion, (4) reduced or absent anterior nasal spine, (5) atrophy of the nasal mucosa and lastly, (6) absence of the frontal sinus (not obligatory).

Later researchers such as Gorlin *et al.* (1976), Munro *et al.* (1979), Rintala and Ranta (1985) and Horswell *et al.* (1987) have also stated the frontal sinus to be hypoplastic.

Horswell *et al.* (1987) have described the clinical appearance, radiographic findings, treatment and aetiological factors in 24 Australian children with maxillonasal dysplasia.

1.2.2 Clinical and Pathological Features

The case reports have described morphological characteristics of BS which are of fundamental importance for correct diagnosis and treatment planning.

Holmström (1986a) studied the clinical and pathological features in 50 Scandinavian patients with BS, whose data were collected over 10 years (25 males and 25 females, aged 8 to 34 years). The diagnosis was based on clinical findings of facial characteristics and on palpation of the base of the nostrils. The pathological features were analysed at operation, when the midfacial region was denuded after intraoral subperiosteal dissection of the maxilla and the lower nasal cavity. Holmström's anatomically detailed observations showed that:

- 1) The alveolar bone sloped into the nasal cavity and the prominent bony crest normally found between the facial skeleton and the nasal cavity was absent.
- 2) The nasal incisure was missing, due to the prenasal fossa (Fig 1.4).
- 3) A crescent-shaped scaphoid depression was found centrally on the inferior border of the piriform aperture forming two low transverse ridges from the nasal septum to the lateral borders. Hölmstrom (1986a) concluded that this depression is the prenasal fossa. According to Hölmstrom (1986a), this fossa was first discovered by Zuckerkandl in 1882. In six percent of the patients the anterior ridge of the fossa was missing and there was a continuous slope from the alveolar process.
- 4) There was a low or absent nasal crest, and a small but not totally missing anterior nasal spine. The hypoplastic anterior nasal spine was different from what is usually claimed to be typical in BS, which is often interpreted as an absent anterior nasal spine as seen on cephalometric radiographs. This highlights the limitations of radiographic evaluation of the nasal floor.
- 5) There are three varieties of "flat noses" : when viewed from the side and from below, the dorsum of the nose may be (i) convex, (ii) undulant or (iii) concave. In addition, the dorsum and columella may be long or short (Fig.1.2).
- 6) The soft tissue morphology of the nose and upper lip region in BS is distinctive: the columella is short, and the upper lip slants backwards so that the angle between the columella and lip is acute (Holmström, 1986a). Viewed from below, these differences are more fully appreciated (Fig. 1.3).

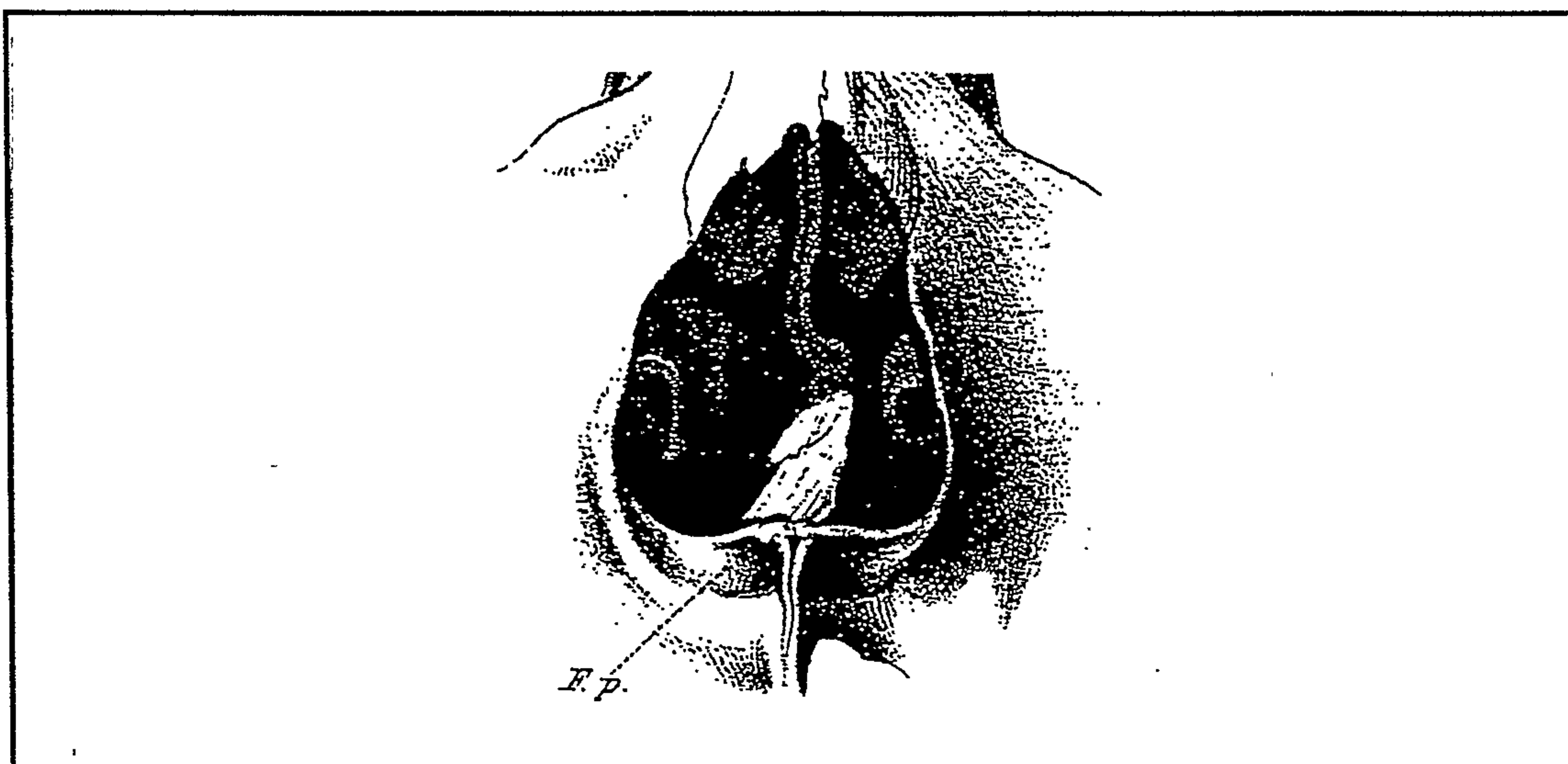


Fig.1.1: Fossa prenasalis (F. p.) in the floor of the piriform aperture. Original illustration by Zuckerkandl in 1882, (from Holmström, 1986a).

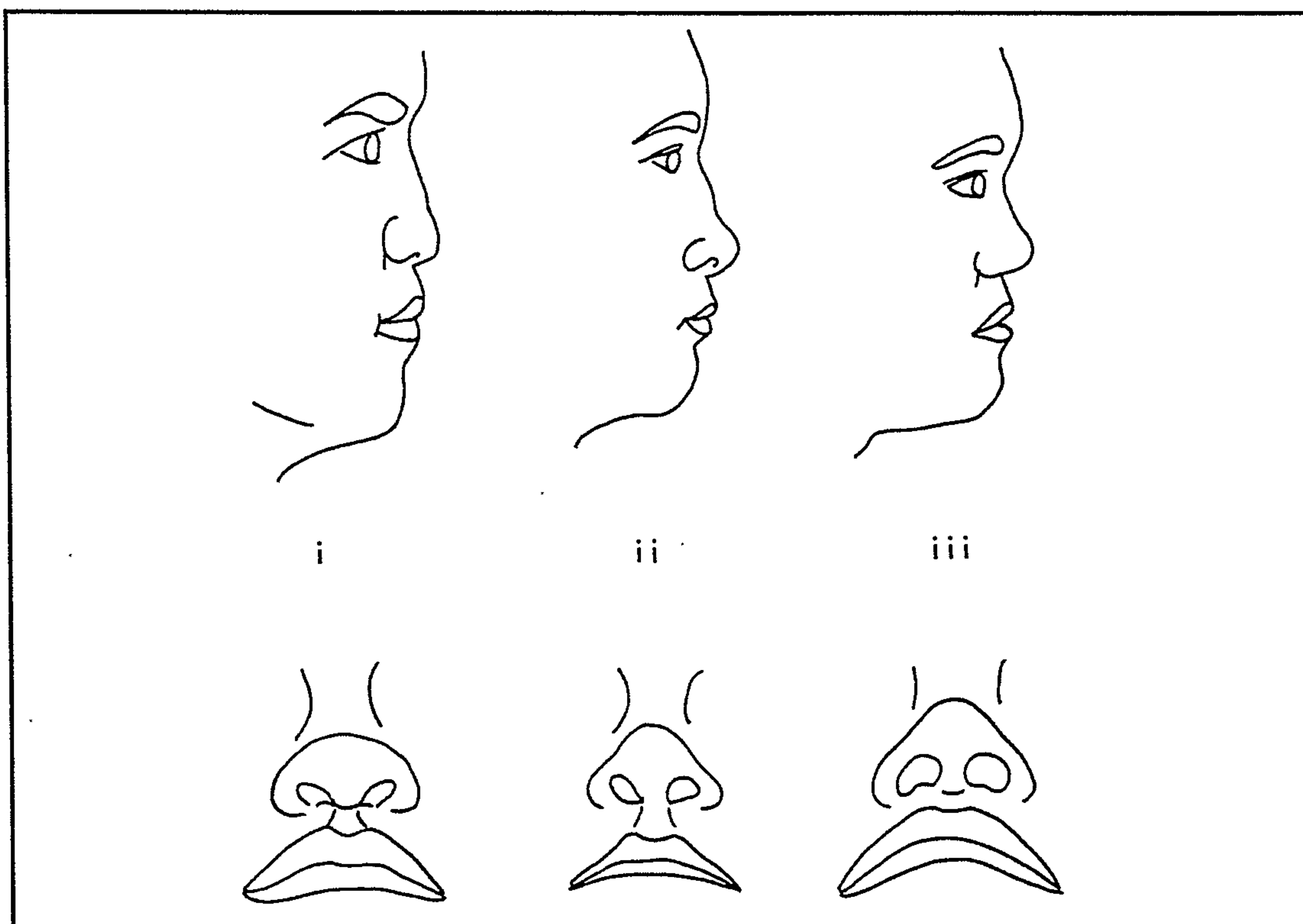


Fig.1.2: The 3 varieties of "flat noses" : when viewed from the side and from below, the dorsum of the nose maybe (i) convex, (ii) undulant or (iii) concave. In addition, the dorsum and columella may be long or short (From Holmström 1986a).

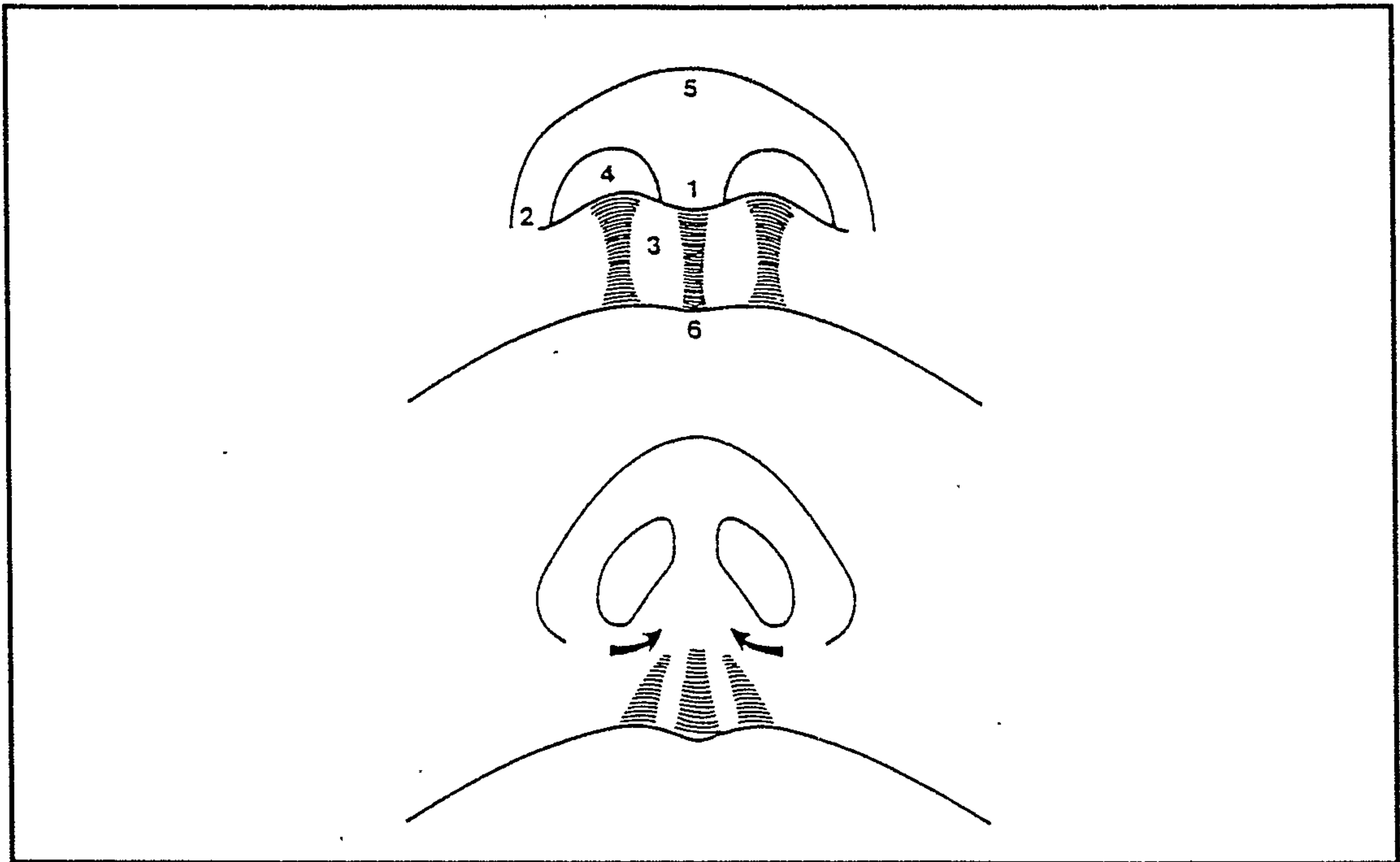


Fig. 1.3: A schematic drawing of the nose and upper lip in Binder's Syndrome seen from below (from Holmström, 1986a).

The normal anatomy is shown below for comparison. Soft tissues converge at the nasal spine. (1) Retracted columella-lip junction and lack of normal triangular flare at the base (produced by the ends of the medial crurae and the anterior nasal spine); (2) perpendicular alar-cheek junction; (3) upper lip is convex with a wide, shallow philtrum; (4) crescent-shaped nostril without nostril sill; (5) low-set and flat nasal tip; and (6) Cupid's bow stretched and shallow.

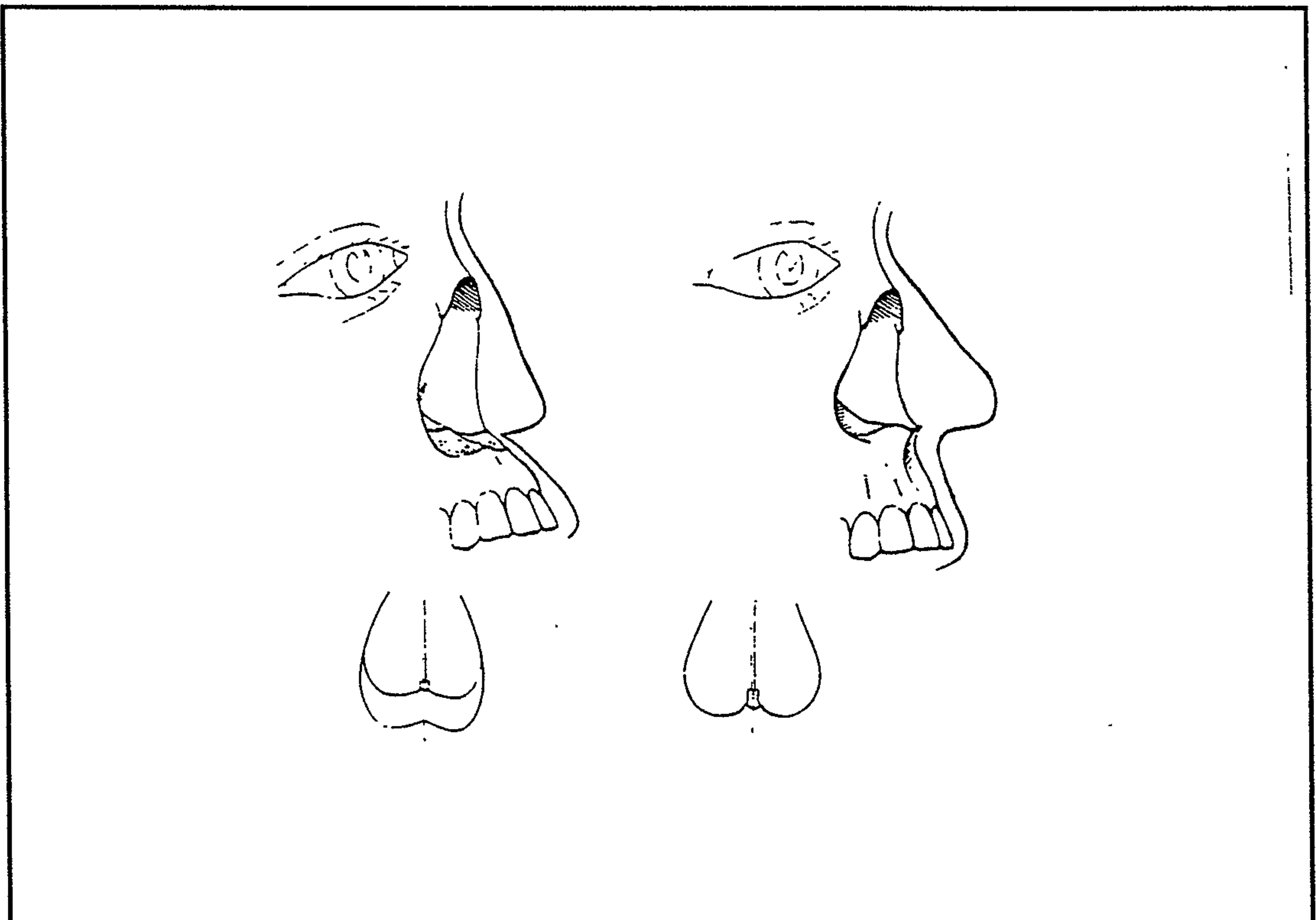


Fig. 1.4: Schematic drawing of the prenasal fossa in Binder's Syndrome. Normal Caucasian morphology is shown on the right. (From Holmström, 1986a).

1.3 Aetiology of Binder's Syndrome and Chondrodysplasia Punctata

Various theories have evolved as to the aetiology of BS and CDP. Some workers believe that BS and CDP represent ends of a larger spectrum of Chondrodysplasias (Sheffield *et al.* 1976), whereas others (Holmström, 1986a) regard them as two separate entities.

Notwithstanding the previous observation as to the different nomenclature of the defect, a general overview of the literature gives the impression of a broad range of severity of the Syndrome, with varying associated anomalies and family histories. There are 4 main theories as to mechanisms that may be acting - either in varying combinations or as isolated factors. These are:

1) Genetic

- Fraser and Scriver (1954), believed CDP to be an autosomal recessive disease and estimated its incidence to be about two per one million births.
- Spranger *et al.* (1971) believe that the Conradi-Hunermann type was a dominant mutation, but that genetic heterogeneity and the influence of environmental factors could not be excluded. The rhizomelic type was believed to be caused by the homozygous state of an abnormal autosomal gene and was a lethal autosomal dominant pattern with variable expression (Horswell *et al.* 1987).

2) Environmental

- Earlier articles claimed it was due to birth trauma (Noyes, 1939; McLaughlin, 1949).

However, teratogens such anticoagulants, anti-epileptic drugs and alcohol have been found to be associated with its pathogenesis in more recent times (Becker *et al.* 1975).

3) Multifactorial / polygenic

- Olow-Nordenram and Valentin (1988) and Horswell *et al.* (1987) believed this to be the case in many instances.

4) Idiopathic

- an explanation given for sporadic cases.

Binder (1962), as cited by Olow-Nordenram and Thilander (1987), had suggested the Syndrome may also be of arhinencephalic origin but of a mild nature. He made the observation that the nasal mucosa was very thin, which, according to Olow-Nordenram and Thilander (1987), has later been confirmed in the literature.

The overall incidence of BS is unknown and cannot be deduced from case reports. Diagnosing the Syndrome at birth is difficult because the involved structures

are not fully developed at that time and because of the resemblance with patients where birth trauma might have occurred (Rival *et al.* 1974, as cited by Olow-Nordenram and Thilander, 1987).

As with BS, the cause of CDP is not known. While many sporadic cases have been reported, there is some evidence of a genetic factor (Fraser and Scriver 1954; Spranger *et al.* 1971).

Some believe it to be an autosomal recessive disease (Fraser and Scriver 1954), while others believe that the Conradi-Hunermann type was probably a dominant mutation (Spranger *et al.* 1971). Nonetheless the genetic heterogeneity and the influence of environmental factors could not be excluded. The rhizomelic type was believed to be caused by the homozygous state of an abnormal autosomal gene and was a lethal condition.

Becker *et al.* (1975) proposed that Warfarin is a possible aetiological factor causing possible foetal haemorrhage and maceration. Evidence from animal models have shown the teratogenic effects of Warfarin. Another factor to be considered is that medications which can potentiate the action of Warfarin (Koch-Weser and Sellers, 1971). If the drug passes through the placental barrier at some critical time during pregnancy and the foetal prothrombin time is substantially lowered, there may be haemorrhages in the developing cartilages leading to calcific lesions in growing cartilage (Becker *et al.* 1975).

Becker's report cited two cases of CDP, and thus could not definitely establish a link between Warfarin and the disease. With the small number of cases involved, it is difficult to state whether this was due to a single gene mutation caused by exposure to the drug. Becker recommended radiographic surveys of infants who had been receiving Warfarin-type anticoagulants during pregnancy, in order to screen for the presence or absence of CDP even in the mild form, and to verify the possible teratogenic link with Warfarin.

1.3.1 The Role of the Premaxilla in Facial Development

The central part of the upper jaw, the premaxilla and its integration into the midfacial bones, has been debated since the 1930's. Despite the disagreement on the pre-maxilla maxillary suture, a true separation into a "premaxillary" and a "maxillary" portion of the upper jaw does not exist at any stage of development. Holmström (1986a) suggested that an inhibition of the "ossification centre" would result in localized hypoplasia of the maxilla, similar to that observed in BS. The incisors were, however, normally developed in all patients, and no signs of midline defects in the central lip were observed in Holmström's study (1986a).

Associated malformations of the cervical vertebrae in about half the patients with BS have been reported from radiological studies (Resche *et al.* 1980; Olow-

Nordenram and Rådberg, 1984). Holmström (1986a) noted six percent of subjects in his study had thoracolumbar spine deviations which required orthopaedic therapy, leading to the suspicion that BS is caused by an unknown agent that acts in the same period as the differentiation of vertebrae. Simultaneous induction of malformations of the spine and BS during week five to week six is developmentally possible. However, Holmström maintained that an arhinencephalic malformation is the result of a disturbance of the enlargement and proliferation of the median nasal prominence as this occurs too early, to have any influence on vertebrae.

Other researchers emphasised that, as in other syndromes, the aetiology must be presumed to be multifactorial (Olow-Nordenram and Thilander, 1987; Langman, 1985; Gorlin and Boggs, 1977).

1.3.2 The Role of the Nasal Septum in Facial Development

In order to appreciate the theories of growth control in midfacial development, one must first look at the development of the chondrocranium in foetal development.

The skeletal elements that form the skull of the foetus develop initially in support of the brain, yet others appear very early in the rapidly developing face as well. The brain is given support by cartilages forming along the cranial base, the chondrocranial elements, whereas the flat bones of the skull, the neurocranial elements, surround the brain.

The chondrocranium is important to the growing face and supports both areas through the development of a bar of cartilage extending uninterrupted along the midline from the anterior nasal region to the foramen magnum (Fig. 1.5). Anteriorly, this cartilage forms a capsule related to the olfactory nerve endings - the nasal capsule. More posteriorly, the cartilage supports the pituitary; laterally, the otic capsules develop around the middle and internal ear structures; and more posteriorly, it forms the occipital cartilages around the foramen magnum. These cartilages form the cranial base as early as the eighth week and will be transformed mostly into bone, with the future ethmoid bone arising from the nasal capsule, and parts of the sphenoid, temporal, and occipital from the more posterior cartilage.

As each of these bones develop, cartilaginous centres remain between them, forming the cranial base synchondroses. These centres will provide for further growth and expansion of the cranial base. The anteriorly located nasal capsule is a large and important cartilage to the developing face and consists of a medial septum component, the mesethmoid, and two lateral cartilaginous wings (Enlow, 1982).

Until bone formation commences, the nasal capsule is the only skeletal support of the upper face. Lateral and inferior to the cranial base cartilages, ossification centres appear in support of these parts of the face as it begins to develop in width during the prenatal period. The nasal, premaxillary, maxillary,

lacrimal, zygomatic, palatine, and temporal ossification centres appear and expand until they appear as bones separated only by sutures (Enlow, 1982).

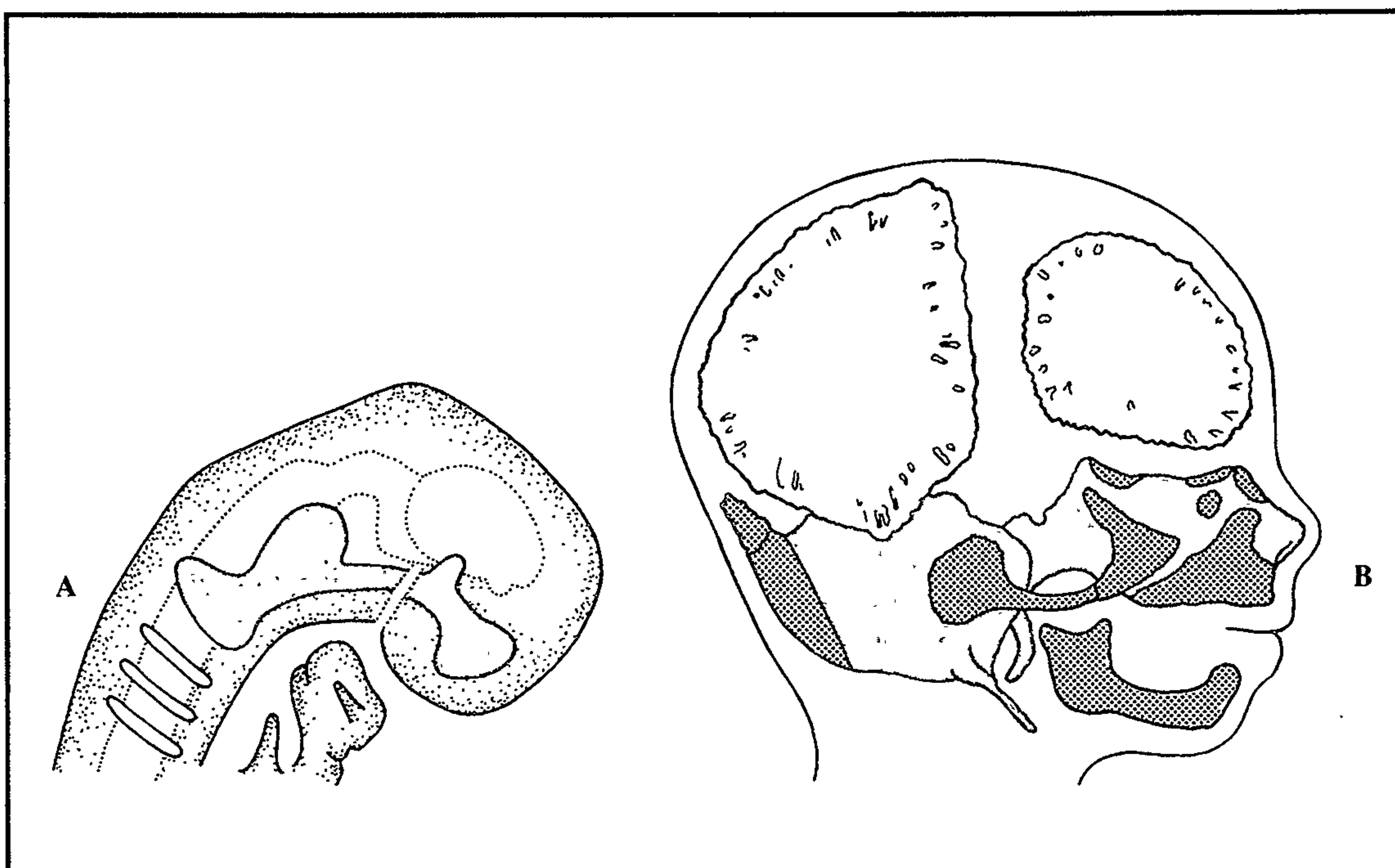


Fig. 1.5: Development and maturation of the chondrocranium:

(cartilage: pink; bone: stippled red).

A: Diagram of 8 week-old human fetus. Note that an essentially solid bar of cartilage extends from the nasal capsule anteriorly through to the occipital area posteriorly.

B: Skeletal development of human at 12 weeks. Ossification centres have appeared in the midline cartilage structures, and, in addition, intramembranous bone formation of the jaws and brain case has begun. From this point on, bone replaces cartilage of the original chondrocranium rapidly, so that only the small cartilaginous synchondroses connecting the bones of the cranial base remain.

[From Proffit WR, Fields HW *et al.* Contemporary Orthodontics, 2nd Edition, Mosby -Year Book 1993, page 30, figure 2-18].

There are two major theories on the mechanisms governing pre- and postnatal growth and development of the human midface, differing in opinion as to whether the nasal septum plays an active or a passive role. The first theory was proposed by Scott (1953), who suggested that the cartilaginous nasal septum is a primary growth centre which pushes the midfacial bones downwards and forwards relative to the cranial base. Latham (1970), from a histological study of human fetuses, reported the presence of a “septopremaxillary ligament” which was a fibrous ligament passing from the anteroinferior border of the nasal septum posteroinferiorly to the anterior nasal spine and the intermaxillary suture of the premaxillary region. Latham concluded that this ligament was a means by which the growing nasal septum might exert a downward and forward traction upon the bones of the upper jaw.

The second theory (Moss *et al.* 1968) using the hypothesis of boxed structures (Badoux, 1966) views the septal cartilage as only a strut in the nasal cavity without a primary morphogenetic role. Moss proposed that in line with his theory of functional matrices, the growth of the nasal septum is secondary and compensatory to expansion of the nasal cavity due to increased functional demands for an increased respiratory volume.

There seem to be numerous data that support the contention that the cartilaginous nasal septum plays an important role in midfacial translative growth. On the other hand, Moss' functional matrix hypothesis attributes midfacial translatory growth to the expansion of spaces, rather than to cartilaginous growth. These two hypotheses are mutually exclusive.

However, the lack of controlled experimental data to support an active role of spaces may well be related to the absence in Moss' theory, of a well-defined physical mechanism that can be tested.

Past attempts to test these theories centred around clinical studies of children with midfacial clefts, traumatic or congenital absence of the nasal septum, or experimental animal studies involving partial or complete nasal septal ablation. Results have been inconclusive mainly due to failure in isolating the nasal septal damage from damage and effects on adjacent structures. For instance, although clefting may separate the septum from the palatal processes of the maxilla, the nasal capsule is still continuous superiorly and posteriorly. Similarly in cases of so-called nasal septal cartilage agenesis there would have been prenatal continuity of the nasal capsule. The animal resection results have been criticised by Moss (1976) and Koski (1968) who concluded that factors such as animal size, age of the animal at the time of operation, site, size and composition of the extirpated portion, surgical damage to adjacent structures, nutritional condition after the operation and growth registration procedure underlie the controversial and contradictory results.

The findings of Latham and Burston (1966) on sections of the palate of a nine month old arrhinencephalic child in which the nasal septum was congenitally absent were significant, in that the vertical height of the nasal region was not affected greatly. However, the anteroposterior dimension of the middle third of the face was clearly deficient. Gange and Johnston (1974) described a similar finding in the albino rat model, following electrocautery of the septopremaxillary ligament. In this well controlled animal experiment, the severing of the septopremaxillary ligament was found to retard the forward advancement of the maxillary and palatine bones without decreasing their size. This procedure failed to impair the vertical growth of the nasomaxillary complex. These studies conform closely to predictions generated by the nasal septum theory.

1.3.3 Evidence from an Animal Model of Human Warfarin Embryopathy

Recent research into an animal model of the human Warfarin embryopathy presents strong evidence for an active role for the nasal septum (Howe *et al.* 1991). Warfarin embryopathy in humans, is characterised by severe maxillonasal hypoplasia. In the rat model of this condition the hypoplasia of the midface is accompanied by ectopic calcification and underdevelopment of the nasal septum (Howe *et al.* 1991). These findings are consistent with the inhibitory effects of Warfarin on vitamin K metabolism. Unlike the septal resection studies of Sarnat (1976), this animal model provides a non-invasive method whereby the nasal septum can be disrupted without interference to the surrounding structures. The results of this animal model indicate an active role for the nasal septum in midfacial development.



Fig. 7 – A 3-week control rat (left) compared with a 3-week experimental rat (right) which received warfarin and vitamin K1 each day from postnatal day 1. The experimental rat has a short upturned snout compared with the control.



Fig. 8. – Nasal septa from control (upper) and experimental (lower) 12-week-old rats. The experimental rat received warfarin and vitamin K1 each day from postnatal day 1. The septa have been stained with alizarin to show calcium. The nasal septum from the experimental rat shows large areas of abnormal calcification (arrows). eth = perpendicular plate of the ethmoid.

Fig. 1.6: (a): Animal model of human Warfarin embryopathy (Rat).
 (b): Nasal septa for control and experimental rats. From Howe and Webster, 1992.

Thus, studies of a rat model of the Warfarin embryopathy suggest a pathogenesis for the maxillonasal hypoplasia induced by vitamin K deficiency (Howe and Webster, 1992). In the rat, Warfarin-induced maxillonasal hypoplasia was associated with abnormalities of the nasal septum. The normally cartilaginous part of the septum was heavily calcified and shorter than normal. Hence, in foetuses made vitamin K-deficient by Warfarin, matrix gla-protein would be present in cartilage in a non-functional form and could lead to uncontrolled calcification, leading to decreased longitudinal growth and subsequent maxillonasal hypoplasia. This data points to a critical period which occurs prenatally, towards the end of the first trimester in the human, and postnatally, in the first few weeks after birth in the rat.

1.3.4 Evidence from Human Phenytoin Embryopathy

Prenatal exposure to Phenytoin, either alone or in combination with other anticonvulsants, can result in severe appearance problems later in life requiring orthodontic and plastic surgery. Failure to appreciate the association between maxillonasal hypoplasia and prenatal Phenytoin exposure may lead to inappropriate counselling and great distress to the patient (Howe *et al.* 1992).

Studies by Howe *et al.* (1992) support the observations of Sheffield *et al.* (1976, 1991) that a mild-form of CDP usually presents as Binder's Syndrome in adulthood, regardless of whether the CDP was associated with prenatal Phenytoin exposure or was genetic in origin. Usually no attempt is made to investigate the maternal drug history of Binder's cases as the Syndrome is considered by most health professionals to be 'genetic' in origin (Olow-Nordenram and Thilander, 1987).

1.3.5 Mechanisms of Phenytoin Teratogenesis

There are at least seven theories that attempt to explain the adverse effects of Phenytoin on prenatal development (Howe *et al.* 1994). These are:

- 1) A direct teratogenic effect of products of Phenytoin metabolism (Martz *et al.* 1977),
- 2) The production of teratogenic free radicals during Phenytoin metabolism (Kubow and Wells, 1989),
- 3) Phenytoin-induced folate deficiency (Meadow, 1968),
- 4) Foetal hypoxia secondary to Phenytoin-induced foetal cardio depression (Danielsson *et al.* 1992),
- 5) Phenytoin-induced vitamin K deficiency (Keith and Gallop, 1979),
- 6) A direct effect of seizures (Majewski *et al.* 1981), or
- 7) A developmental effect associated with the genotype for epilepsy (Durner *et al.* 1992).

Clinical and experimental studies suggest that Phenytoin causes maxillonasal hypoplasia by inducing vitamin K-deficiency in the developing embryo.

1.3.6 Vitamin K Deficiency as a Consequence of Phenytoin Exposure

The association between Phenytoin exposure and vitamin K deficiency has been demonstrated in several studies (Keith and Gallop, 1979; Mountain, Hirsch and Gallus, 1970; Bleyer and Skinner, 1976) and the mechanisms described (Deblay *et al.* 1982).

1.3.7 Similarities between Warfarin and Phenytoin Embryopathies

Warfarin is a well established vitamin K antagonist and several authors have drawn attention to the similarities of the facial and skeletal abnormalities (such as epiphysial stippling and distal digital hypoplasia) of the Warfarin and Phenytoin embryopathies (Keith and Gallop, 1979; Hall *et al.* 1980). The similarities suggest that the abnormalities are due to a common deficiency in vitamin K in the foetus.

Nasal and midfacial hypoplasia are very common outcomes of Warfarin use during pregnancy.

The reported prevalence of nasal and midfacial hypoplasia after prenatal exposure to Phenytoin is also likely to be influenced by the method of facial examination and the length of time to follow-up. Very severe cases are easily discernible at birth; such cases are probably rare and related to multiple drug therapy.

Evidence from prospective studies using large sample sizes indicate that many, if not most cases, of mild to moderate maxillonasal hypoplasia are not diagnosed at birth. Hence, the same facial, skeletal and haemorrhagic disorders are seen after prenatal exposure to Phenytoin or Warfarin. Since Warfarin's effect appears to be exclusively through the induction of vitamin K deficiency, it is reasonable to conclude that the effects of Phenytoin on facial and digital development are also the result of vitamin K deficiency. Other signs of Phenytoin teratogenesis have been described but are unlikely to be due to vitamin K deficiency, as they do not occur after Warfarin exposure (Howe *et al.* 1994).

1.3.8 Other Anticonvulsants

Other anticonvulsants may also lead to "Phenytoin facies", as they share common pharmacological pathways, leading to increased incidence of facial dysmorphology and haemorrhage with multiple drug therapy (Howe *et al.* 1994).

1.3.9 Prevention by Vitamin K Supplementation

Warfarin is a potent and intentional cause of vitamin K deficiency and is effective in well nourished individuals. Phenytoin is probably a less potent cause of vitamin K deficiency and only impacts on vitamin K status when there are other predisposing factors. One of these factors appears to be pregnancy.

A number of investigators have proposed that pregnant women on anticonvulsants should receive vitamin K supplementation towards the end of pregnancy in an attempt to prevent haemorrhage in the foetus (Owen *et al.* 1967; Bleyer and Skinner, 1976; Deblay *et al.* 1982; Gimovsky and Petrie, 1986; Moslet and Hansen, 1992; Delgado-Escueta and Janz, 1992; Cornelissen *et al.* 1993a,b). Cornelissen *et al.* (1993a) showed that supplements of 10 mg vitamin K1 per day from 36 weeks gestation had a beneficial effect on women taking anticonvulsants. Delgado-Escueta and Janz (1993), in developing consensus guidelines at a 1990 workshop on anti-epileptic drugs and pregnancy, reported that a majority considered it prudent to administer vitamin K1 (20 mg per day) prophylactically in the last month of pregnancy to protect the infant against vitamin K1 responsive bleeding.

Scientific evidence points to the fact that it may be possible to prevent the disfigurement of midfacial hypoplasia by vitamin K1 supplementation in early pregnancy. The critical time for this supplementation, according to Howe *et al.* (1994), would appear to be from six weeks gestational age.

**SECTION II:
LITERATURE REVIEW ON PATIENT
EVALUATION TECHNIQUES**

CHAPTER 2.

CEPHALOMETRIC EVALUATION

2.1 Cephalometric Analysis

Radiographic evaluation of the skull in orthodontics began in 1931, when Broadbent in the USA and Hofrath in Germany independently devised a standardised system of cephalometric radiography. Subsequent research into facial growth and the results of orthodontic treatment had a major impact on orthodontic theory and practice (Houston, Stephens and Tulley, 1994).

In clinical practice, cephalometric analysis is of value in assessing facial and dentoskeletal relationships as an aid to treatment planning, as well as monitoring changes attributable to growth and treatment. Broadbent (1937) emphasised the three-dimensionality of facial relationships and recommended that postero-anterior (PA) as well as lateral skull views should be obtained. However, cephalometric analysis has largely come to imply the measurement of lateral skull radiographs. This has been attributed in part to the facial variations of greatest orthodontic importance being in the sagittal plane, and in part because other views are difficult to interpret and measure. Few landmarks that are important in a lateral skull radiograph can also be reliably identified in the PA view, thus limiting a three-dimensional analysis utilising this method (Houston, Stephens and Tulley, 1994).

Cephalometric radiographs are to be taken in a standardised manner to permit comparative measurements between patients and for the same patient on different occasions. The head is held in a cephalostat so that the mid-sagittal plane is at a fixed distance from, and parallel to the film. The target of the X-ray tube is also at a fixed distance from the film, with the central ray directed through the ear rods of the cephalostat so that the enlargement at the midsagittal plane is constant. Unfortunately, there has not been agreement on international standards for these distances so that enlargement would be standardised. Provided that the magnification is known, linear enlargement can be compensated for. To this end it is recommended that a metal scale of known length be suspended at the midsagittal plane of the head so that it provides a permanent record of the enlargement of that film (Houston, Stephens and Tulley, 1994).

If the goal of cephalometric analysis is to provide an accurate description of dentofacial relationships, it must be specified exactly what those relationships are of interest. The consideration of the head and face as being made up of five major units aids diagnosis. These units are: (1) the cranium and cranial base, (2) the nasomaxillary complex, (3) the mandible, (4) the maxillary dentition, and (5) the mandibular dentition. The object of diagnosis is to detect and quantify

disproportionate relationships among these craniofacial units, and lateral cephalometric analysis offers a quantitative way of doing this in the antero-posterior and vertical planes of space. The most important result of the cephalometric analysis is a judgement as to the proportionate relationship of the parts, and not the angular or linear measurements *per se*. The present discussion will be concerned with the skeletal measurements, as this is the focus of the cephalometric hard tissue analysis used. The relevant aspects of the anatomy will be mentioned in the description of the landmarks used in Chapter 5. The soft tissue cephalometric criteria will be discussed in Chapters 3 and 5.

2.2 Antero-Posterior Relationship of the Jaws to the Cranium

The orthodontic tradition of Angle emphasised that the position of the maxilla and maxillary dentition was invariant, making the location of mandibular position the key diagnostic element (Angle, 1907). Thus, when Downs (1948) initially developed the cephalometric analysis, he focussed on the relationship of the maxilla to the mandible, which was indirectly linked back to the cranium via the facial angle.

In time, it was clear that Angle had underestimated the variability of the maxillary position relative to the cranial base. In 1952, Riedel suggested the use of the sella-nasion (SN) line and its angular relationships to the anterior maxillary and mandibular landmarks the angles SNA and SNB. He used the ANB angle as an indicator of the relative antero-posterior relationship of the jaws. These measurements are still widely used, however the merits or limitations of ANB remains a subject of debate (Jarvinen, 1986).

The problem of relying on angular measurements to SN as an indicator of the antero-posterior position of the jaws is that it assumes a normal inclination of the anterior cranial base, which is not always the case. Because the inclination of the SN line to true horizontal is so variable, the uncritical acceptance of SNA and SNB can lead to an error in determining which jaw is at fault in a skeletal discrepancy. A method of compensating for this is by using a true horizontal orientation: the Frankfort plane, or better, the true horizontal line from an "Natural Head Position"[NHP] radiograph, and correct the SN line to its normal inclination to true horizontal (Proffit and White, 1991).

McNamara (1984) published a useful and direct way of relating the jaws to a true vertical line (perpendicular to true horizontal) dropped from nasion. McNamara's published data are based on the Frankfort plane rather than on a true horizontal, which was not available for his Michigan growth study reference sample.

For most normal Caucasian populations, the nasion perpendicular line (N-Perp.) passes slightly behind point A and 2 to 3 mm anteriorly to point B. The normal A-B difference (the distance between A and B points when projected to the

true horizontal line) is approximately 4 mm. Thus the measurements to N-Perp. line can establish the antero-posterior relationships of the jaws to the cranium and to each other. An A-B difference greater than 6 mm or less than -4 mm indicates a discrepancy. If there is a discrepancy, one can decide which jaw is at fault by comparing their position to the N-Perp. line.

A jaw discrepancy can be the result of a problem in the size of the jaw or its position. Harvold's (1974) approach is one of the most straightforward in evaluating relative jaw size, which is incorporated into McNamara's analysis. Harvold (1974) used a measurement from the temporomandibular joint 'tm', an averaged point on the contour of the glenoid fossa where the line indicating maximum mandibular length intercepts the contour of the fossa to the anterior nasal spine (inferior point where ANS is 3 mm thick) for the maxillary unit length. For mandibular unit length, he used the measurement from 'tm' to prognathion or 'pgn' a point on the contour of the bony chin close to pogonion, indicating maximum mandibular length measured from the temporomandibular joint. Although the absolute distances are interesting as a reflection of jaw size, the difference in the mandibular and maxillary lengths is more significant. If the mandibular length is more than 30 mm, or 15 mm less than the maxillary length, the discrepancy may be too great for orthodontic correction. In using this approach, it must be kept in mind that the vertical separation of the jaws affects the measured distances.

2.3 Vertical Relationship of the Jaws to the Cranium

Early cephalometric analyses did not address vertical relationships as much as antero-posterior relationships. This deficiency reflects the bias in classical orthodontics toward antero-posterior relationships that was exemplified by the Angle classification. Traditional treatment modalities limited the treatability of the vertical dimension, so it was viewed as untreatable. Surgical-orthodontic treatment has developed to the extent that vertically dysplastic cases can be treated with a greater degree of predictability (Proffit and White, 1991).

Sassouni (1969) contributed to the idea that in a vertically well proportioned face, the horizontal reference planes of the cranial base (SN), Frankfort, palatal, occlusal and mandibular planes project toward an approximate common intersection located near the back of the skull. He showed that if these planes are nearly parallel, so that their point of convergence is well away from the face, anterior and posterior facial heights will be nearly the same. This in turn produces a predisposition toward a deep bite anteriorly. Conversely, if these planes converge just behind the face, there is an open bite predisposition, as anterior facial dimensions will be considerably longer than the posterior ones. He further emphasised that if one part of

the face is vertically disproportionate, the plane through it will not converge with the others.

Measuring contributions of the maxilla and mandible to total facial height is a valuable tool in establishing whether anterior vertical disproportions are primarily in the maxilla or mandible. These measurements must be made along a true vertical line, to avoid errors from angles which tend to increase the distance measured.

Linear measurements from cranial base reference points (sella and ethmoid points) can be useful in establishing the vertical position of the posterior maxilla (PNS) and mandible (Go). Measurements of this type rarely have been included in published cephalometric analyses, despite their importance, and so tabulated data for standards are not widely available. The best source is the Michigan growth study (Riolo *et al.* 1974).

The interpretation of such measurements must be tempered by an appreciation that they should be interpreted primarily in the context of proportional relationships within the same individual. If the upper face height is large, the lower face height must also be, in order to give balance. Likewise, one would expect these proportions to be linked to body stature; that is, a tall person is more likely to have larger upper and lower face height measurements than a short person. If anterior dimensions are large, posterior measurements should also be, to produce a well-proportioned face (Proffit and White, 1991).

2.4 Cross-Sectional Studies of Binder's Syndrome and Chondrodysplasia Punctata

An early study was that of Hopkin (1963), who described five cases where trauma and infection was excluded as a cause for absence of the anterior nasal spine. Radiographically, he found a deficiency in thickness of the maxillary frontal cortical plate of the alveolar bone and a more downward than forward angulation of the nasal bone. There was also a reduction in maxillary length. Speculations were made over the causes of the abnormal development compared with similar, less severe, findings observed in children with bilateral total clefts.

McWilliam and Linder-Aronson (1976) studied 31 children with hypoplasia of the middle third of the face between the ages of eight and 18 years with special reference to their ability to breathe through their nose and normal airflow. Their cephalometric findings showed the eight significant differences: (1) short anterior cranial base, (2) small cranial base angle, (3) reduced sagittal development of the nose, (4) straight profile, (5) retrognathic maxilla, (6) short maxilla, (7) reduced sagittal depth of the nasopharynx, and (8) reduced vertical growth of the maxilla. However, the above two studies did not take sex and age into account and the sample sizes were relatively small. Linder-Aronson (1979) presented a follow up

study that seemed to show that disturbed nasal respiration can affect the facial morphology.

The study by Olow-Nordenram, Sjöberg and Thilander, (1986) was a far more extensive investigation. Their study analysed the craniofacial morphology of 97 Swedish children (51 boys and 46 girls) with BS in a retrospective cross-sectional study. The children studied were all untreated, and came from three different geographical areas. Craniofacial morphology was studied using 36 cephalometric variables and were compared with that of normal Swedish children matched for sex and age. For several of the cephalometric variables, highly significant differences were obtained, describing the distinct craniofacial characteristics of BS. Of particular interest was that significant differences were found in the younger age groups, groups two and three below, indicating a severe form of the Syndrome, with greater need for orthodontic treatment.

The Swedish children were divided into four age groups:

- 1) 6-8 years -Boys (n=17); Girls (n=10) - Total N=27
- 2) 9-11 years -Boys (n=20); Girls (n=15) - Total N=35
- 3) 12-14 years -Boys (n=8); Girls (n=10) - Total N=18
- 4) 15-17 years -Boys (n=6); Girls (n=11) - Total N=17

The three different geographical regions from which Olow-Nordenram, Sjöberg and Thilander, (1986) pooled their sample meant three different magnification factors for their radiographs, which required adjustment. The error of measurement by the three different orthodontists were "negligible". In addition, the point termed 'ss,' the deepest point on the anterior contour of the upper alveolar arch at the level of the upper central incisor or Burstone's 'A' point (1978), had to be specially constructed, because point 'ss' was very difficult to find due to the absence of the anterior nasal spine (Fig. 2.1). The reference line 'NL', the midline through the palate through 'ptm', was also a constructed point for the same reason (Fig. 2.1). Olow-Nordenram, Sjöberg and Thilander, (1986) stated that the error in definition of 'ss' and 'NL' was negligible, and that both the error of the method and of the measurements could be ignored.

More recently, Eliasson and Holmström (1994) described a lateral cephalometric analysis to describe midface prognathism in BS, which they termed the "theoretical midface ratio" or "TMFR". The TMFR, which is independent of the vertical position of point A was described as a valid substitute for SNA, as in BS subjects, point A is difficult to determine. Of the 35 adults with BS studied, they found only less than half had maxillary retrognathia (11 subjects), and the majority (22 subjects), had an orthognathic maxilla and two had a prognathic maxilla. Since their study had a poor description of how these points were measured, their methodology remains obscure.

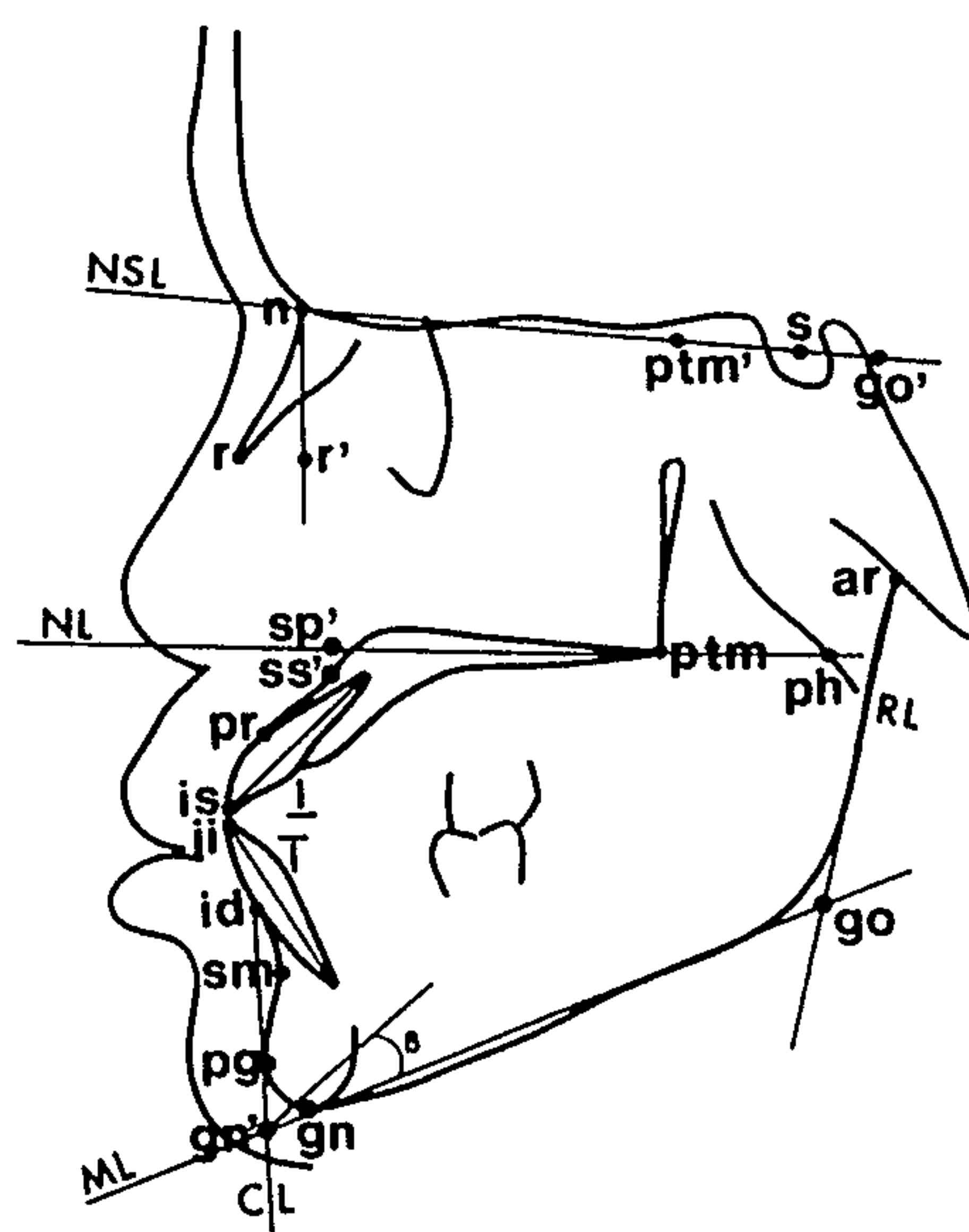


Fig. 4. Special reference points and lines used in this investigation in addition to those introduced by Björk.¹⁴ CL = Chin line, the tangent to the chin through id (gn'-id); gn' = intersection between the chin line (CL) and the mandibular line (ML); go' = projection of gonion on the nasion-sella line (NSL); ptm' = projection of ptm on NSL; ss' = deepest point on the anterior contour of the upper alveolar process at the level of the apices of the central incisors; sp' = projection of ss' on the nasal plane (NL); NL = midline of the palate through ptm; ph = intersection between NL and the contour of the posterior pharyngeal wall; and r = rhinion, the most anteroinferior point on the nasal bone.

Fig. 2.1: Reference points and lines used by Olow-Nordenram in the cephalometric study of Binder's Syndrome.

(From Olow-Nordenram and Thilander, 1989).

Rune *et al.* (1982) discussed the difficulty of analysing the facial morphology by traditional cephalometric methods as the measurement points: nasion, the anterior nasal spine, and subspinale, point A, are missing or displaced. Thus, Delaire and associates, as cited by Rune *et al.* (1982), used the "architectural and structural craniofacial analysis, lateral view" of Delaire to describe the abnormalities of thirty-four patients.

Olow-Nordenram *et al.* (1986) showed that the most characteristic sign was the retrognathic face, especially for the maxilla, resulting in a concave profile. This finding is in agreement with earlier studies (Hopkin 1963; McWilliam and Linder-Aronson 1976). The rest of their findings may be summarised:

- (1) Significantly short anterior cranial base.
- (2) Significantly smaller pharyngeal depth.

- (3) The shortness of the maxilla can be explained by the absence of the anterior nasal spine.
- (4) Due to the above factors (1) and (3), the nasal septum will be in a posterior position, causing retroclination of the nasal bones. Olow-Nordenram *et al.* (1986) defended this speculative statement by citing the experimental studies of Moss *et al.* (1968); Stenström and Thilander (1970 and 1972), concluding that the origin of the absent anterior nasal spine was prenatal, together with retarded growth of the anterior cranial base during early childhood.
- (5) The significantly shorter mandibular body (as compared with the control group) was found in only two age groups between nine and 14 years for both sexes. The high significance of some differences in the lower facial variables possibly indicates posterior rotation of the mandible.
- (6) The proclination of the upper incisors, which is highly significant in all age groups and both sexes, is probably a compensatory effect for the retrognathic and small maxilla. Where compensation of lower incisor inclination also occurred, the interincisal angle was within normal limits.

The most significant differences in the lower face was found in the younger age groups and in particular from nine to 14 years. Olow-Nordenram *et al.* (1986) believed that this could be due to "catch-up" growth with increasing age. In this cross-sectional study, a more plausible explanation could be that the younger age group was referred early, as their malocclusions were more severe due to more marked signs of BS, but the teenagers had little or no need for orthodontics, and did not seek treatment unless surgery was required for aesthetic reasons.

2.5 Longitudinal Studies of Binder's Syndrome and Chondrodysplasia Punctata

Olow-Nordenram and Thilander (1987 and 1989) conducted further investigations on the growth and development of BS children in two longitudinal groups: one with 13 orthodontically untreated children (Olow-Nordenram and Thilander, 1987) and the second study with 15 orthodontically treated patients at corresponding ages (Olow-Nordenram and Thilander, 1989). The untreated children were compared with the same reference material as in their cross sectional study. The study looked at serial headfilms taken from early childhood until growth was 'completed'. It was shown that all linear dimensions changed with age parallel with normal growth and approximately to the same extent, although on another level for some variables.

Specific findings from the untreated longitudinal sample of seven boys and six girls were (Olow-Nordenram and Thilander, 1989):

- 1) **Cranial Base:** The anterior cranial base (s-n) was initially shorter but grew in the same way and at the same rate as the controls. The cranial base angle (n-s-ar) was smaller in boys than in normal subjects and did not change during growth.
- 2) **Upper Face:** The length of the maxilla (sp-pns) - subspinale was used instead of A point. The upper face was considerably shorter in the BS subjects but grew in parallel with the controls. The upper anterior face height (n-sp) was lower than in the controls at early ages but increased progressively while the posterior height (ptm-ptm') stayed within norms. The maxillary apical base was smaller than in the controls. The inclination of the nasion plane to anterior cranial base (NSL) varied individually.
- 3) **Lower Face:** The length of the mandible (go-gn, ar-go) was shorter in most subjects initially but followed the same growth rate as the control group. Lower anterior face height remained normal. The gonial angle and mandibular plane angles varied individually.
- 4) **Total Face:** The anterior face height (n-gn) was in most subjects within norms.
- 5) **Profile:** The profile angle (n-'ss'-pog) was larger in the BS subjects and became more accentuated with age, thus increasing the profile concavity with age. The inclination of the nasal bone (s-n-r) was considerably smaller than in the controls and remained so.
- 6) **Pharyngeal Depth:** This variable (ptm-ph) was smaller than normal and was remarkably stable during growth.
- 7) **Dental relationships:** All values for dental relationship varied greatly in both sexes and showed the greatest variations. The most striking feature was the proclination of the upper incisors, which was already large at first registration and became more pronounced with age. The lower incisors were also more protrusive than the controls or within normal limits.

Comparison between the two longitudinal samples also provided information on the effects of orthodontics on BS cases by Olow-Nordenram and Thilander (1989). Unfortunately, no positive influence of orthodontic treatment on craniofacial growth in BS subjects could be stated. However, the graphical description of the groups, utilising facial polygons, revealed marked divergences even at early ages, remaining throughout the growth period. The maxillary retrognathia of the more severe cases was aggravated by a craniofacial Class III tendency in the lower face. In such cases, combined orthodontics and orthognathic surgery was performed after completion of adolescent growth.

CHAPTER 3:

SOFT TISSUE PROFILE EVALUATION

Individuals with the unusual facies of BS or CDP present to orthodontists, oral-maxillofacial surgeons and plastic surgeons to correct their facial deformity and obtain relief from their psychosocial distress. The extent of an individual's facial deformity is bound up in the aesthetic value placed on "ideal" facial proportions.

Aesthetics is defined in "Webster's Collegiate Dictionary" as "the branch of philosophy dealing with the beautiful, chiefly with respect to theories of its essential character, tests by which it may be judged, and its relation to the human mind." The word "aesthetics" was derived from the Greek word "*aisthetikos*", meaning "perceptive, especially by feelings". Therefore, in discussing aesthetics, we are concerned essentially with subjective reactions, that is, how we feel about things that we see (Lusterman, 1963).

Powell and Rayson, (1976) defined facial aesthetics as the study of the variations that may occur in facial appearance on the one hand and the individual response of observers to these variations on the other.

Peck and Peck, (1970) believed that the formalised studies of psychology and sociology have helped transform aesthetic judgment from simply a visual "feeling" to an understanding exercise in visual perception. They believed that the study of the face as the "aesthetic stimulus", and the nature of the "aesthetic response" or the observer's perception are of equal importance.

Burstone (1958) stated that facial form may be divided into two planes of space: frontal and sagittal. The midsagittal plane produces an outline which commonly is referred to as the profile. So, with the lateral surface of the face oriented perpendicular to the camera, the profile along the median sagittal plane may be recorded photographically (Peck and Peck, 1970).

As mentioned by Luckner (1980), although the face is commonly seen from a multitude of perspectives and angles, angular views and three-dimensional representations complicate precise measurement of the face. Facial features have commonly been studied in full frontal and profile views. Powell and Rayson (1976) advocated using the three-quarter facial view for a more complete analysis of the face. Burstone (1958), Peck and Peck (1970) and Barrer and Ghafari (1985) believed that although the complexities of facial aesthetics may not be totally expressed in one method of analysis, a profile view of the face provides information necessary for diagnosis and treatment planning of dentofacial problems. So, emphasis has been placed on the profile view, since many dentofacial malformations as well as therapy changes are more evident in this plane of space, especially in orthodontics.

In the present study, the soft tissue characteristics seen in the two syndromes will be examined from the profile view. Photographic records will be used exclusively for the soft tissue evaluation of BS and CDP.

3.1 The Soft Tissue Covering of the Face

During orthodontic treatment, dentoalveolar changes will impact the soft tissue aesthetics in a negative or positive way, implemented by treatment and concomitant growth.

In investigating integumental extension patterns, Burstone (1959) found that the soft tissue mass of the face lying inferior to subnasale is relatively thick compared to the that of the glabellar region. This difference reflects, in part, the high degree of development of the orbicularis oris complex. The upper lip thickness gradually reduces from subnasale to labrale superius. The horizontal extension at inferior labial sulcus and pogonion which Burstone referred to as "menton" is less than any other region of the lower face. Burstone found the greatest variation in horizontal and vertical extension values in the lower face, especially in the lip region.

Many investigators have studied profile changes during growth and treatment and generally agree that the relationship between hard and soft tissues is very complex. This implied that it is necessary to study directly the integumental contour of the face, since hard tissue measurements can deviate considerably from the facial form which the patient expresses with the soft tissues.

3.2 The Development of Concepts on Facial Aesthetics in Orthodontics

The early orthodontists had been very much influenced by the facial aesthetics of the Greek sculptures. Angle (1907) modelled his concept of an ideal profile and facial beauty through his association with the noted art teacher E.H. Wuerpel. Angle said of Apollo Belvedere: "...every feature is in balance with every other feature and all the lines are wholly incompatible with mutilation or malocclusion."

Facial aesthetics in orthodontics have been influenced by ancient art, influential clinicians such as Grieve (1944) and Tweed (1953) and cephalometric norms (Riedel, 1957). Three primary sources of aesthetic idealism were found:

- 1) The first source of aesthetic idealism was probably derived from ancient art.
- 2) The second developed through the influence of such men such as Grieve and Tweed, who developed concepts of aesthetics based on accepting as pleasing or satisfactory, a face with a stable dentition and incisors in an uncrowded upright position.

- 3) The third concept of aesthetics was based upon orthodontic cephalometric standards or norms based on "normal occlusion" developed by orthodontists such as Margolis (1943), Steiner (1953), Downs (1948) and others.

However, a compromise in facial appearance must sometimes be accepted to secure occlusal stability.

3.3 Methods of Soft Tissue Evaluation

There are many methods of hard tissue analysis based on the lateral cephalogram. In comparison, there are fewer comprehensive soft tissue analyses available to the clinician. This is partly due to the problem of lip posturing and the reliability and reproducibility of the various soft tissue landmarks (Wisth and Boe 1973; Hillesund *et al.* 1978). There is also the difficulty in predicting the eventual soft tissue outcome with growth and treatment.

Soft tissue profile analysis can be conducted from lateral profile photographs (Stoner, 1955; Neger, 1959; Peck and Peck, 1970; Larrabee *et al.* 1985; Satravaha and Schlegel, 1987; Yuen and Hiranaka, 1989) or from the soft tissue outline on lateral cephalograms.

Simon (1926) as cited by Burstone (1958), and Hellman (1939) advocated systematic measurement and analysis of the face. They constructed lines and angles, and made measurements directly from the patient and photographs for use in orthodontic diagnosis and classification. After the clinical introduction of radiographic cephalometrics, the profile photograph became a rather passive facial record. However, a soft tissue profile analysis carried out both clinically and photographically can provide valuable information regarding facial aesthetics, and as a supplement to other diagnostic records.

Peck and Peck (1970) recommended that profilometric analysis be conducted on oriented facial photographs. In this way the orthodontist may evaluate subjective factors such as facial topography, muscle contours, and the structural elements of the side of the face while constructing the angular measurements. They advised that the photographs be taken with the subject in a cephalostat. Standardised head positioning is thus assured for comparative purposes.

3.4 Total Facial Profile Analysis

3.4.1 Antero-Posterior Assessment of the Facial Profile

Profitt and Ackerman (1986) in their clinical assessment, placed the patient in natural head position, either sitting upright or standing, and looking at a distant object, to establish the following:

- 1) Divergence of the face, which is defined as an anterior or posterior inclination of the lower face relative to the forehead, was a term originally coined by Hellman (1939). Anterior divergence, where the face slopes anteriorly, or posterior divergence where the face slopes posteriorly, can be compatible with good facial aesthetics and proportions; in fact a degree of facial divergence is a racial and ethnic characteristic. For example, American Indians and Orientals tend to have an anteriorly divergent face, while Caucasians of Mediterranean ancestry tend to have a posteriorly divergent face.
- 2) Profile convexity or concavity is detected by viewing the relationship between two lines, one dropped from the bridge of the nose to the base of upper lip, a second extending from that point downward to the chin. These line segments form a nearly straight line. An angle between them indicates profile convexity (upper jaw prominent relative to chin) or profile concavity (upper jaw behind chin).

The “soft tissue angle of convexity” was described by Wylie (1955) as the angle that is roughly the soft tissue equivalent to the bony landmarks which defined Downs’ angle of convexity (nasion - point A - pogonion). The angle involves the prominence of the forehead, the most prominent point on the upper lip, and the most prominent point on the soft tissue chin. The acceptable range was found to be between 6.5 to 16 degrees.

Burstone (1958, 1967, 1975), Legan and Burstone (1980) and Hunt and Rudge (1984) described the overall horizontal soft tissue profile, by using the angle of facial convexity (glabella - subnasale - soft tissue pogonion). Legan and Burstone (1980) suggested that the mean for adults is 12 ± 4 degrees, which indicates that the soft tissue profile normally demonstrates definite convexity. As the angle of facial convexity becomes a smaller positive or a negative value, the profile is suggestive of a Class III skeletal and dental relationship. A clockwise angle is positive, and a counterclockwise angle is a negative. As the positive angle increases, the profile becomes more convex, suggesting a Class II skeletal and dental relationship.

Subtelny (1959) employed two methods to evaluate the convexity of the soft tissue profile:

1. Soft tissue profile convexity (soft tissue nasion - subnasion - soft tissue pogonion): this is designed to focus upon soft tissue structures which are

closely analogous to the structures utilised for measuring the degree of convexity of the skeletal profiles.

2. Total soft tissue profile convexity, including the nose (soft tissue nasion - tip of nose - soft tissue pogonion): this is devised so that the nose can be included in the measurements of facial convexity, as the nose has a marked influence upon the total soft tissue profile.

The soft tissue facial angle was used by Stoner (1955) as part of his facial contour analysis from profile photographs. The facial angle is formed by the intersection of facial plane, which is the line drawn from the depth of concavity at the base of the nose to a tangent to the chin, and Frankfort horizontal, drawn from tragon or the lowest margin of the tragus as it passes posteriorly on the ear to orbitale. For what is identified as an excellent Caucasian profile, the mean is 87.7 ± 2.9 degrees; the range is from 79 to 92 degrees.

Neger (1959) in his soft tissue profile analysis from oriented profile photographs, also used this same angle, which he called pogonial angle (angle Pg). In his sample of good, acceptable, average profile with excellent occlusion, the mean is 88.1 ± 2.9 degrees; the range is from 81 to 95 degrees.

The soft tissue facial angle was defined by Holdaway (1983a) as the angular measurement of a line drawn from soft tissue nasion (where the sella-nasion line crosses the soft tissue profile) to the soft tissue pogonion, relative to the Frankfort horizontal plane. A measurement of 91 degrees is ideal, with an acceptable range of ± 7 degrees. This gives an assessment of the soft tissue chin prominence in the facial profile.

Peck and Peck (1970) in their profilometric analysis study from lateral profile photographs employed the soft tissue orientation plane. This is constructed by drawing the facial line from nasion to pogonion (N-Pg) and bisecting it. The midpoint (P) is then connected with tragon (T) to form the orientation plane. The facial angle (F) is formed by the intersection of the orientation plane with the facial line at point P. It is read as the inside inferior angle and serves as an index of profile orientation. In their sample of aesthetically pleasing faces, the mean facial angle is 102.5 ± 2.7 degrees; the range is from 96.0 to 106.5 degrees.

Satravaha and Schlegel (1987) cited the work of Schwarz (1951) for profile form and profile flow analyses. For these two analyses, the Frankfort horizontal plane (PHP), the line joining tragon and soft tissue infraorbitale, is used as the horizontal reference plane. Two vertical lines, line A from infraorbitale and line B from nasion, are drawn perpendicular to Frankfort horizontal plane.

- 1) Profile form: compares the horizontal relationship of soft tissue subnasion to line B. If subnasion lies anterior to line B, the facial profile is considered to be

prognathic; if subnasion coincides with line B, the profile is orthognathic; and if subnasion lies posterior to line B, the profile is retrognathic.

- 2) Profile flow: compares the horizontal relationship between soft tissue pogonion and lines A and B. If pogonion is positioned more anteriorly towards line B, this is called a "forward shift" of the chin; if pogonion is located midway between lines A and B, the profile is orthognathic; if pogonion is located more posteriorly, this is called "backward shift".

The nose has a significant influence on the soft tissue profile. There have been many angular and linear measures developed to characterise the nose. There are two soft tissue angles most commonly used in plastic and orthognathic surgery: the nasofacial and the nasolabial angles.

The nasofacial angle is the angle formed by the intersection of the line joining glabella and soft tissue pogonion with the line which defines protrusion of the nose from the facial plane. The ideal nasofacial angle for Caucasians is between 36 to 40 degrees, according to Powell and Humphreys (1984). The nasofacial angle is illustrated in Fig. 5.2 of the present study.

Legan and Burstone (1980) and Hunt and Rudge (1984) considered the nasolabial angle an important measurement in assessing the antero-posterior maxillary dysplasia. The nasolabial angle takes into account the inclination of the columella of the nose and the position of the upper lip. An obtuse nasolabial angle suggests a degree of maxillary hypoplasia or lack of lip support, whereas an acute nasolabial angle indicates upper lip prominence. The nasolabial angle is illustrated in Fig. 5.2 of the present study.

There are various definitions of the nasolabial angle. Burstone (1967) and Lo and Hunter (1982) defined the nasolabial angle as the angle formed by the intersection of a line originating at subnasale tangent to the lower border of the nose and a line from subnasale to labrale superius. Legan and Burstone (1980) defined the nasolabial angle (Cm Sn - Ls) as the angle formed between the line connecting the columella point and subnasale and the line from subnasale to labrale superius. The mean value is 102 ± 8 degrees.

Powell and Humphreys (1984) described the nasolabial angle as the angle from a line tangent to the most anterior point of the columella (Cm) to the subnasale (Sn), and a line intersecting the subnasale and the muco-cutaneous border of the upper lip. The range is between 90 to 120 degrees.

Hunt and Rudge (1984) measured the nasolabial angle as the anterior angle formed between the Frankfort plane and a line tangent to the upper lip through subnasale. This angle normally approximates 90 degrees. They suggested that the nasolabial angle should be studied relative to the Frankfort horizontal plane, since the shape of the columella would influence the magnitude of this angle.

According to Holdaway (1983a), nose prominence can be measured by means of a tangent at the vermilion border of the upper lip perpendicular to the Frankfort horizontal plane. This measures the nose from its tip in front of the line and the depth of the incurvation of the upper lip to the line. Arbitrarily, those noses under 14 mm are considered small, while those above 24 mm are in the large or prominent range.

3.4.2 Vertical Assessment of the Facial Profile

Angular and linear vertical measurement of the integumental profile can also be performed using soft tissue landmarks from either photographs or lateral cephalograms.

Legan and Burstone (1980) in their vertical facial height proportionality assessment suggested that the ratio of the distances glabella to subnasale (G-Sn) and subnasale to soft tissue menton (Sn-Me'), measured perpendicular to the Horizontal Plane, should be approximately 1:1 in normal adults. The lower third of the face (Sn-Me') can be further divided into thirds. The length of the upper lip, which is measured from subnasale to stomion superius (Sn-Stm_s) should be approximately one third of the total, and the distance stomion inferius to soft tissue menton (Stm_i-Me') should be about two thirds. Stated another way, this ratio of Sn-Stm_s : Stm_i-Me' should be 1:2.

Powell and Humphreys (1984) used another method for evaluating vertical facial height proportions. The ratios for the midfacial height (Na'-Sn) is 43 percent and the lower facial height (Sn-Men') is 57 percent of the total length from nasion to menton. They suggested that the nasion (Na') point marks the upper limit of the nasal length and is far more reproducible than glabella (G).

Ricketts *et al.* (1979) as cited by Hunt and Rudge (1984), suggested that in the anterior soft tissue height assessment, the distance from the eyebrow to subnasale and from subnasale to soft tissue menton should be approximately equal. When measured perpendicular to Frankfort plane the mean for this distance is 66 mm in normal Caucasian males and 60 mm in females. The distances from subnasale to lower lip vermilion and lower lip vermilion to soft tissue menton should be equal for ideal balance in the lower third of the face. The clinical average is 33 mm in males and 30 mm in females.

Peck and Peck (1970) utilised four angular measurements which can be applied to measure vertical height:

1. The nasal angle (Na) measures nasal height from nasion to pronasale. The mean is 23.3 degrees and the range is 20 to 27 degrees.
2. The maxillary angle (Mx) measures maxillary height from pronasale to labrale superius. The mean is 14.1 degrees, with a range of 12 to 17 degrees.
3. The mandibular angle (Mn) measures mandibular height from labrale superius to pogonion. The mean is 17.1 degrees, with a range from 14 to 20 degrees.
4. The total vertical angle (TV) is the total dimension from nasion to pogonion. The mean value is 54.5 degrees with a range of 47 to 62 degrees. The vertex of all these angles is at tragion.

Cutcliffe (1974) as cited in Worms, Isaacson and Speidel, (1976) suggested as a general rule, that the total facial height between the eye and the soft tissue menton can be divided into fifths. The upper facial height (eye - subnasale) is $\frac{2}{5}$, the upper lip length (subnasale - stomion) is $\frac{1}{5}$, and the lower lip length the remaining $\frac{2}{5}$. Another proportionality that is helpful in assessing the lower facial height (subnasale - menton) is that the upper lip length is one half of the lower lip length.

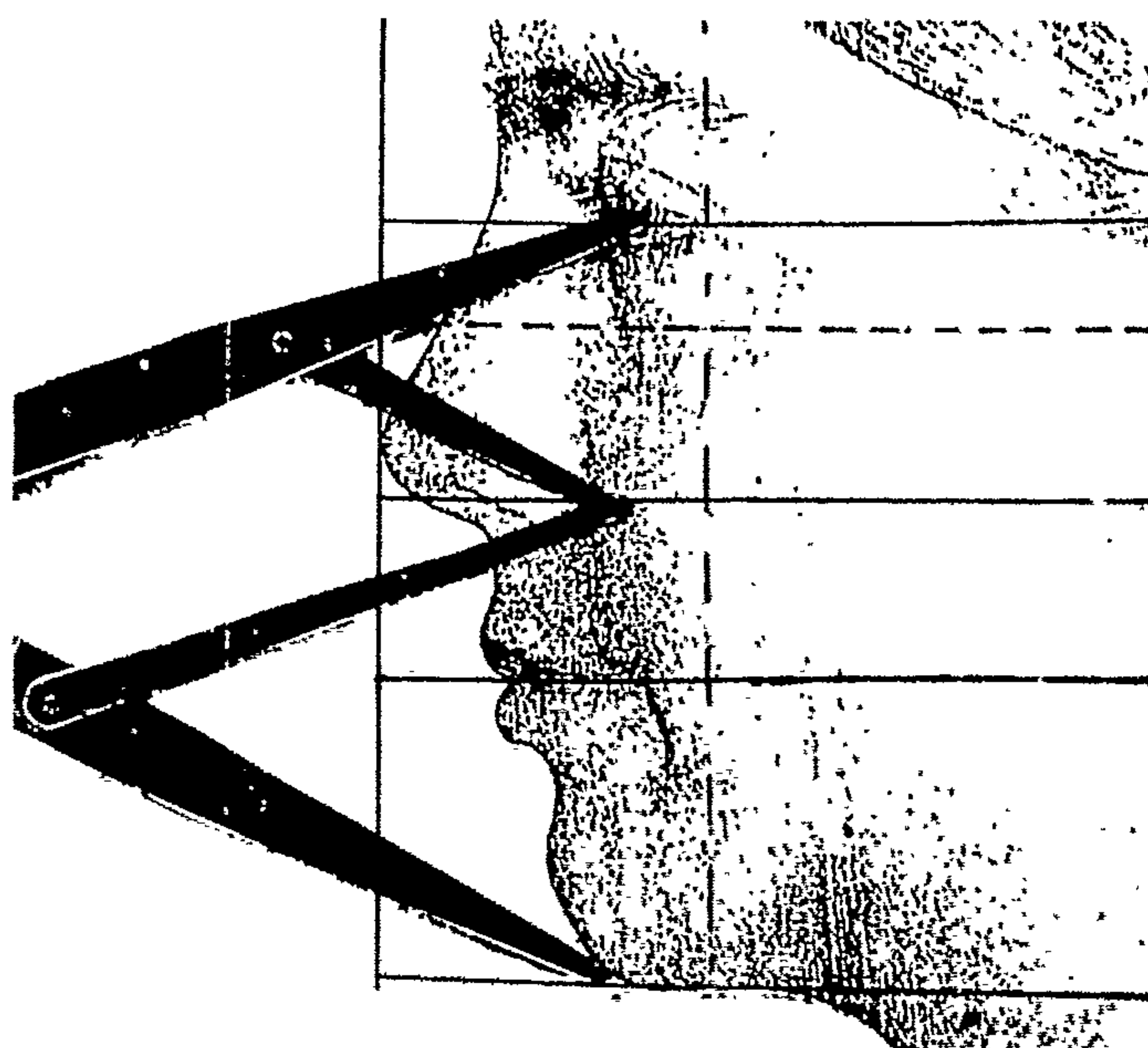


Fig 3.1: The “golden proportion” from eye-nose-chin (Ricketts, 1981).

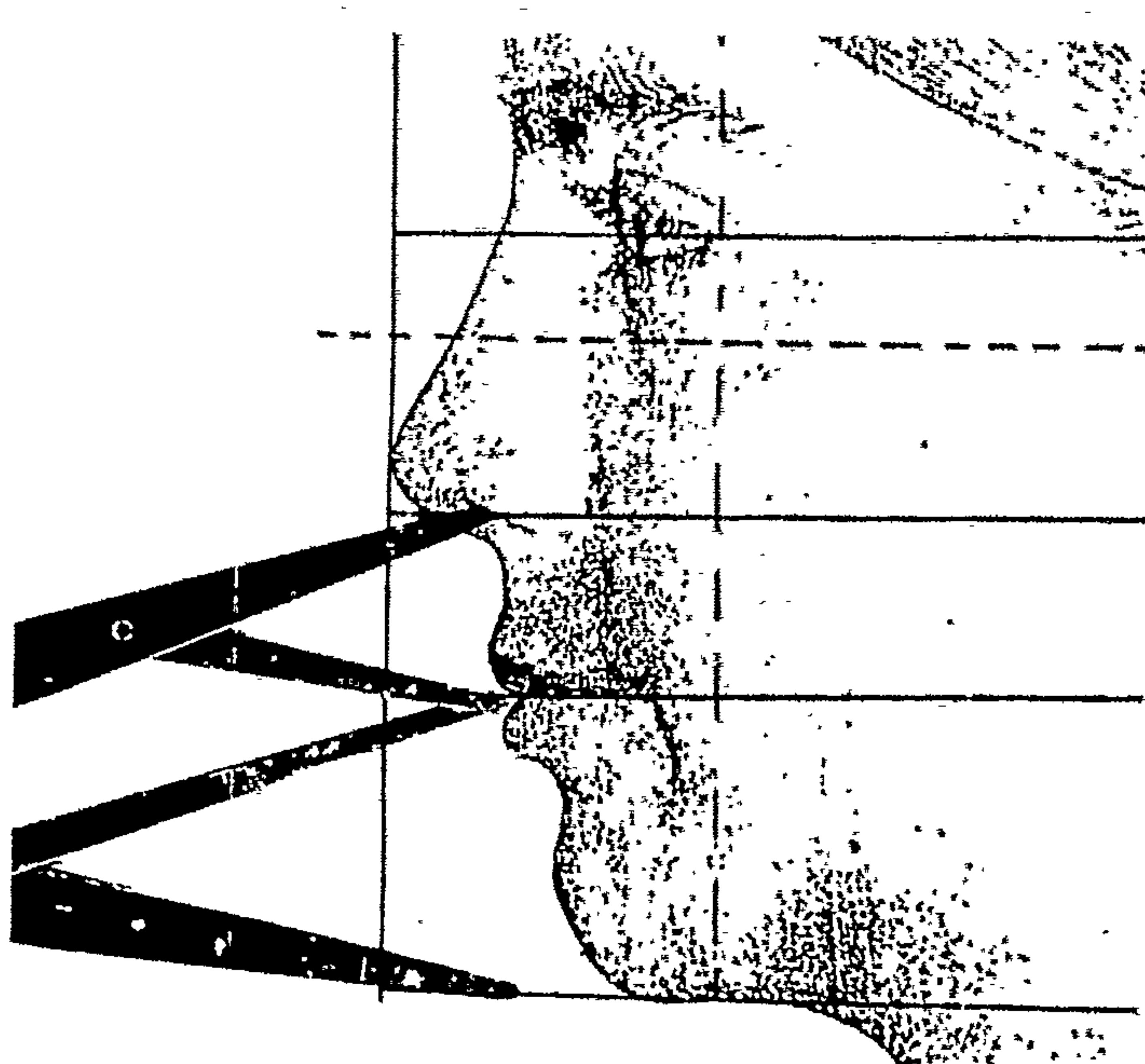


Fig 3.2: The “golden proportion” from nose-mouth-chin (Ricketts, 1981).

Ricketts (1981, 1982) proposed the concept of the "Golden Proportion," the "Divine Proportion" or "Phi" for the analysis of structural harmony and balance of the face in all dimensions. For appreciation of beauty, it has been suggested that the human mind functions at the limbic level in attraction to proportions in harmony with the Golden Section, which is a proportion of 1.618. That which is known as the golden section has been known at least since the time of the ancient Egyptians and was extensively used in Greek art and architecture. The divine proportion is also called "Phi", after the first letter in the name of a famous Greek sculptor, Phidias, who used the golden section extensively in his art.

This golden relation, or Fibonacci number (1:1.618 or its reciprocal 0.618) can be measured with the golden divider. Upon widening the divider, the longer side is 1.618 times the shorter side and the shorter side is 0.618 the length of the longer. In turn, the longer side is 0.618 the length of the total outer measurement. For vertical proportion in the profile aspect, a divine proportion is recognised in the eye - alar of the nose - chin positions (Fig 3.1); the alar of the nose - lip embrasure - chin positions (Fig. 3.2).

The numbers used by Ricketts were obtained from the original work of Filius Bonacci, also known as Leonardo of Pisa, who published his work, *Liber Abaci*, in 1202, which was to change the Roman numerical world into the Hindu-Arabic numeral system. Bonacci was famous for the Fibonacci Sequence of numbers, which is basically a ratio of addition related to biology (Ricketts 1981, 1982).

3.5 Lower Facial Profile Assessment

The soft tissue contour of the lower third of the face has been studied extensively by orthodontists, as orthodontic treatment by altering the dentoalveolar region may produce changes in the external or integumental contours of the face (Burstone, 1958; Hambleton, 1964; Spradley *et al.* 1981).

Evaluation of lip protrusion can be conducted by relating the lips to a true vertical line passing through the concavity at the base of the upper lip (soft tissue A point) and through the similar concavity between the lower lip and chin (soft tissue B point). The lip should lie along or only slightly in front of this line. If the lip is significantly forward from this line, it can be judged to be protrusive; if the lip falls behind the line it is retrusive. Lip protrusion, like facial divergence, is strongly influenced by racial and ethnic characteristics. Caucasians of Northern European origin have relatively thin lips and minimal lip prominence. Caucasians of Southern European and Middle Eastern origin normally have more lip prominence than their Northern cousins.

Burstone (1958) in his analysis of the lower third integumental profile, used contour angles to describe the profile components to each other.

1. Maxillomandibular contour (ACDF): angle formed by the intersection of upper facial and anterior lower facial components.
2. Labiomandibular contour (CDF): angle formed by the intersection of interlabial and mandibular components.
3. Maxillary sulcus contour (ABC): angle formed by intersection of subnasal and superior labial components.
4. Mandibular sulcus contour (DEF): angle formed by the intersection of inferior labial and supramental components.

The landmarks used are: subnasale (A), superior labial sulcus (B), labrale superius (C), labrale inferius (D), inferior labial sulcus (E), and menton (F). It should be noted that Burstone's soft tissue menton is equivalent to soft tissue pogonion.

Ricketts (1957, 1960, 1968, 1981) proposed a line tangent to the chin and the nose, which he called the "esthetic plane" or "E" plane to evaluate lip relation to the other profile structures. In Caucasians, by adulthood, the lips should be contained within this line, with the contour of the lips smooth, and the mouth closed with no strain. The upper lip is ideally located 2 mm further behind the line than the lower lip. As the nose grows and the chin develops, the lips gradually appear to contract into the face. In the juvenile stages, the lips may start slightly ahead of the "esthetic plane"; by adolescence, the lower lip drops behind this line and continues to retract in adulthood.

Ricketts (1968) noted that most people object to lips that protrude beyond the E plane. Lip prominence seemed to be an undesirable trait and an unacceptable situation, particularly in adults. However, fullness of the lips and mouth prominence are characteristics of the young. He also noticed that many women object to excessively flat mouths or puckered lips later in life because the prominent dentition and the full mouth constitute a mark of youth, whereas flat mouths suggest old age.

Steiner (1960) suggested the "S plane", that is drawn from the chin to the middle of the "S" formed by the lower border of the nose and the upper lip. He believed that in "good" faces, the lips often fall on this line and that lips ahead of it would, on an average, be too full, whereas those falling behind it would give too flat an appearance as related to other parts of the profile.

As cited by Hambleton (1964), Holdaway suggested the use of the "H angle" for soft tissue profile diagnosis. The "H angle" is formed by the "H" line, a line tangent to the soft tissue chin and the upper lip, and the line NB. Holdaway believed that when the ANB angle is 1 to 3 degrees this "H" angle should be 7 to 9 degrees to produce a pleasing profile. Changes in the ANB angle would also mean similar changes to the "H" angle. Holdaway (1983a) redefined his "H" angle as the angle measurement of the "H" line to the soft tissue nasion - pogonion line or soft tissue

facial plane. He found this angle to be a better indicator of the prominence of the upper lip in relation to the overall soft tissue profile, and it takes into account the variability of the chin area that is not considered by the ANB angle. For Caucasians, the ideal "H" angle is 10 degrees, when the skeletal profile convexity measurement, defined as the linear measurement from point A to the hard tissue line nasion - pogonion, is 0 mm. The best range is from 7 to 15 degrees as dictated by the convexity present. As indicated by Holdaway, there is no single "H" angle that can be set as an ideal for all types of faces, but it will increase proportionately as the skeletal convexity increases.

The upper lip form is important in the study of profile form, especially with regard to orthodontic treatment. Holdaway (1983a) took the upper lip form or curl as the linear measurement from the superior sulcus depth to a perpendicular to Frankfort horizontal and tangent to the vermilion border to the upper lip. The ideal superior sulcus depth is 3 mm, with a range of 1 to 4 mm.

Holdaway (1983a) noted that the lower lip is considered ideal when it is zero to 0.5 mm anterior to the "H" line. The contour in the inferior sulcus area should fall into harmonious lines with the superior sulcus form. The inferior sulcus is taken as the point of greatest incurvation between the vermilion border of the lower lip and the soft tissue chin and is measured to the "H" line.

Merrifield (1966) suggested a line drawn tangent to the soft tissue chin and the most procumbent lip extending superiorly to intercept the Frankfort Horizontal plane. The inferior angle formed by the intersection is called the "Z" angle, which indicates the extent of lip protrusion. The mean is 81.4 degrees, with a range of 71 to 89 degrees.

Burstone (1967, 1975) used a plane that connected subnasale and soft tissue pogonion to evaluate the relative protrusion or retrusion of the lips. Lip protrusion or retrusion is measured as a perpendicular linear distance from the subnasale-pogonion plane to the most prominent point on the upper and lower lips. In a normal adolescent sample, the upper and lower lips lie ahead of the subnasale - pogonion plane. The mean for the upper lip is 3.5 mm anterior to the line, and the lower lip is 2.2 mm anterior to the line. Burstone selected the subnasale-pogonion plane of reference as he believed that it is a plane of minimal variation with orthodontic treatment in non-growing individuals. Burstone felt that although an "esthetic plane" joining the nose and the chin is useful for evaluating the facial form, there is so much variation in nose form and length that the nose should be avoided in the evaluation for lip protrusion.

3.6 The Structural Basis for Ethnic Variations in Craniofacial Form

Among most of the world's different racial groups, either the brachycephalic or dolichocephalic type of head form tends to predominate in any given group. However, a distribution range from one extreme to the other also usually exists within a group, even though one particular side of the range is more common. An intermediate type of head form, mesocephalic, can also occur (Enlow, 1982).

Dolichocephalic head form tends to predominate in the northern and southern edges of some parts of Europe, England, Scotland, Scandinavia, Northern Africa, and some Near and Middle Eastern countries, such as Iran, Afghanistan, India, Iraq and Arabia. The brachycephalic head form tends to predominate in Middle Europe as the "Alpine" head form.

A third type of head form, termed Dinaric can be found at the geographic interface between the dolicho - and brachycephalic regions of the world. Such areas include regions located between Middle and Northern Europe, between Middle and Southern Europe, and between Europe and the Near East. This head form is characterised by the posterior part of the skull being brachycephalized and flattened, whilst the anterior part of the skull has retained the relative narrowness that characterised the dolichocephalic pattern.

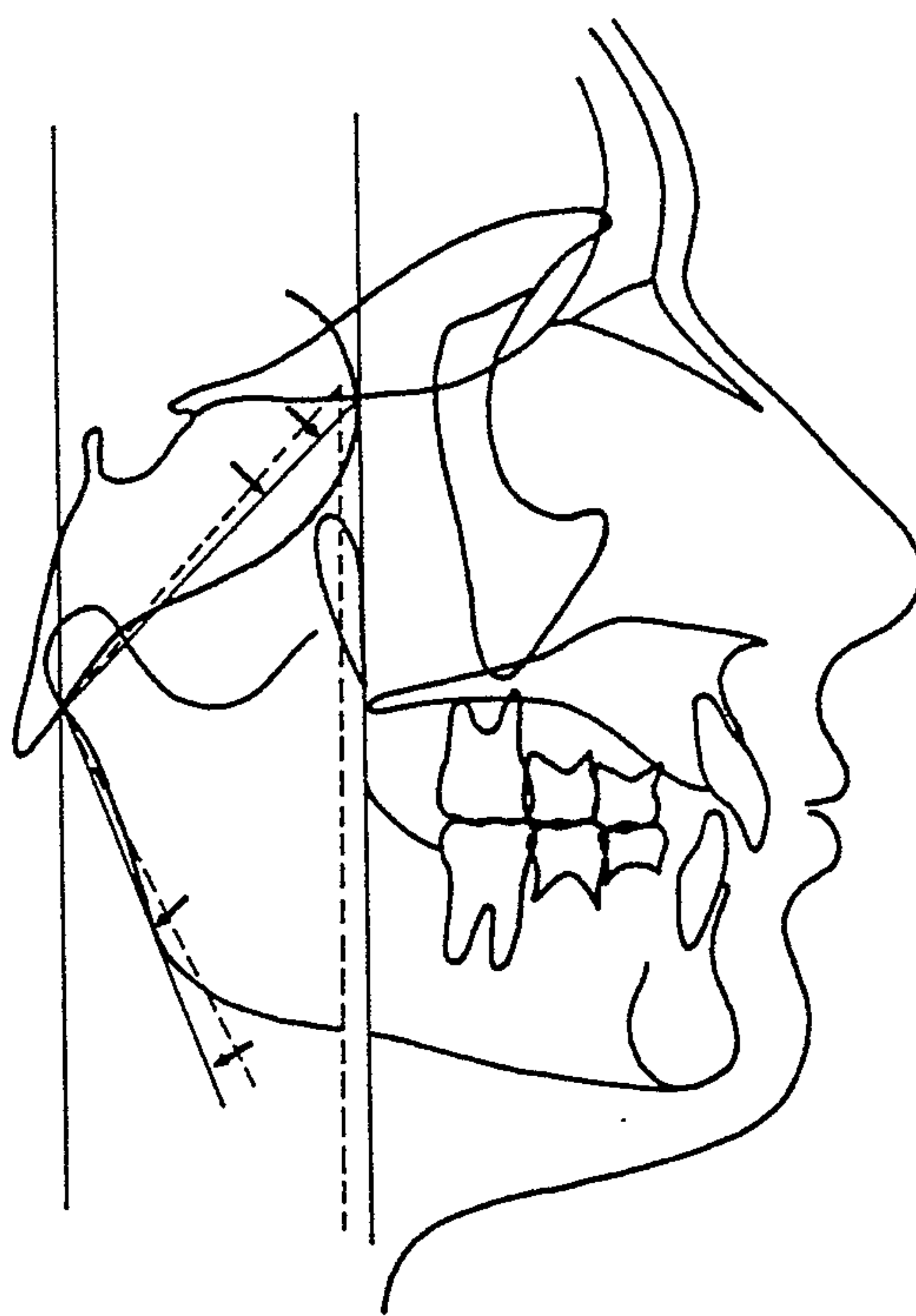


Fig. 3.3: The facial pattern that is characteristically seen in many Caucasian groups (Enlow 1982).

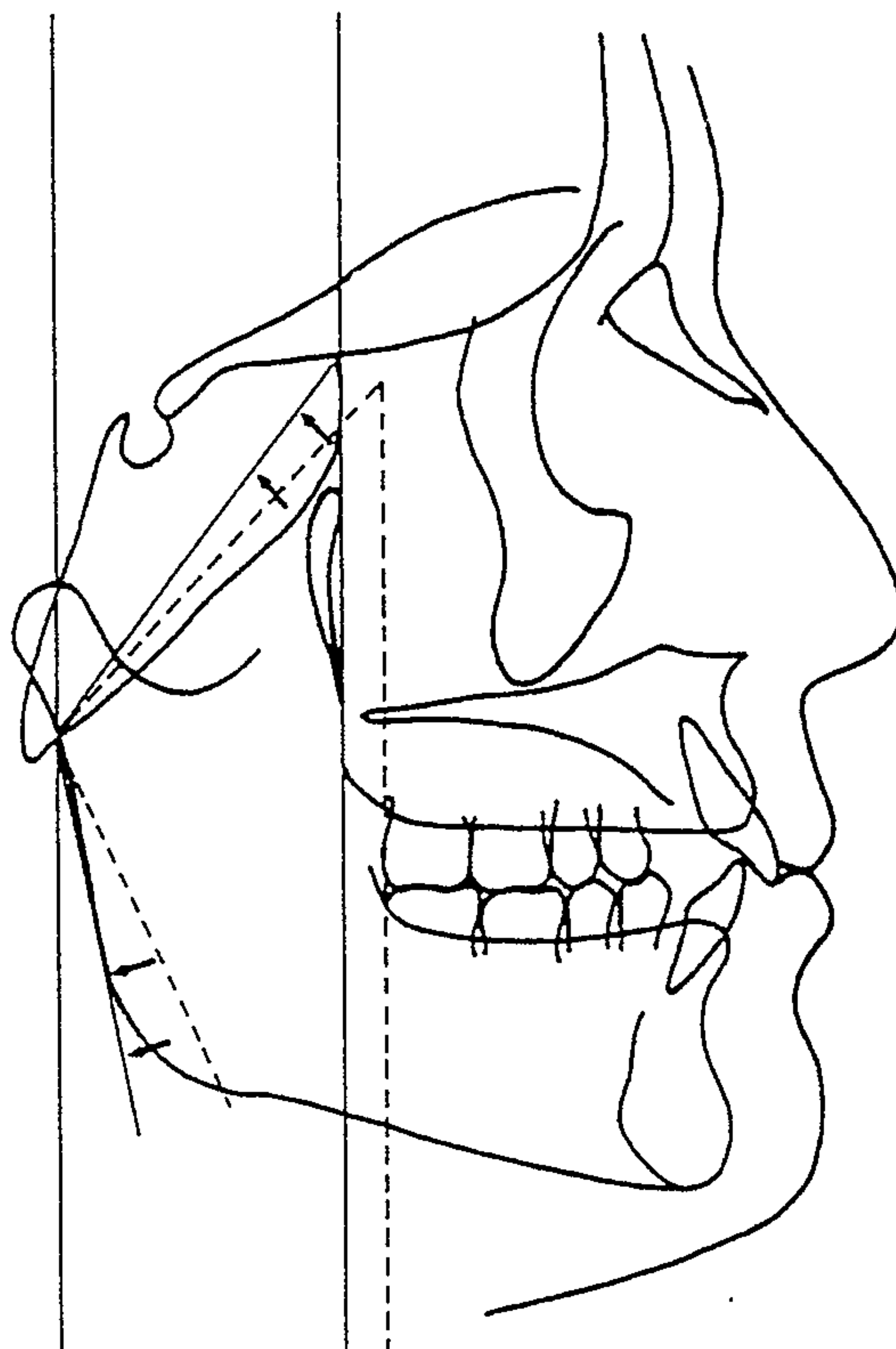


Fig. 3.4: The facial pattern that is characteristically seen in many Oriental groups (Enlow, 1982).

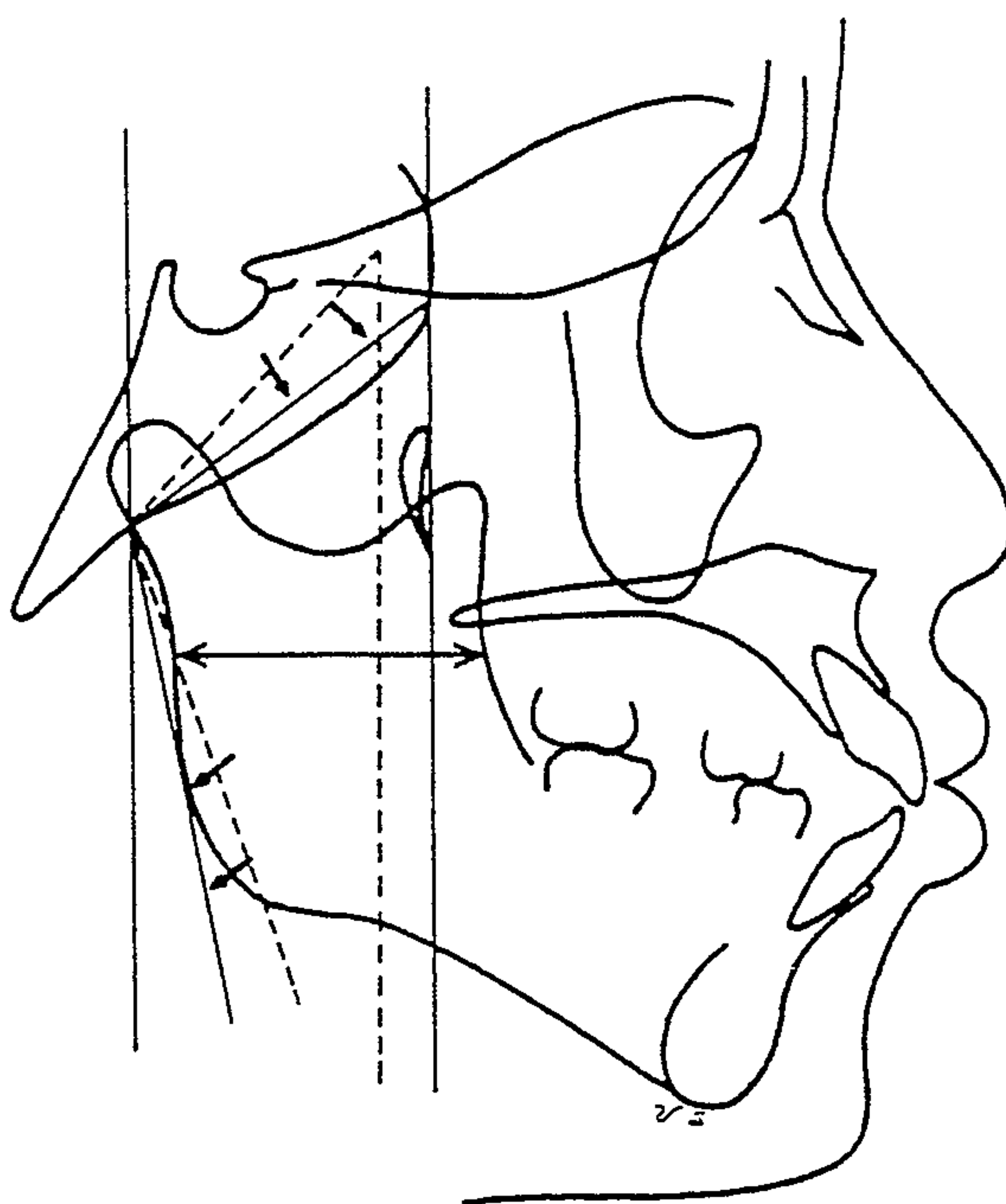


Fig.3.5: The facial pattern that is characteristically seen in Negroid groups (Enlow, 1982).

The dolichocephalic head form present in many Caucasian groups is usually associated with a more open or "flat" cranial base flexure (Fig. 3.3), which tends to produce a more protrusive upper face and a more retrusive lower face. The open cranial base flexure results in the whole nasomaxillary complex being more forwardly positioned and lowered relative to the mandibular condyle. As the condyle is "higher", there is the tendency for a downward and backward rotation of the whole mandible. Hence, there is a greater frequency of the retrognathic type of profile and a Class II tendency among these groups. There is also a high incidence of a "broad" ramus to compensate for the built-in tendency toward mandibular retrusion.

The brachycephalic head form, present in many Mongoloid or Oriental groups, is characterised by a more closed, upright cranial base flexure (Fig. 3.4). The broad head form also sets up a wider but antero-posteriorly shorter upper and midfacial region. The result is that the whole upper and midfacial region is less protrusive. There is often a "vertical" character to the whole face; the cheekbones are more prominent because the upper and middle face is less protrusive. There is a greater likelihood for this group to have a Class III malocclusion. Bimaxillary protrusion occurs when the mandible tends to be slightly long horizontally, and this causes a forward tipping and proclination of the upper incisors. The result is a protrusion of both the upper and lower incisor regions. However, the nasomaxillary complex can also be vertically long, resulting in a downward and backward rotation of the ramus. The face does not appear as long due to a greater bizygomatic width. The mandibular corpus tends to be shorter relative to the maxillary arch, and this factor, together with the backward ramal rotation, contributes to a compensation for the built-in tendency toward prognathism and bimaxillary protrusion (Enlow 1982).

The Negroid group tends to have a dolichocephalic head form, although there are broader-faced individuals, just as among Caucasians. The cranial base flexure is even more open or flat than Caucasian groups (Fig. 3.5). This factor, together with a vertically long nasomaxillary complex causes the mandible to rotate markedly downward and backward. The mandibular corpus tends to be long relative to the maxillary length, which is similar to the Caucasian pattern. However, the upper part of the face in the Negroid groups expands much less and is therefore not nearly so protrusive, a feature which corresponds to that of the Oriental. The mandibular ramus is characteristically broad, more so than in other groups; thereby offsetting an intrinsic tendency for mandibular retrusion and a Class II malocclusion. The broad ramus places the mandibular corpus, which can also be long relative to the maxilla, in a resultant protrusive position.

Thus the protrusive mandibular corpus causes the maxillary incisors to tip labially, creating a bimaxillary protrusion (Enlow 1982).

The characteristic facies of BS and CDP have been likened to Oriental facial form with regard to the nasomaxillary region. If this is the case, Oriental BS or CDP patients would be more difficult to diagnose. However, Munro *et al.* (1979) were amongst the first to state that the deformity associated with the Syndrome was of such a severe nature that Oriental groups could still be diagnosed with BS or CDP.

3.7 Soft Tissue Profile Changes with Growth in Normal Patients

From some of the growth studies (Björk 1947; Lande 1952; Subtelny 1959, 1961; Björk and Skieller 1983), it may be said that with continuing growth, the skeletal profile becomes less convex. The chin usually assumes a more forward and downward position with relation to the forehead.

Björk (1947) found that an increased prognathism of both the maxilla and the mandible is characteristic of profile changes with age; but the increase is greater in the mandible than the maxilla. This accounts for the straightening of the facial profile from the age of 12 to 20 years in his sample comprising of Swedish males only. Lande (1952) also observed that the mandible tends to become more prognathic between the ages of 7 to 17 years, while the maxilla shows very little change and this reduces the convexity of the face.

The nose grows in a downward and forward direction. Subtelny (1959) noticed that from one to 18 years of age, the rate of nasal growth does not seem to decrease appreciably with age, as is typical of the growth pattern established for the skeletal facial structures. This nasal growth contributes to the total soft tissue profile increase in convexity with age. Generally, the total profile of the nose is closely related the path of the growth of the nasal bone; the bridge of the nose and the cartilaginous aspect of the nose seem to maintain contour conformity with age. Sometimes there may be a tendency for the nasal bone to deviate from its downward and forward direction. In these instances, there is frequently a hump or elevation of the bridge of the nose (Subtelny 1959). Chaconas (1969) and Chaconas and Bartroff (1975) reported that this humping of the bridge of the nose, as a result of a forward positioning of the nasal bones, is probably caused by a forward growth of the supporting nasal cartilages. Subtelny (1959) indicated that this elevation of the bridge would be less apparent if there is a comparable and associated forward positioning of the more inferior aspect of the nasal profile.

The growth of the lips follows the general growth curve for muscle and other connective tissue within the body (Scammon *et al.* 1930 as cited by Subtelny 1959). The upper and lower lips show a progressive increase in lip length until approximately fifteen years of age; after this time it appears to slow down markedly.

With this increase in lip length there is also an increase in thickness, especially in the vermilion region of the upper lip. The increase in thickness of both lips is considerably greater at the vermilion level than in the regions overlying skeletal points A and B.

Subtelny (1959, 1961) found that the posture of the lips is strongly dependent on the position of the underlying dentoalveolar complex. After about nine years of age, when the upper central incisors have fully erupted, the upper lip maintains a fairly constant vertical relationship to its alveolar process, prosthion, and the upper incisal edges. Similarly the lower lip shows the same relative stability in its vertical relationship to infradentale and the lower incisal edges. The upper lip normally covers about 61 percent to 67 percent of the upper central incisor crown. The remainder of the tooth is covered by the lower lip. The antero-posterior posture of the lips is also closely related to their supporting hard tissue structures, ie. the teeth and alveolar processes.

Björk (1947), Lande (1952) and Subtelny (1959, 1961) found that alveolar processes do not keep pace with the growth of the skeletal bases in a horizontal direction. Subtelny noticed that point A and point B exhibit some stability in their antero-posterior relationship to each other after nine years of age, while their supporting skeletal bases continue to grow and change in antero-posterior relationship. So when the dentoalveolar structures receded and uprighted relative to the skeletal base, the lips, especially the lower lip were observed to concomitantly become more retruded relative to the facial profile. So, Subtelny concluded that there is a strong interrelationship between the lips and the dentoalveolar structures.

Ricketts (1960) found that the average convexity of the face decreases with age and the lips become progressively more retracted in relation to the "Esthetic Plane". He suggested that the lips may lie on or ahead of the E-Plane in the early ages of childhood; on it, in middle childhood; and behind it in later life.

3.8 Soft Tissue Profile Changes with Treatment in Normal Patients

In the early orthodontic literature, concern over facial features and facial aesthetics is evident. Hunter (1835), as cited by Angelle (1973), felt that one of the prime objectives of orthodontic treatment was to improve and beautify the appearance of the mouth. Case (1921) stressed that facial outline should be an important guide in determining treatment objectives and procedures. He advocated extraction in some cases of bimaxillary protrusion to retract the procumbent lips. Many studies have since been conducted to determine profile changes concurrent with orthodontic treatment, and this in part, reflects the importance of facial aesthetics, and the role the orthodontist might have in influencing facial balance.

Lip posture and tonicity and the position of the soft tissue chin are of considerable importance in the aesthetics of the facial profile. Research into the effect of changes in position of the teeth on the soft tissues have been largely concentrated on changes in the lip profile because it seems that most changes in tooth position during treatment alter lip position.

However, there appears to be some controversy as to the amount of soft tissue changes possible as a result of orthodontic treatment. Some of the controversy relates to the relationships of the hard and soft tissues. Some investigators, like Downs (1948, 1956), Steiner (1953) and Riedel (1957), stressed that the soft tissue profile is closely related to the underlying skeletal and dental structures. On the other hand, Subtelny (1959, 1961) from his longitudinal study of soft tissue profile changes, found that not all parts of the soft tissue profile directly follow the underlying skeletal profile; however, the upper and lower lips closely follow the contour of their underlying dento-alveolar structures. Thus orthodontic treatment which is aimed at dentoalveolar changes, will concomitantly change the soft tissue profile in this region. However, Burstone (1958, 1959) and Hambleton (1964) indicated that a direct relationship could not always exist because of individual variation in the thickness, length, and postural tone of the soft tissue covering the dentoskeletal framework.

Neger (1959) found that a proportionate change or improvement in the soft tissue profile does not necessarily accompany extensive dentition changes.

Soft tissue treatment changes are also complicated by the fact that changes are brought about not only by the treatment alone but also by the normal growth of the soft tissue itself. Koch *et al.* (1979) believed that the possibility of improving the soft tissue profile is limited. The disproportionate growth of the nose and the forward growth of the chin have a marked influence on the facial profile but the change of lip profile that can be effected by orthodontic treatment is relatively small.

Koch *et al.* (1979) found that the soft tissue profile did not directly reflect the change in the underlying skeletal profile. There was great variability in response of the soft tissue to the retraction of the upper incisor. It is evident that when incisors are retracted, lip tonicity is decreased and the lip becomes thicker but is retracted together with the hard tissues. Soft tissue change is not easy to measure and, because of individual variations in lip morphology and tonicity, even though the incisor positions may be similar, both the appearance and measurements can differ considerably.

Other studies by Baum (1961), Bloom (1961), Rudee (1964), Hershey (1972), Wisth (1972), Anderson *et al.* (1973), Angelle (1973), Garner (1974), Roos (1977), Oliver (1982), Rains and Nanda (1982), Lin *et al.* (1985), Shue (1985) and many others have shown that the upper and lower lips, forming the lower part of the soft

tissue facial profile can be influenced by orthodontic treatment, though it can be extremely variable.

Although, Hambleton (1964) felt that a direct relationship between hard and soft tissue cannot be relied on, the lips can be influenced by orthodontic treatment. Retraction of the upper teeth can produce a dramatic lip change. He noticed that as the lip is retracted, there is also some thickening of the lip. Ricketts (1957) supported this observation, especially in severely protruding cases, whereby the upper lip appears to be thin and stretched; so, as the teeth are retracted, the lip thickness increases. A 2 to 4 mm thickening of the upper lip can be expected in severely protruding cases, and about a 1 to 2 mm increase in cases where the upper incisors are not going to be moved excessively. The lower lip does not thicken but curls backward as a result of upper incisor retraction. An increase of soft tissue on the chin occurs because of loss of lip strain and loss of chin elevation by the mentalis muscle (Ricketts, 1960).

Holdaway (1983a) took a morphological assessment of 'upper lip strain' into account to assess the amount of upper lip retraction possible. He suggested that when there is a taper in the upper lip immediately anterior to the incisor, the lip will thicken following retraction until it approaches the thickness at point A (within 1 mm of the thickness). According to Holdaway, when the lip strain has been eliminated, further retraction of the incisor will cause the lip to follow the incisors in a one to one ratio. This, he felt, was predictable in adolescents when the lip thickness at point A is within the normal range (14 to 16 mm).

The exceptions are:

1. When the tissue thickness at point A is very thin (9 to 10 mm), the lip may follow the incisor immediately and still retain the taper;
2. When the tissue at point A is very thick (18 to 20 mm), the lip may not follow incisor movement at all.

Adult tissue reaction is similar to the first exception. Even though there may be lip taper, the lips will usually follow the teeth immediately.

Waldman (1982) in his cephalometric study of the influence of hard tissue changes on lip contour, found that the nasolabial angle was increased with uprighting, lingual tipping, of the incisors. Lo and Hunter (1982) found that the increase in nasolabial angle is significantly correlated with the amount of maxillary incisor retraction in the treatment of Class II, division I malocclusion.

Rains and Nanda (1982) looked at the upper and lower lip response to upper and lower incisor retraction in 30 late adolescent and early adult female patients. They found that the lower lip was more variable than the upper lip to differences in

the upper incisor movement. The upper lip at labrale superius was found to be more variable with increased retraction of the upper incisors.

Lin *et al.* (1985) studied the soft tissue changes during Class III traction on Chinese children. They found that changes in the upper and lower lip relation closely followed changes in angulation of the upper and lower incisors. They also noticed that soft tissue change was often greater than that of the skeletal tissue during orthodontic treatment; so that it is possible for some skeletal Class III patients to obtain an acceptable or more satisfactory soft tissue profile than their skeletal pattern would appear to permit. Shue (1985), also found an improvement in the overall profile with orthodontic treatment of Class III patients, with positive changes in the angles of convexity of both the hard and soft tissues. However, the changes were much smaller than that reported by Lin *et al.* (1985).

3.9 Interdisciplinary Treatment of Binder's Syndrome and Chondrodysplasia Punctata

In BS and CDP the severity of malocclusions is intimately connected with the severity of the Syndrome. In mild cases, orthodontic treatment may not be necessary, because of compensatory effects in the dental arches, while in the most severe cases the maxillary underdevelopment is aggravated by true mandibular prognathism and must be treated with combined therapy.

Many surgical protocols have been presented, all based on the severity of the deformity and the age of the patient. Munro *et al.* (1979), emphasised that surgery must be directed at three defects in BS: (1) the short maxilla with resulting Class III malocclusion, (2) perialar flatness and (3) hypoplastic nose with decreased columella.

Staging the procedures is important. Children suffering from BS or CDP should be followed up for psychosocial and orthodontic reasons.

CHAPTER 4: PHOTOGRAMMETRIC EVALUATION

“Photogrammetry” of the face is an indirect anthropometrical technique adapted for quantification of surface features from standard one fifth, one fourth, one third, one half, or life-sized photographs (Farkas, 1981). In direct anthropometry, measurements are taken directly from the subject’s face, whereas in indirect anthropometry or photogrammetry, they are taken from the standard photographs. Standardised photography providing standard print sizes and views implies photogrammetry is scientific, accurate, and documentary (Farkas, 1981). Standardised photographs as used in photogrammetry may provide considerably more benefit (Farkas, 1981) as non-standardised ones are no more objective than visual assessment in complementing a patient’s history. Facial photographs have advantage over radiographs, in that patients are seen as they are.

Facial photogrammetry has been used widely (Stoner 1955; Neger 1959; Peck and Peck 1970) although assessment of the reliability of the indirect measurements obtained by this method has been limited (Tanner and Weiner, 1949; Gavan *et al.* 1952; Fraser and Pashayan 1970).

4.1 Standardised Photography

Standardised photographic techniques and subject position allow accurate longitudinal serialisation and comparison of treatment stages. If the camera position, the patient-to-camera distance, and the subject’s head orientation are not standardised, comparison of photographs will be inaccurate (Fricker, 1978 and 1982).

Lucker (1980) recommended standardisation of photographs and radiographs for all patients for comparative studies by:

1. Fixed camera to target distance to ensure size constancy for photographic and radiographic records.
2. Standardised head position, and pose, for example in lip posture, with head holders.

Morrello *et al.* (1977) and Crawford (1987) stressed the standardisation of orthodontic and surgical photographic records, by consistency of camera settings, careful lighting adjustment, alignment, camera and subject positioning, as well as standardisation of the lens. The recommended lens is the 105 mm lens fitted to a 35 mm, single-lens, reflex camera, as this focal length produces the least distortion in facial photography (Morrello *et al.* 1977).

Brodbelt (1978) described standardisation through a mirror system to obtain an anterior view and both left and right profiles with one exposure.

Yuen and Hiranaka (1989) profile photography, using a fixed camera to subject distance with a reference ruler attached to a plumb line.

Farkas (1981) described another standardised photographic technique developed at The Hospital for Sick Children, Toronto, Canada. The lens-subject distance used in their photography was 3352.8 mm, which was 2.2 times greater than the minimum recommended by Gavan *et al.* (1952) to reduce photographic error to less than one percent. This calculation is based on the maximum breadth of the subject being photographed, in their study the head width at the level of the parietal and temporal bones (eurion-aurion) measurement was used, which in adult men is an average of 153 mm. Thus they minimised the distortion in the plane on which the camera was focussed. However, distortions in other planes were not reduced. Two life-size photographs were taken using this procedure.

For the frontal view, the head frame, placed around the head of the subject, facilitated both focusing and positioning of the head. The vertical arms of the frame were placed at the level of the exocanthions so that the plane of focus was approximately level with the lower orbital rim. When profile pictures were taken, one arm of the head frame was removed, and the other was used to position and maintain the facial midline in the vertical. The scale on the frame arms, which was on the same plane as the tip of the nose, was used to identify the focusing plane.

4.2 Standardised Head Position

Burstone (1958) stated that a standard method of subject positioning is essential and recommended the following procedure:

1. The sagittal plane is at right angles to the film with Frankfort horizontal parallel to the floor.
2. The teeth are in centric occlusion, for increasing or decreasing vertical dimension would influence the soft tissue structures.
3. The lips are lightly closed, neither overly relaxed nor tightly closed. In extreme malocclusions, such as Class II, division 1 cases, light closure does not allow the upper and lower lips to have complete contact.

Neger (1959) used a similar method for evaluating the soft tissue profile in a quantitative manner on a profile photograph. The head is positioned with the Frankfort plane parallel to the floor and the median sagittal plane of the patient parallel to the plane of the film, with the optical axis of the camera passing through orbitale. The patient is instructed to place his teeth in occlusion and to keep the lips relaxed and closed without exerting any undue force. If, because of the severity of

the malocclusion, the lips cannot be closed, the picture is taken with the lips in a parted position while maintaining occlusal contact.

4.2.1 Frankfort Horizontal Plane

The Frankfort Horizontal plane (FH) was defined at a conference of craniometrists held in Frankfort in 1884 as approximating to the true horizontal when the head is held in the normal postural position. It was originally designed to allow comparison of the skulls of different species, as well as races of man, and has been widely used for orientation of the head in cephalometric recordings (Houston, Stephens and Tulley, 1994).

However, Farkas (1981), found that placing the head in the FH position was uncomfortable and unnatural for many subjects. In most healthy subjects the rest position of the head is about 5 degrees above the FH. Therefore Farkas used 5 degrees above the FH as standard for his photography, obtained by a commercial angle meter and checked repeatedly throughout photography sessions.

4.2.2 Natural Head Position

Broca (1862) as cited by Moorrees and Kean (1958), defined the natural head position as "when a man is standing and when his visual axis is horizontal, he (his head) is in the natural position."

Moorrees and Kean (1958) investigated the constancy of the natural head position and found it extremely reliable for use in radiographic and photographic analyses, ie. a suitable standard of reference from which to measure the relationship of other craniofacial lines. Other studies have also shown the natural head position to be highly reproducible (Bean *et al.* 1970; Solow and Tallgren, 1971 and 1976; Foster *et al.* 1981; Lundstrom, 1981; Cooke and Wei, 1988b).

The methods used for obtaining the natural head position vary among authors. The method most commonly used has been to allow the subject orient the head by looking straight into a mirror or into the distant view through a window. Other studies used a light source (Cleall *et al.* 1966), while in others, no external reference was used.

Solow and Tallgren (1971) pointed out that determination of the head position by means of an external reference such as a mirror has the disadvantage, that the position obtained may not be the one habitually used by the subject in real life.

Solow and Tallgren (1971, 1976) investigated two different methods of recording natural head position:

1. Self-balance position, which is determined by the subject's own feeling of a natural head balance. This method has no external reference.
2. Mirror position which is determined by the subject looking straight into his eyes in the mirror, being the external reference.

The radiographs were recorded with the subject standing in the "orthoposition", defined as the intended position from standing to walking. Solow and Tallgren (1971, 1976) selected the orthoposition because it is a habitual symmetrical standing position and reproducible in postural investigations. Both recordings were made with the teeth in occlusion, but head postures achieved by two different methods showed significant differences from each other. The mirror position kept the head higher than in the self-balance position.

In some studies, the natural head position was recorded with subjects sitting in a relaxed position and looking into their own eyes in a mirror located at a fixed distance away from them (Moorrees and Kean, 1958; Bean *et al.* 1970; Larrabee *et al.* 1985).

It has been suggested that the physiologically determined head posture in a standing position is the appropriate orientation for lateral skull radiographs or profile photos. The natural head posture orientation has the advantage of being the position in which we normally observe the individual. This should present us with a profile view considered typical for the subject under investigation. It has been shown that lateral cephalometric radiographs or profile photographs arranged with respect to a particular cephalometric reference line risks incurring a forward or backward tip of the head (flexion or extension) from the natural head posture (Lundstrom, 1981).

Foster *et al.* (1981) believed that the true horizontal in cephalometric assessment would be more meaningful in aesthetic judgement. Contemporary orthodontics places more emphasis on facial profile and on incisor inclinations. These are judged clinically in relation to a true horizontal or vertical, with the head in its natural upright posture. They believed that the use of intracephalic reference lines was probably developed to overcome the variation of head positioning within the cephalostat. Variation in the reference line relationships was much greater than that due to head positioning errors. If the head is carefully placed in a natural posture position in the cephalostat, a true horizontal or vertical on the radiograph would give a more reliable assessment than the use of intracranial reference lines. There is the added benefit that the radiographic assessment would be based on the same standards as the clinical judgement on the patient.

Cooke and Wei (1988a,b) stated that natural head posture is the reproducible, natural, physiologic position of the head obtained when the relaxed subject looks ahead at an external eye reference - for example, a wall mirror.

Alternatively, a comfortable "self-balance" position of the head may be defined without any external eye reference. The true vertical and the true horizontal derived from it are usually used to define natural head posture and are themselves defined by the free position taken up by a weighted plumb line. Cooke and Wei

(1988b) found that analyses based on natural head position and the true horizontal are more clinically relevant, as they would more closely describe the morphology and appearance of the subjects as they truly appeared in life.

Cooke (1990) found that the natural head position deteriorated over time, but stabilises after 1 to 1 1/2 years. However, the variance of natural head position is found to be significantly less than the variance of intracranial reference planes with respect to the true vertical. Larrabee *et al.* (1985) used the natural head position to obtain reproducible photographic records. No head-holding apparatus was used as they believed, that they interfered with the visualisation of the face. In their profile analysis for facial plastic surgery, the true horizontal has substituted for the Frankfort horizontal plane, as they found that the former is more reliable than the Frankfort horizontal as estimated from photographs.

4.3 The Problem of Lip Posturing and the Reliability of Soft Tissue Landmarks

In the study of soft tissue profiles using standardised photography or cephalometric radiographs, there is the problem of lip posturing and the reliability and reproducibility of the various soft tissue landmarks. Lip position is affected by a number of variables including skeletal relationships, dental positions, soft tissue thickness as well as function. Postural variations of the lips introduce inaccuracies in the evaluation of the lip profile (Burstone, 1958 and 1967; Hillesund *et al.* 1978).

Burstone (1958, 1967) conceded that lip posture is a variant which cannot be completely standardised. Lip posture, like body posture, is a muscle-determined position. Therefore, it cannot have the reproducibility that is associated with measurements on hard structures.

Burstone (1967) described two postural positions of the lips:

1. Relaxed lip position, is when the lips are apart and hanging loosely with no effort made at lip contraction. In this position, an interlabial gap is present between the inferior surface of the upper lip and the superior surface of the lower lip. This gap varies with length and the occlusal vertical dimension as well as the magnitude of dental protrusion. Burstone believed that the relaxed lip position is reasonably reproducible, but, like all muscular positions is somewhat variable. Emotional states can strongly influence the contraction and relaxation of the muscle fibres of the lip.
2. Closed lip position, is when the lips are lightly touching in order to produce an anterior seal of the oral cavity.

Although, Burstone (1967, 1975) noted that the starting place for evaluating lip posture is the relaxed position, the patient normally does not assume this in daily activity. Rather, an effective lip seal is maintained to facilitate swallowing, protect the teeth and gingiva, and add retaining forces to maintain anterior teeth position. So, the postural lip position for the patient is closed during the day and apart during the night.

Burstone (1967) observed that there is a great variation in the reproducibility of lip position in patients with dentofacial disharmonies, such as in Class II division I and Class III malocclusions. Furthermore, there is no definite way of testing if the lips are truly relaxed. Burstone also noted that determination of rest position of the mandible is likewise not highly reproducible or easily obtained. If lip posture is to be evaluated, he believed that the vertical dimension of the jaws should be standardised. The simplest procedure is to have the teeth in occlusion. However, in certain conditions, such as marked overjet, in centric occlusion, the lower lip may be

deflected by the maxillary incisors. In such case, mandibular rest position so that a truer representation of lower lip posture may be achieved.

Hillesund *et al.* (1978) indicated that there is no significant difference in reproducibility whether lips are closed or in a relaxed state. In fact, the reproducibility of the lower lip in the vertical plane is significantly better when closed than when relaxed in the overjet and in the normal occlusion group, and concluded that to recommend any particular method (closed or relaxed lips) in the recording of soft tissue profile is not justified. However, if profile changes associated with orthodontic treatment are to be evaluated, it is best recorded in relaxed lip position, as the flattening of the lips in closed position will camouflage lip response to incisor retraction, particularly with large overjets.

In many soft tissue profile studies, there is disagreement as to the preferred lip position of the subjects. In attempts to study the effect of orthodontic retraction of incisors on the lips, some authors (Anderson *et al.* 1973; Oliver, 1982; Holdaway 1983a,b), took cephalograms with the lips in light contact both before and after treatment; whilst some (Bloom, 1961; Hershey, 1972; Rudee, 1964) took the radiographs with lips relaxed; and others (Garner, 1974; Roos, 1974; Huggins and McBride, 1975), did not specify whether a standardised lip position had been used.

Some authors (Wisth and Boe, 1975; Carlson and Wisth, 1972) indicated that soft tissue landmarks are more difficult to locate accurately than hard tissue on consecutive cephalograms of the same subject due to less well-defined anatomical structures, and variations in facial expressions. However, their investigation of the reliability of cephalometric soft tissue measurements, found that the errors of landmark location are generally the same for both soft and hard tissue, but variations in facial expressions can change the vertical position of the lip sulcus. They concluded that soft tissue variables showed the same errors as hard tissue variables. Thus, they can be used in clinical evaluations and scientific works with the same reservations as for the hard tissue variables, if cephalograms (or photographs) were taken with the jaws brought together in habitual occlusion and with lightly closed lips.

Tulley (1953) showed that orbicularis oris muscle activity reached a minimum when the mandible is in the habitual rest position. However, Wyke (1974) and Bando *et al.* (1972) found that this habitual rest position of the mandible is not a constant and varies in individuals constantly depending on factors such as head and body posture and emotional state.

Houston (1983) believed that the term reliability should be used to encompass both validity and reproducibility. The occurrence of systematic errors and random errors were discussed. Soft tissue cephalometric studies are particularly prone to

random errors induced by patient poses and the lack of precision in anatomical definitions that causes difficulty in landmark identification.

4.4 The Reliability of Photogrammetric Measurements

Accurate measurements that could be obtained by photogrammetry have benefits to the patient, the orthodontist and the oral-maxillofacial surgeon, as well as being economical.

Farkas' system of anthropometry involves many direct facial measurements. He determined how many of these measurements could be obtained reliably from photographs, that is, indirectly (Farkas *et al.* 1980). The study consisted of 36 healthy young Canadian Caucasians, comprising of 18 men and 18 women, whose soft tissue landmarks were marked on the skin of each subject with small ink dots (Figs. 4.1). Points in the facial midline, such as the profile line landmarks: tr, g, n, prn, sn, gn; were marked by a horizontal V-shaped sign. The tip of the sign touched the landmark, and the wings extended to the left.

Farkas took 104 direct measurements from the head, the face, and the ears. Following this he took standard frontal and left lateral black-and-white photographs of the subjects. Life-size photographs were printed. From these, another set of measurements were obtained and compared, in order to test the validity of the second, or indirect, set.

Kodak (Toronto, Canada) registration markers that matched the markers photographed on the head frame were applied to ensure that prints were life-size (1:1). Linear distances between landmarks were measured by a sliding calliper. Inclinations and angles were determined by a transparent protractor, and the degree of inclination calculated from the horizontal or vertical as defined by the FH.

Each measurement obtained from the print was compared with the corresponding direct measurement, and the difference was registered. The differences in all 18 subjects of each sex were then averaged.

Farkas regarded a measurement reliable only if the average difference between indirect and direct measurement was not greater than 1 mm or 2 degrees. Otherwise it was considered inaccurate, which were consistently longer or consistently shorter than the corresponding direct measurements or were combination of the two, depending on the individual. In selected measurements the extent of error was expressed as a percentage of the direct measurement. Since the analysis of measurements taken separately in boys and girls showed the same trend, the averages of the differences were calculated jointly.

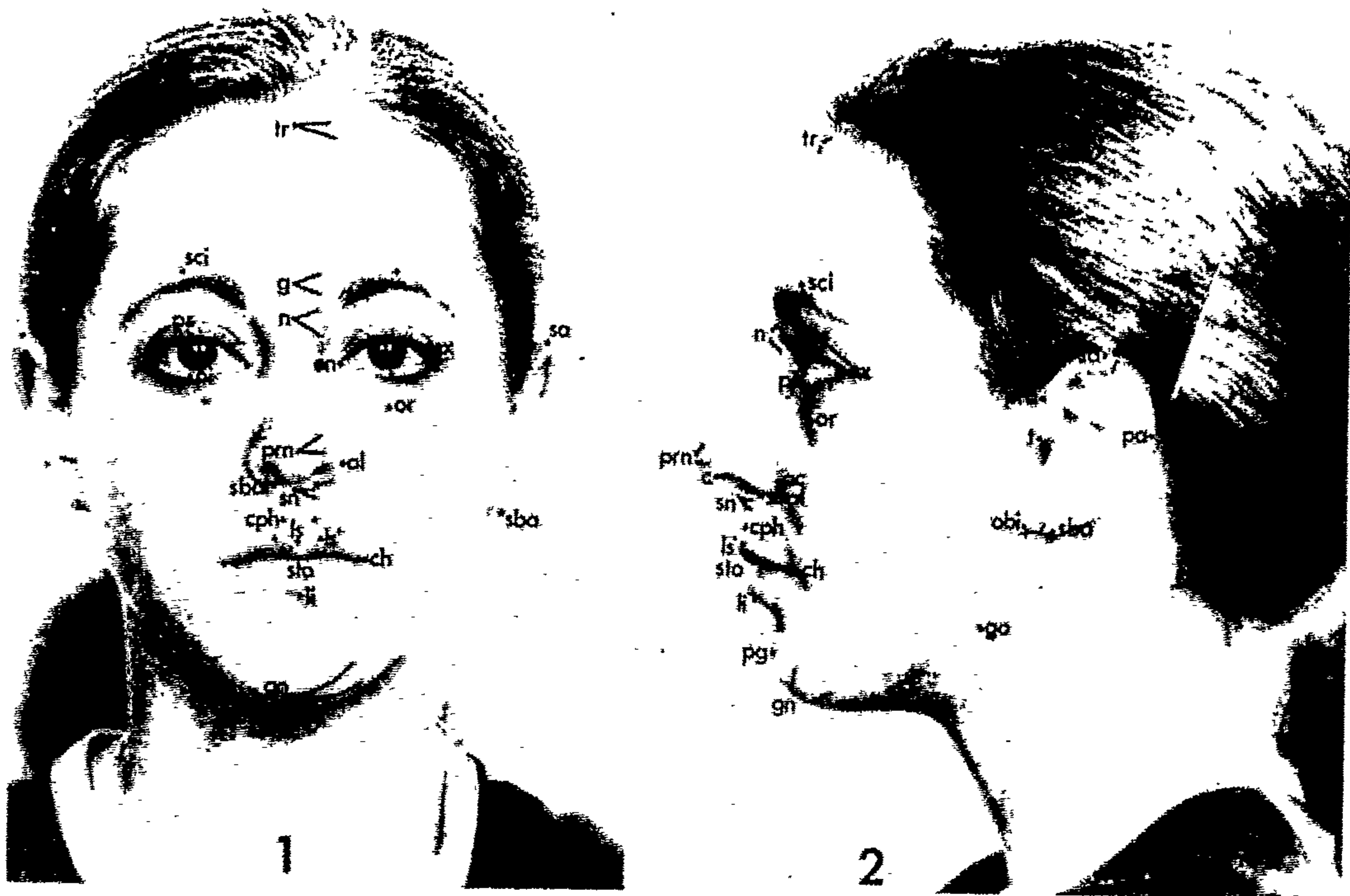


FIGURE 70. Landmarks on (1) the frontal print and (2) the left lateral print of the face. [Reprinted, by permission, from Farkas et al. (1980)]

Fig. 4.1: Landmarks on (1) the frontal print and (2) the left lateral print of the face. Note the use of V shaped markers. The tip of the sign touched the landmark, and the wings extended to the left (Reproduction of figure from Farkas et al. 1980).

The measurements taken from the 36 subjects in Farkas' study were both anthropometric and photogrammetric. There were 13 head measurements, 22 face measurements, 20 orbital measurements, 23 nasal measurements, 14 lip and mouth measurements and 12 ear measurements (Farkas *et al.* 1980).

Only 62 measurements were obtained from the two prints: 40.4 percent fewer than that obtained by direct measurement. The greatest reduction was in the head (53 percent), and the least the lips (seven percent).

4.4.1 Accuracy of Indirect Measurements

According to Farkas (1980), of the 62 indirect measurements, 20 (32.3 percent) were reliable, seven of which were from the lips and mouth. No accurate measurements of the ears were registered. The largest number (nine) of reliable measurements was of inclinations. Of the 62 measurements, 41 were unreliable. Three were consistently longer and 22 shorter than the identical direct measurements; the remaining 16 were mixed, since they were longer in some and shorter in others.

Consistently Longer Measurements: three measurements were consistently longer: the width of the face (zy-zy), by an average of 3.6 mm; the width of the nose (al-al), by 2.4 mm; and the width of the lower face (go-go), by 21.6 mm. All of these measurements were horizontal and obtained from frontal prints.

Consistently Shorter Measurements: 11 measurements were consistently shorter; more were horizontal (15) than vertical (seven). The greatest shortening (17.6 mm) was in the distance between the nasion (n) and the tragion (t) on the profile print. On the frontal print, the greatest difference (5.5 mm) was in halves of the labial fissure (ch-sto).

Mixed Differences in Measurements: 16 measurements had mixed differences, depending on the individual facial characteristics of the subjects. Of these, 12 (seven of which were vertical measurements) were taken from profile prints, and 4 (three of which were horizontal measurements) were taken from frontal prints. The greatest differences in measurements were in angles. The linear measurements differed little from the equivalent direct measurements. In general, vertical measurements showed smaller differences than horizontal measurements (Farkas *et al.* 1980).

4.4.2 Identification of Profile Landmarks

Landmarks of the facial profile and the forehead (n, prn, sn, sto, pg, gn, tr, g) are not always visible on the lateral print, even when marked on the skin before photography (Farkas, 1981). In profile, flattening along the axial line, namely the root of the nose, tip of the nose and chin, and in some cases, a slight axial depression at the base of the columella, in the middle of the border of the upper vermilion and on the chin make landmarks invisible on lateral prints. From the side, the glabella can be blocked by a bushy eyebrow, and the trichion can be hidden by the hair. Dots on landmarks on the profile line cannot be utilised. However, with the exception of the glabella, which often is covered by hair, the use of V-shaped markers as described by Farkas, are helpful in locating these landmarks (Farkas *et al.* 1980).

A) Measurement from Bony Landmarks

Direct measurements between bony landmarks (e. g., zy-zy, gn-go) are taken by calliper, with the tips of the instrument pressed to the bony surface. As this cannot be conducted on prints, errors occur even when landmarks are marked on the skin before photography. Farkas (1980) stated that errors may be much greater if the anatomical points are left unmarked.

B) Difficulty in Identifying Landmarks Located on the Edges, or Contours, of Anatomical Features

Some landmarks cannot be marked, such as the inner or outer commissure of the eye fissure, and others, such as the most lateral point of the ala are not always seen clearly because the anatomical feature bearing the landmark may not be sharp enough on the photograph. This may be due to the differing degrees of reflection in various facial surfaces, but such errors are usually minimal (Farkas, 1981).

C) Landmarks Covered By Hair Or Hidden Behind Facial Features

Indirect measurement of the length, width, or height of the head produced significant errors unless the subject was bald. However, locating the head landmarks (v,eu,op) may be possible using a head frame device developed by Farkas (1981).

Farkas stated that the widest diameter of the head (eu-eu) can be determined from the frontal print if the perpendicular arms of a frame (which had ruler markings) are positioned in the closest possible contact against the head at its greatest width during photography. In a similar manner, the horizontal bar of the frame can be used to identify the vertex on the profile photograph if it was lowered to the patient's vertex.

The view of other landmarks may be blocked by some features. For example, on the profile photograph, the porion landmark may be hidden by the tragus, or the commissure of the lip may be covered by a skin fold. Similarly, on the frontal print, the exocanthion landmark cannot be seen clearly in all subjects.

4.4.3 Inaccuracies in Measurement of Angles

Inaccuracies in angle measurement occur because the lateral print does not reflect precisely the true facial contour. If there is flattening or depression in the midline of the face, the print profile will differ from the true profile. This affects the accuracy of the inclinations and the angles measured on the print, and explains the relatively wide range of error (± 5 degrees) in assessing the nasolabial and nasofrontal angles (Farkas *et al.* 1980).

4.4.4 Errors Introduced by Photographic Distortion

Even when photographic distortion is reduced to acceptable levels, a certain amount of distortion cannot be avoided in the plane on which the camera is focussed, due to the uneven topography of the face.

In the study conducted by Farkas (1981), the greatest distortion occurred between the distance between the base of the columella (subnasale) and the tip of the nose (pronasale). This was 46 percent shorter on the print than on the subject because of the great difference of level between the two landmarks. On the other hand, the measurement between the root of the nose (n) and the labial fissure (sto) was accurate because of the similar relationship of the two landmarks to the focusing plane.

When photographing facial profile, Farkas (1981) recommends specific geometric areas for focusing. The degree of distortion depends on the differences in the level of the individual facets. Because the print is two-dimensional, measurement of the surface length of the face or of its parts, such as the supraorbital rim, nasal ala length, maxillary and mandibular arcs, is impossible. Sagittal measurements such as nasal protrusion (sn-prn) taken from the frontal-view prints are markedly distorted or cannot be taken at all, as in the case of the depth of the nasal root.

4.4.5 Which View is of the Greatest Value in Photogrammetry?

Farkas' study (1981) concluded:

- 1) Frontal-view and profile prints each allowed 10 reliable measurements. Of the 20 reliable measurements, nine were inclinations; eight of these were vertical and three were horizontal. Frontal-view prints supplied the most precise measurements of orbits, lips, and the mouth. However, the profile prints generally were more useful, since a number of vertical measurements in these differed from the corresponding direct measurements by little more than 1 mm.
- 2) Another useful view is the base view of the soft nose, which shows the structures surrounding the nostrils. In order to obtain accurate measurements from this special view the plane of the nasal ala must be parallel to the camera lens.

Three reports compared the reliability of photogrammetry to anthropometry (Tanner and Weiner, 1949; Gavan *et al.* 1952; Fraser and Pashayan, 1970) but mentioned only 16 of Farkas' 104 measurements. Only some of these 16 measurements show the same trend as in Farkas' study, but the discrepancies may be caused by differences in marking technique and head positioning (Tanner and Weiner, 1949; Fraser and Pashayan, 1970). The sample of Gavan *et al.* (1952), which consisted of two subjects, was too small for valid conclusions, but it is the only study that expresses differences quantitatively.

Photogrammetric landmarks in the profile view were chosen for the present study because these were the most reliable ones according to Farkas. The details of these are given in Chapter 7.

4.4.6 Photogrammetry and Facial Proportions

The relative proportions of individual features determine the rich variations in facial structures. Farkas (1981) felt that subtle differences of size and shape easily identified by measurement can be recognised visually only by a trained examiner. Greater disproportions are easy to detect, although pointing to the specific dimension(s) causing the unusual relationship is difficult. To study proportions, a facial dimension such as eye fissure length or upper lip height, that has minimal changes during postnatal development is selected, and the remaining measurements are expressed as percentages of that dimension.

Photogrammetry has been used in many studies of facial proportions (Broadbent and Mathews, 1957; Gonzalez-Ulloa, 1962; Seghers *et al.* 1964; Patterson and Powell, 1974) without the support of direct measurements. The inaccuracies that were observed in Farkas' (1981) study indicate some of the problems associated with evaluation of proportions from photographs. In addition, before photographs are used for analysing proportions, the formulas for determining proportions need to be verified. Classic formulas of idealistic proportions of the head, face, and ears in adults were developed by the Egyptians and adopted by ancient, Medieval and Renaissance artists. However, Farkas (1981) felt that the formulation of new rules for determining ranges for normal variation with the aid of direct measurements are also necessary.

The use of a proportion index comprising a few anthropometric data permits complex judgment of the face and establishes a base for a detailed comparison superior to the analysis of single measurements (Ainsworth *et al.* 1979). Farkas tested the validity of two basic facial proportions formulas using data for six and 18 year olds. Contrary to classic rules, they found that the three parts of the face were not equal, and in 97 of the 101 girls and in all 102 boys the labial fissure was below ideal level. Although standard photographs help visualise the areas of marked change in a patient's face, they are not adequate substitutes for the live face. Therefore face proportion must be judged carefully from photographs as disproportion can result from or be increased by photographic distortion.

Farkas (1981) concluded: "...At this time the number of reliable measurements is too small to justify the exclusive use of photogrammetry in the study of facial proportions. However, with improved photographic techniques, photographs may provide an ideal tool for studying the proportions of the head, the face, and the ears."

**SECTION III:
ORIGINAL WORK**

CHAPTER 5.
PART I: CEPHALOMETRIC STUDY

5.1 Aims of the Study

A cephalometric study of 11 patients of Caucasian background(five females aged between six and 22 years and six males aged between 11 and 25 years) was conducted to increase our understanding of the extent of underdevelopment of the facial skeleton in CDP and BS and to evaluate whether the Syndrome affected males and females differently.

In the present study, the majority (nine) were diagnosed with CDP (four females and five males) and two with Phenytoin embryopathy (a female and a male) who were diagnosed with BS, possibly because they presented later in life and radiographic evidence of punctate calcification in infant bones were missed had it been present (Table 5.1).

Table 5.1: Diagnoses for cephalometrically measured sample.

PATIENT / sex / age	DIAGNOSIS
(A) / Female / 6yrs	CDP
(B) / Male / 11yrs	CDP
(C) / Female / 12yrs	CDP
(D) / Female / 14yrs	BS (PHENYTOIN)
(E) / Male / 14yrs	CDP
(F) / Male / 17yrs	CDP
(G) / Male / 19yrs	BS (PHENYTOIN)
(H) / Female / 20yrs	CDP
(I) / Male / 21yrs	CDP
(J) / Female / 22yrs	CDP
(K) / Male / 25yrs	CDP

5.2 Materials and Method

All cephalograms chosen in the experimental cohort were standardised by being taken at the same institution, the Royal Children’s Hospital in Melbourne, between 1987 and 1990, with the majority taken in 1987 and 1988. All cephalograms were taken with a 1980 *Siemens Orthoceph 5* machine Model 542600 ZX; Serial number 07651507; Model number 777340 V1010. The unit had the following specifications : an Ortho G regular cassette, an A1 filter block, and an aluminium 3+ filter which is used for soft tissues of the facial profile. There was a forehead, nose and chin vertical. This soft tissue filter was designed to be thinner medially towards the bony skeleton and gradually thicker as one approaches the forehead, nose, lips and chin.

The protocol for lateral cephalograms used is the one standardised for that hospital in 1980. The patient is positioned in true lateral position, with lips together

and teeth lightly touching, with their head held in a cephalostat with ear-rods and a scale placed on the nasal bridge. If the subject is older than 5 years of age, they are told to stand, but if there is a small child or an invalid, it is more practical to have them seated.

The exposures used were also standardised using age as a guide. For example, an adult 18 years old and over had 70 kV for 1.6 seconds. An 8 year-old had 70 kV for 1.2 seconds, and a child 3 years old had 60 kV for 1.0 seconds. These were adjusted also based on relative body mass and build.

A limitation to the present thesis data was that all the radiographs in the present study were copies of those taken from the Royal Children's Hospital in Melbourne. Despite these drawbacks, the radiographs measured were of an appropriate diagnostic quality for hard and soft tissue analysis, as any poorly duplicated radiographs were rejected. A standardised enlargement factor of 13.3% was used, and calculated by the known subject to film distance, fixed at 18 cm, and the anode to subject distance, fixed at 135 cm. This enlargement factor compared well with that of the normative data from Riolo (1974), which was 12.7%.

The hard tissue cephalometric analyses proposed by Leonard and Walker (1977); Zide, Grayson and McCarthy (1981); Horswell *et al.* (1988) were used in this study, as this analysis indicates proportionately the relations between the midface, cranial base and mandible, and how these structures may be affected in conditions such as CDP or BS. Vertical hard tissue facial measurements were added to this study, to assess proportions of the anterior and posterior facial heights.

Cephalometric measures were compared to normative data from Riolo (1974) and McNamara (1984). The angular measurement (SNO) was compared with the normal SNO measurement as reported by Leonard and Walker (1977). The skeletal angle of convexity standard (N-A-Pog) was taken from a study from The Child Research Council at the University of Colorado (Burstone 1978).

For the analysis of soft tissues, the 11 measurements used in Holdaway's analysis (1983a) were chosen, due to its common usage amongst orthodontists. To supplement these soft tissue measurements, the nasofrontal and nasolabial angles were used, the normal ranges for these two angles were taken from Powell and Humphreys (1984).

5.3 Selection of Hard Tissue Reference Points

The following points are used in this cephalometric analysis:

1) S-N (Sella-Nasion): Anterior cranial base length.

This measurement joins the "sella", defined as the centre of Sella Turcica by inspection, and the "nasion," which is the junction between the frontal and nasal

bones at the frontonasal suture, seen as the most posterior point on the curve of the bridge of the nose.

2) SE-PNS (Spheno-ethmoidal juncture to posterior nasal spine):

Posterior maxillary height.

This measurement joins the "ethmoid registration point," which is intersection of the sphenoidal plane with the averaged greater sphenoid wing, and the "posterior nasal spine" or the most posterior point at the sagittal plane on the bony hard palate (Riolo *et al.* 1974).

3) PNS-A (Posterior nasal spine to point A): Total maxillary length.

This measurement joins the "posterior nasal spine" with "point A", defined as the most posterior point on the curve of the maxilla between the anterior nasal spine and supradentale (Riolo *et al.* 1974). Due to the diminished size or absence of the anterior nasal spine in these syndromic subjects, point A was even more difficult to identify.

4) Co -A (Condylion to point A): Effective maxillary length.

This measurement joins the "condylion" and "point A". "Co" or condylion is the most posterior superior point on the curvature of the average of the right and left outlines of the bony condylar head. Determined as the point of tangency to a perpendicular construction line to the anterior and posterior borders of the condylar head. The "Co" point is, therefore, located as the most superior axial point of the condylar head rather than the most superior point on the condyle (Riolo *et al.* 1974).

5) Co-Gn (Condylion to Gnathion) : Effective mandibular length.

This measurement joins the "condylion" and "gnathion" the most anterior-inferior point on the contour of the bony chin symphysis, and is determined by bisecting the angle formed by the mandibular plane and a line through pogonion and nasion (Riolo *et al.* 1974).

6) SNO (Sella-nasion-orbitale angle): Orbital position relative to S-N.

This measurement was first introduced by Leonard and Walker (1977) and comprised of the angle formed by two lines: one joining sella and nasion (the anterior cranial base) and the other joining nasion and orbitale, which is the lowest point on the average of the right and left borders of the bony orbit (Riolo *et al.* 1974).

Leonard and Walker (1977) regarded this measurement as a "...significant guide to the malar-maxillary relationship and will assist in complete cephalometric evaluation of these patients [individuals with midfacial hypoplasia at the Le Fort II level] ." Prior to this cephalometric criterion for diagnosing midfacial hypoplasia had been principally dependant on an SNA of less than 79 degrees. However, this measured the retrusion of point A relative to the anterior cranial base and meant that the maxilla at that level (Le Fort II) is diminutive provided that the SN plane is

horizontal to Frankfort Horizontal or within 8 degrees of being so. Thus it does not follow that the malar eminences and infraorbital rims are diminutive.

In the search for a measurement that could aid in the diagnosis of midfacial hypoplasia at the Le Fort II level, Leonard and Walker (1977) tested the hypothesis that it should be possible to establish a normal range for their new angle, angle SNO, if SNA is in the normal range (83 ± 1 SD = 80 to 86 degrees).

Records were obtained from 168 Caucasian American women aged 15 years or older with an SNA of 69 to 79 degrees. They felt that there was "good evidence to show that, at this age, any further growth of the upper part of the face has virtually ceased in a woman and that there is little difference in the maxillary growth between the sexes after this age" (Leonard and Walker, 1977). Two groups were studied: the first group consisted of those with an SNA between 80 and 86 degrees and the second group had an SNA < 79.9 degrees. Patients in group 1, had an average value for SNO of 57.8 degrees with a standard deviation of ± 4.18 degrees. Patients in group 2, had an average value for SNO of 54.5 degrees with a standard deviation of ± 3.8 degrees: thus an average retruded case could be expected to have an SNO angle between 50 and 58 degrees. If SNO < 50 degrees, then an unusual degree of maxillary retrusion at orbitale was expected.

7) N-A-Pog (Nasion-point A-pogonion angle): Skeletal angle of facial convexity.

The Down's skeletal angle of convexity describes the concavity or convexity of the profile, and is comprised of the angle formed by two lines - one from nasion to point A and the other from pogonion (the most anterior point on the contour of the bony chin, determined by a tangent through nasion) to point A. The mean is 180 degrees ranging from +10 degrees to -8.5 degrees, with a standard deviation of 5.1 degrees. This compares with a study by Riedel (1952) of an adult group aged 18 to 36 years and a group of children aged eight to 11 years; yielding +1.6 degrees for the adults and +4.2 degrees for the children. Normally, there is a steady decrease of this angle with age, as the face becomes less convex.

8) Co-A to Co-Gn: (Mandibular-maxillary unit length differential).

The differential measurement between mandibular and maxillary lengths to assess relative jaw proportion. This is calculated by the maxillary unit length (Co-A) being subtracted from the mandibular unit length (Co-Gn). Differences toward either end of the sample range is indicative of a mismatch between maxillary and mandibular lengths (dysplasia). The norms used were calculated from Michigan standards (Riolo *et al.* 1974).

9) N-ANS (Nasion-Anterior nasal spine) : Upper anterior face height.

The linear measurement between nasion and the anterior nasal spine. The anterior nasal spine is the tip of the median, sharp bony process of the maxilla at the lower margin of the anterior nasal opening.

10) ANS-Me (Anterior nasal spine-Menton): Lower anterior face height.

The linear measurement between the anterior nasal spine and menton (the most inferior point on the symphysial outline of the mandible). Lower anterior face height is an indicator of mandibular vertical position. Harvold (1971) measured this distance from a point on the superior contour of the anterior nasal spine, where the vertical thickness is 3mm.

11) N-Me (Nasion-Menton): Anterior face height.

The linear measurement between nasion and menton.

12) S-Go (Sella-Gonion): Posterior face height.

The linear measurement between sella turcica and gonion, the midpoint of the angle of the mandible. Found by bisecting the angle formed by the mandibular plane and a plane through articulare posterior and along the portion of the mandibular ramus inferior to it.

13) S-Ar (Sella-Articulare)

The linear measurement between sella turcica and articulare (the point of intersection of the inferior cranial base surface or basisphenoid and the averaged posterior surfaces of the mandibular condyles).

14) Ar-Go (Articulare posterior-Gonion)

The linear measurement between articulare and gonion.

15) PFH:AFH (Posterior face height [S-Go] to anterior face height [N-Me] ratio)

The ratio formed by dividing the posterior face height by the anterior face height and expressing the ratio as a percentage. A subnormal ratio indicates that the posterior face height is relatively shorter than the anterior face height giving an anterior divergence to the profile. A ratio in excess of the normal value indicates that the posterior face height is proportionately long for the anterior face height giving posterior divergence to the profile.

16) UFH:LFH (Upper anterior face height [N-ANS] : Lower anterior face height [ANS-Me] ratio)

The ratio formed by dividing the upper anterior face height by the lower anterior face height and expressing the ratio as a percentage. A subnormal ratio indicates that the upper face height is short for the lower face height, and a greater than normal ratio indicates that the upper face height is long relative to the lower face height.

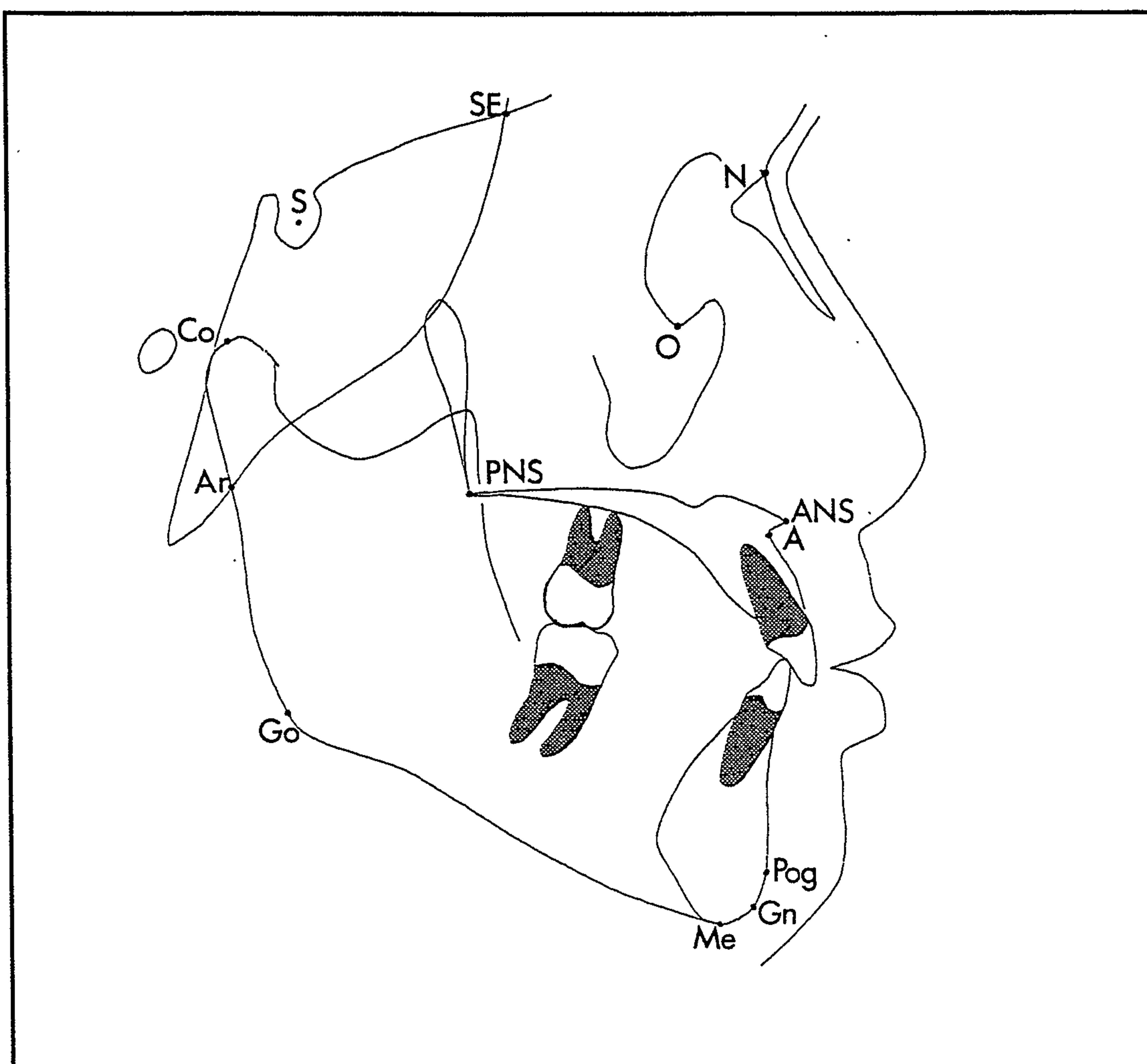


Fig. 5.1: Diagram of hard tissue points used in this study.

5.4 The Measurements Used in the Soft Tissue Analysis

- 1) ***Soft Tissue Facial Angle***: this is an angular measurement of a line drawn from soft tissue nasion, where the sella-nasion line crosses the soft tissue profile, to the soft tissue chin at a point overlying the hard tissue suprapogonion of Ricketts measured to the Frankfort horizontal plane (Holdaway 1983a).
- 2) ***Nose Prominence***: measured by a line perpendicular to Frankfort horizontal and running tangent to the vermilion border of the upper lip. This measures the nose from its tip in front of the line and the depth of the incurvation of the upper lip to the line (Holdaway 1983a).
- 3) ***Superior Sulcus Depth***: this is a measurement of the upper lip form or curl, and is measured to a perpendicular to Frankfort and tangent to the vermilion border to the upper lip (Holdaway 1983a).
- 4) ***Soft Tissue Subnasale To H-Line (Harmony Line)***: the distance of soft tissue subnasale to the Harmony line (drawn tangent to the soft tissue chin and upper lip). In cases to be found on either extreme of the facial convexity spectrum, the ideal measurements to the H-line lose their significance because of the change in the cant of the H-line (Holdaway 1983a).
- 5) ***Skeletal Profile Convexity***: this is a linear measurement from point A to the hard tissue line Na-Pog or facial plane. This is not really a soft tissue measurement but convexity can be directly interrelated to harmonious lip positions and, therefore, has a bearing on the dental relationships needed to produce harmony of features of the face (Holdaway 1983a).
- 6) ***Basic Upper Lip Thickness***: this is near the base of the alveolar process, measured about 3 mm below point A. It is at a level just below where the nasal structures influence the drape of the upper lip (Holdaway 1983a).
- 7) ***Upper Lip Strain Measurement***: the usual thickness at the vermilion border level is 13 to 14 mm. Excessive taper is indicative of the thinning of the upper lip as it is stretched over protrusive teeth; also, excessive vertical height may produce more than 1mm of taper due to lip stretching. When lip thickness at the vermilion border is larger than the basic thickness measurement, this usually identifies a lack of vertical growth of the lower face with a deep overbite and resulting lip redundancy (Holdaway 1983a).
- 8) ***H- Angle***: this is an angular measurement of the H-line to the soft tissue Na-Pog line or soft tissue facial plane (Holdaway 1983a).
- 9) ***Lower Lip to H-Line***: measured from the most anterior point on the lower lip to the H-line, the ideal being 0 to 0.5 mm anterior, but -1 mm behind to +2 mm in front is considered acceptable (Holdaway 1983a).

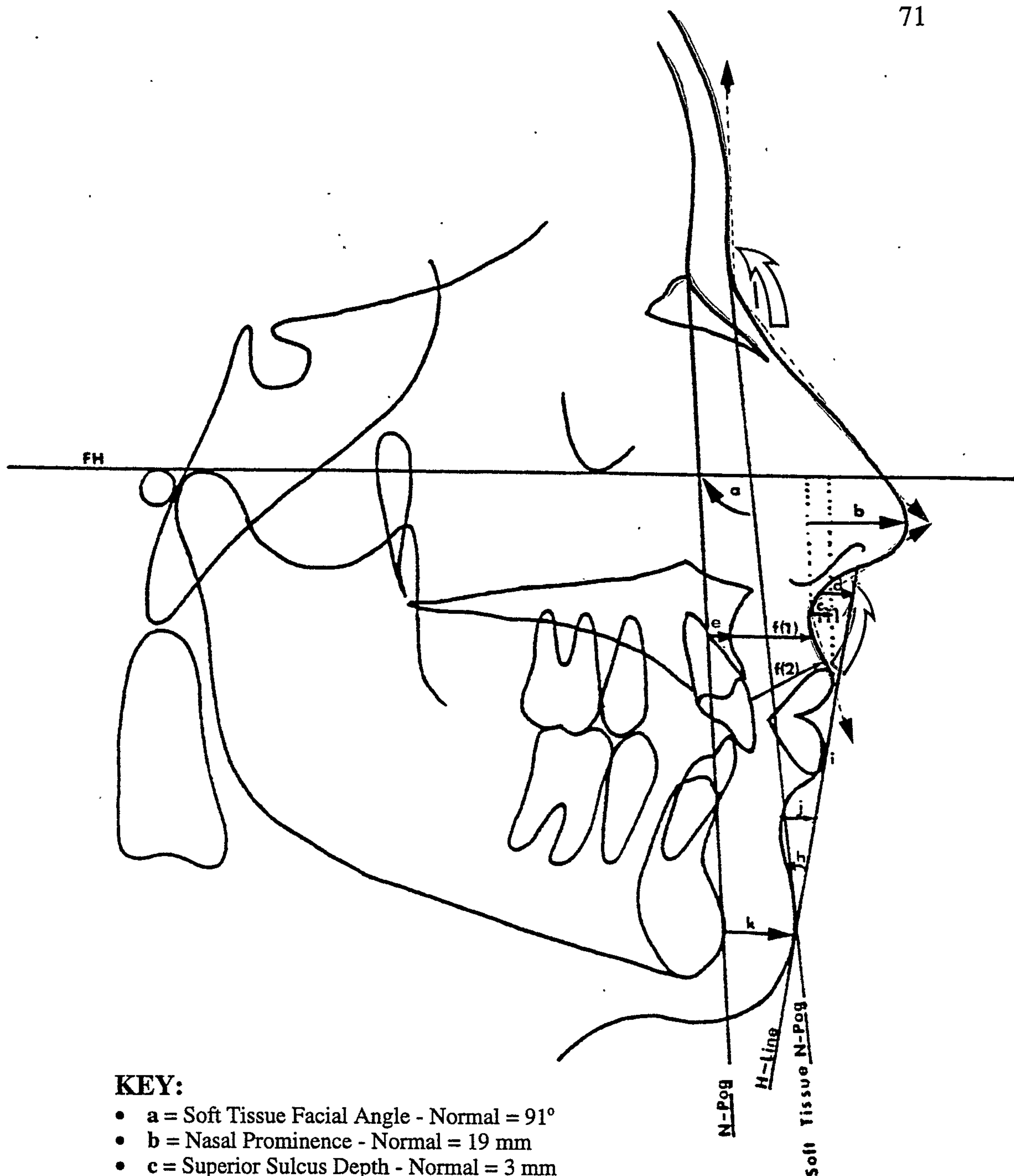
10) ***Inferior Sulcus to H-Line***: measured at the point of greatest incurvation between the vermillion border of the lower lip and the soft tissue chin and is measured to the H-line (Holdaway 1983a).

11) ***Soft Tissue Chin Thickness***: recorded as a horizontal measurement and is the distance between the two vertical lines (10 - 12 mm average) representing the hard tissue and soft tissue facial planes at the level of Ricketts' suprapogonion (Holdaway 1983a).

12) ***Nasofrontal Angle (N-Fr)***: An important angle of the upper face, the nasofrontal angle is a distinct, smooth transition between the nasal dorsum to the glabellar forehead region. It is created by the line tangent to the glabella through soft tissue nasion, intersecting with a line tangent to the dorsum. The range of an aesthetic measurement for this angle, according to Powell and Humphreys (1984), is from 125 degrees to 135 degrees. The importance of this measurement lies in the understanding that a change in angulation will affect nasal aesthetics, namely nasal projection. A change in the position of the angle's vertex superiorly or inferiorly will lengthen or shorten the nasal length respectively.

13) ***Nasolabial Angle (LS-Sn-Cm)***: This angle is measured from a line tangent to the most anterior point of the columella (Cm) to the subnasale (Sn), the point where the columella merges with the upper lip, and a line intersecting Sn and the mucocutaneous border of the upper lip (LS), after Powell and Humphreys (1984).

This important angle measures the inclination of the columella in relationship to the upper lip. A dental or skeletal maxillary or mandibular disharmony can cause a marked impact on this angle, which ideally should be in the range of 90 degrees to 120 degrees (Powell and Humphreys 1984).



KEY:

- **a** = Soft Tissue Facial Angle - Normal = 91°
- **b** = Nasal Prominence - Normal = 19 mm
- **c** = Superior Sulcus Depth - Normal = 3 mm
- **d** = Soft Tissue Subnasale to H-Line - Normal = 5mm (± 2)
- **e** = Skeletal Profile Convexity - Normal = 1mm
- **f(1)** = Upper Lip Thickness at A-Point - Normal = 14 mm
- **f(2)** = Upper Lip Thickness at Upper Incisor - Normal = 13 mm
- **g** = Upper Lip "Strain" = $f(1) - f(2)$ - Normal = 0-1 mm
- **h** = H - Angle - Normal = 10°
- **i** = Lower Lip to H-Line - Normal = 0 mm
- **j** = Inferior Sulcus Depth - Normal = 3 mm
- **k** = Soft Tissue Chin Thickness - Normal = 14 mm
- **l** = Fronto-Nasal Angle - Normal = $125^\circ - 135^\circ$
- **m** = Nasolabial Angle - Normal = $90^\circ - 120^\circ$

Fig. 5.2: Diagram of measurements used in the soft tissue analysis.

(Norms for **a** - **k** from Holdaway, 1983a; **l** and **m** from Powell and Humphreys, 1984).

5.5 Method Used for Measurement

All 11 radiographic cephalograms used were standardised by being taken at the Royal Children's Hospital in Melbourne in true lateral position (Frankfort Horizontal parallel to the floor) with teeth and lips lightly touching.

An important limitation to this project was that all radiographs were copies of those taken between 1987 and 1990. The radiographs used were of appropriate diagnostic quality for hard and soft tissue analysis, as poorly duplicated radiographs were rejected.

Cephalometric measures were compared to normative data from Riolo (1974) and McNamara (1984). The angular measurement (SNO) was compared to the normal SNO measurement (Leonard and Walker 1977). The skeletal angle of convexity standard (N-A-Pog) was taken from a study from The Child Research Council at the University of Colorado (Burstone 1978).

All cephalograms were traced manually by the author on a *Rayvue* variable-intensity lightbox in a darkened room, using a 0.3mm 3H mechanical pencil onto 0.003" matte acetate orthodontic tracing paper which had been affixed to the radiograph with clear adhesive tape. A matte black card with a cut out window was used to blank out peripheral light around the radiograph. The radiograph was oriented with the Frankfort plane parallel to the bottom edge of the screen, because a number of landmark definitions depend on head orientation. All tracings were completed within the same day, to minimise systematic error (Houston 1983). An Ormco cephalometric ruler-protractor was used for all hard and soft tissue measurements, which were recorded to the nearest millimetre.

5.6 Reliability of Measurements

An error study was carried out on the 11 radiographs for all the measurements, which were re-traced and re-measured on a separate occasion by the author.

Paired Student t-tests were computed at the 95 percent confidence interval for each of the following soft tissue and hard tissue variables to test the null hypothesis that there was no difference between the two sets of measurements. The results, showing the mean difference, standard error of the mean, t-test value and the probability for each of the variables measured are listed in Tables 5.2 and 5.3. The degrees of freedom are 10. The results indicate that the tracing, landmark location and measurement method is reliable.

Table 5.2: Results of double determinations on 11 lateral cephalograms for soft tissue variables.

SOFT TISSUE VARIABLE	MEAN [†]	SE	t	p
1. Facial Angle	0.600	0.356	1.69	0.13
2. Nose Prominence	-0.045	0.081	-0.056	0.59
3. Superior Sulcus Depth	0	0.095	0	1.00
4. Subnasale to H-Line	0	0	0	---
5. A to N-Pog	0	0	0	---
6. Upper Lip Thickness	0	0	0	---
7. Upper Lip Strain	0.045	0.142	0.32	0.76
8. H-Angle	0	0	0	---
9. Nasolabial Angle	0	0	0	---
10. Frontonasal Angle	0	0	0	---
11. Lower Lip to H- Line	0	0	0	---
12. Inferior Sulcus to H-Line	0	0.095	0	1.00
13. Soft Tissue Chin Thickness	0.045	0.045	1.00	0.34

[†] = mean values of 0 indicate that there were no differences between any of the 11 double-determinations (to nearest millimetre).
* = Statistically significant difference at p <0.05
** = Statistically significant difference at p <0.01

Table 5.3: Results of double determinations on 11 lateral cephalograms for hard tissue variables.

HARD TISSUE VARIABLE	MEAN	SE	t	p
S-N	-0.091	0.356	-0.26	0.80
Co-Gn	0.045	0.125	0.36	0.72
SNO	0.273	0.141	1.94	0.082
PNS-A	0.218	0.139	1.57	0.15
N-A-Pog	-0.009	0.148	-0.06	0.95
Se-PNS	-0.136	0.097	-1.40	0.19
Co-A	0.227	0.304	0.75	0.47
N-ANS	-0.136	0.136	-1.00	0.34
ANS-ME	-0.364	0.166	-2.19	0.054
N-ME	0.136	0.119	1.15	0.28
S-Go	-0.091	0.222	-0.41	0.69
S-Ar	0.091	0.113	0.80	0.44
Ar-Go	0.136	0.136	1.00	0.34
PFH-AFH	-0.255	0.148	-1.72	0.12
UFH:LFH	-0.009	0.170	-0.05	0.96

* = Statistically significant difference at p <0.05
** = Statistically significant difference at p <0.01

As Houston (1983) recommended, in order to reduce those random errors which may arise from point identification rather than in measurement, the tracings were replicated and the two sets of measurements were averaged for the comparative studies between the sample groups and the norms.

CHAPTER 6:
FINDINGS OF THE CEPHALOMETRIC STUDY

In this study, a total of 11 lateral cephalometric radiographs were measured, with five females aged six to 22 years and six males aged 11 - 25 years.

6.1 Hard Tissue Comparison between the Sample Group in the Present Study and Normal Values Matched for Age and Sex

Double determinations of the patients' hard tissue cephalometric variables were averaged and then compared with normal values matched for age and gender. Paired Student t-tests of the mean differences revealed eight out of the sixteen hard tissue variables were significantly different from the norm. The results are tabulated in Table 6.1.

Table 6.1: Results of paired Student t-tests for comparison between sample group in the cephalometric study and normal values.

Variable	N	Mean	SD	SE	t	p
S-N	11	6.76	5.00	1.25	5.40	0.0001**
Co-Gn	11	-1.32	8.13	2.03	-0.65	0.52
SNO	11	-2.91	5.29	1.32	-2.20	0.044*
PNS-A	11	4.96	4.10	1.03	4.83	0.0002**
N-A-Pog	11	10.49	10.67	2.76	3.80	0.0019**
SE-PNS	11	-2.76	5.42	1.36	-2.04	0.060
Co-A	11	9.26	6.29	1.57	5.89	0.0005**
Mx-Md.	11	-14.80	10.28	2.57	-5.76	0.0005**
N-ANS	11	-2.85	4.29	1.29	-2.21	0.052
ANS-Men	11	-3.40	4.95	1.49	-2.28	0.046*
N-Men	11	1.70	8.55	2.58	0.66	0.52
S-Go	11	-0.72	5.28	1.59	-0.45	0.66
S-Ar	11	-0.250	3.136	0.946	-0.26	0.80
Ar-Go	11	0.30	5.22	1.57	0.19	0.85
PFH:AFH	11	-1.232	3.114	0.939	-1.39	0.22
UFH:LFH	11	8.41	5.96	1.80	4.68	0.0009**

* = Statistically significant difference at p <0.05
** = Statistically significant difference at p <0.01

Statistically significant differences ($p < 0.05$) were found for the following seven variables, the graphs of which are seen from Figs. 6.1 onwards:

- 1) (Sella - Nasion): Reduced anterior cranial base length (S-N). Seen in Fig. 6.1.
- 2) (Sella - Nasion - Orbitale): Position of orbitale relative to SN was recessive. Seen in Fig. 6.3.
- 3) (Posterior nasal spine to point A): Reduced total maxillary length (PNS-A). Seen in Fig. 6.2.
- 4) (Nasion - point A - pogonion angle): Reduced skeletal angle of facial convexity (N-A-Pog). Seen in Fig. 6.6.
- 5) (Condylion to point A): Reduced maxillary length (Co-A). Seen in Fig. 6.4.
- 6) Mandibular-Maxillary differential (Md-Mx): Calculated as $Co - Gn : Co - A$, the difference between horizontal lengths of the jaws was increased. Seen in Fig. 6.5.
- 7) (Anterior nasal spine-Menton): Increased lower anterior face height (ANS-Men or LFH). Seen in Fig. 6.8.
- 8) (UFH:LFH): Increased upper to lower anterior face height ratio. Seen in Fig. 6.9.

The variables SNO (Fig 6.3), N-ANS and Se-PNS show some evidence that the observed values may be different from the normal values, but must be interpreted cautiously as the sample size is small.

6.2 Hard Tissue Comparison between the Sexes in the Sample Group

Two-sample t-tests were carried out at the 95 percent confidence interval to assess whether males and females varied in the way they differed from the normal values, in CDP. Five females and six males were compared cephalometrically and there was no statistically significant difference for all variables except for the anterior cranial base length (S-N), in which the male patients had larger values than the female patients ($p = 0.015$). The results are tabulated in Table 6.2.

Table 6.2: Comparison between sexes in cephalometric evaluation of patients in sample. Results of Student t-tests.

Variable	Sex	N	Mean	SD	SE	DF	t	p
N-ANS	Male	6	60.08	4.80	1.8	7	0.81	0.45
	Female	5	57.50	5.85	2.6			
ANS-ME	Male	6	72.33	8.24	3.4	8	1.76	0.12
	Female	5	65.00	5.53	2.5			
N-ME	Male	6	132.8	12.0	4.9	8	1.37	0.21
	Female	5	122.8	12.0	5.4			
S-Go	Male	6	83.00	6.20	2.5	8	1.93	0.09
	Female	5	75.20	7.06	3.2			
S-Ar	Male	6	37.17	2.99	1.2	6	1.91	0.10
	Female	5	32.80	4.31	1.9			
Ar-Go	Male	6	52.08	7.59	3.1	8	1.54	0.16
	Female	5	46.10	5.25	2.3			
PFH-AFH	Male	6	62.67	3.15	1.3	8	0.88	0.41
	Female	5	61.26	2.15	0.96			
UFH-LFH	Male	6	83.60	7.27	3.0	8	-1.23	0.25
	Female	5	88.46	5.86	2.6			
NF angle	Male	6	122.5	10.8	4.4	8	-0.81	0.44
	Female	5	128.2	12.3	5.5			
NL angle	Male	6	96.2	24.7	10	8	0.28	0.79
	Female	5	92.4	21.5	9.6			
S-N	Male	6	75.17	2.29	0.94	6	3.40	0.015*
	Female	5	68.60	3.78	1.7			
Co-Gn	Male	6	132.2	10.2	4.2	5	1.50	0.19
	Female	5	118.2	18.7	8.3			
SNO	Male	6	54.33	4.08	1.7	6	-1.12	0.31
	Female	5	57.80	5.85	2.6			
PNS-A	Male	6	49.75	4.42	1.8	8	1.64	0.14
	Female	5	46.28	2.47	1.1			
N-A-Pog	Male	6	-9.4	12.3	5.0	8	-0.96	0.36
	Female	5	-1.8	13.7	6.1			
SE-PNS	Male	6	54.83	5.00	2.0	7	1.40	0.20
	Female	5	49.70	6.82	3.0			
Co-A	Male	6	88.83	4.54	1.9	6	1.87	0.11
	Female	5	82.20	6.76	3.0			
Md-Mx	Male	6	43.3	13.4	5.5	8	1.12	0.29
	Female	5	34.0	14.0	6.3			

* = Statistically significant difference at p < 0.05

6.3 Soft Tissue Comparison between the Sample Group in the Present Study with Ideal Values

Double determinations of the patients’ soft tissue cephalometric variables were averaged and then compared with ideal values (Holdaway, 1983a). Paired Student t-tests of the mean differences revealed four out of the eleven soft tissue variables were significantly different from the norm. The results are tabulated in Table 6.3.

Table 6.3: Results of soft tissue cephalometric study comparing sample mean against normal values using paired Student t-tests.

Variable	N	Mean	SD	SE	t	p
Facial Angle	11	94.18	5.48	1.65	0.93	0.083
Nose Prominence	11	13.795	3.037	0.916	-4.59	0.001**
Superior Sulcus Depth	11	3.545	1.048	0.316	1.73	0.11
Subnasale to H-Line	11	6.318	2.639	0.796	1.66	0.13
A-N Pog						
Norm=1	11	-3.55	6.96	2.10	-2.17	0.055
Upper Lip Thickness	11	15.955	2.706	0.816	2.40	0.038*
Upper Lip “Strain”						
Norm=1	11	2.614	1.514	0.456	3.54	0.0054*
H-Angle	11	9.09	7.29	2.20	-0.41	0.69
Lower Lip to H-Line	11	3.591	1.562	0.471	6.56	0.00005**
Inferior Sulcus to H-Line	11	3.591	1.231	0.371	1.59	0.14
Soft tissue Chin Thickness	11	14.477	2.378	0.717	0.67	0.52

* = Statistically significant difference at p <0.05
** = Statistically significant difference at p <0.01

- 1) Reduced nose prominence (p<0.01). Graph of data seen in Fig 6.10.
- 2) Increased upper lip thickness (p<0.05). Graph of data seen in Fig 6.11.
- 3) Increased upper lip “strain” (at normal set at 1mm for strain p<0.05). Graph of data seen in Fig 6.12.
- 4) Prominent lower lip to H-line (p<0.01). Graph of data seen in Fig 6.13.

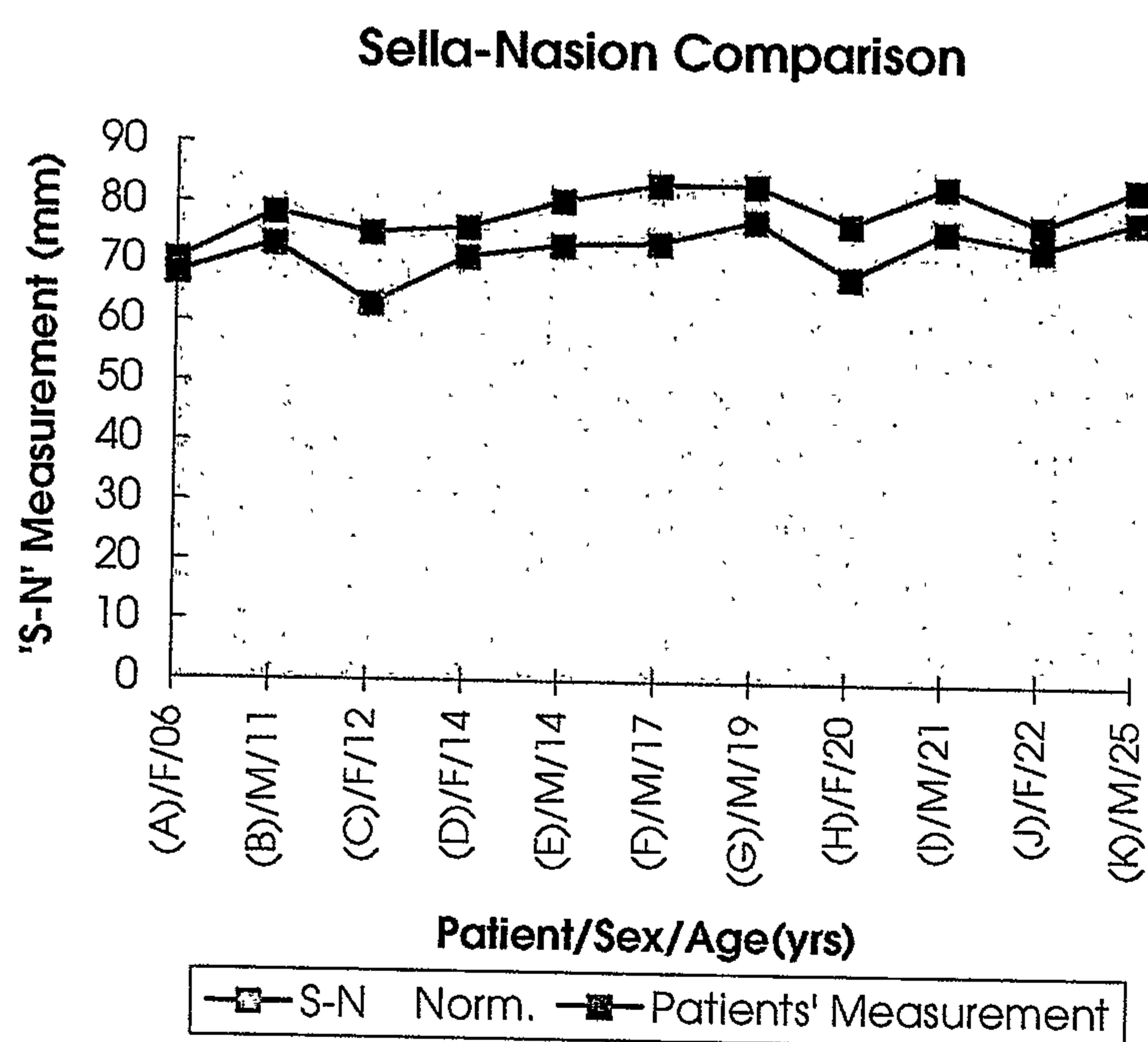


Fig. 6.1: Graph showing comparison between the patient sample and the norms for anterior cranial base length (S-N).

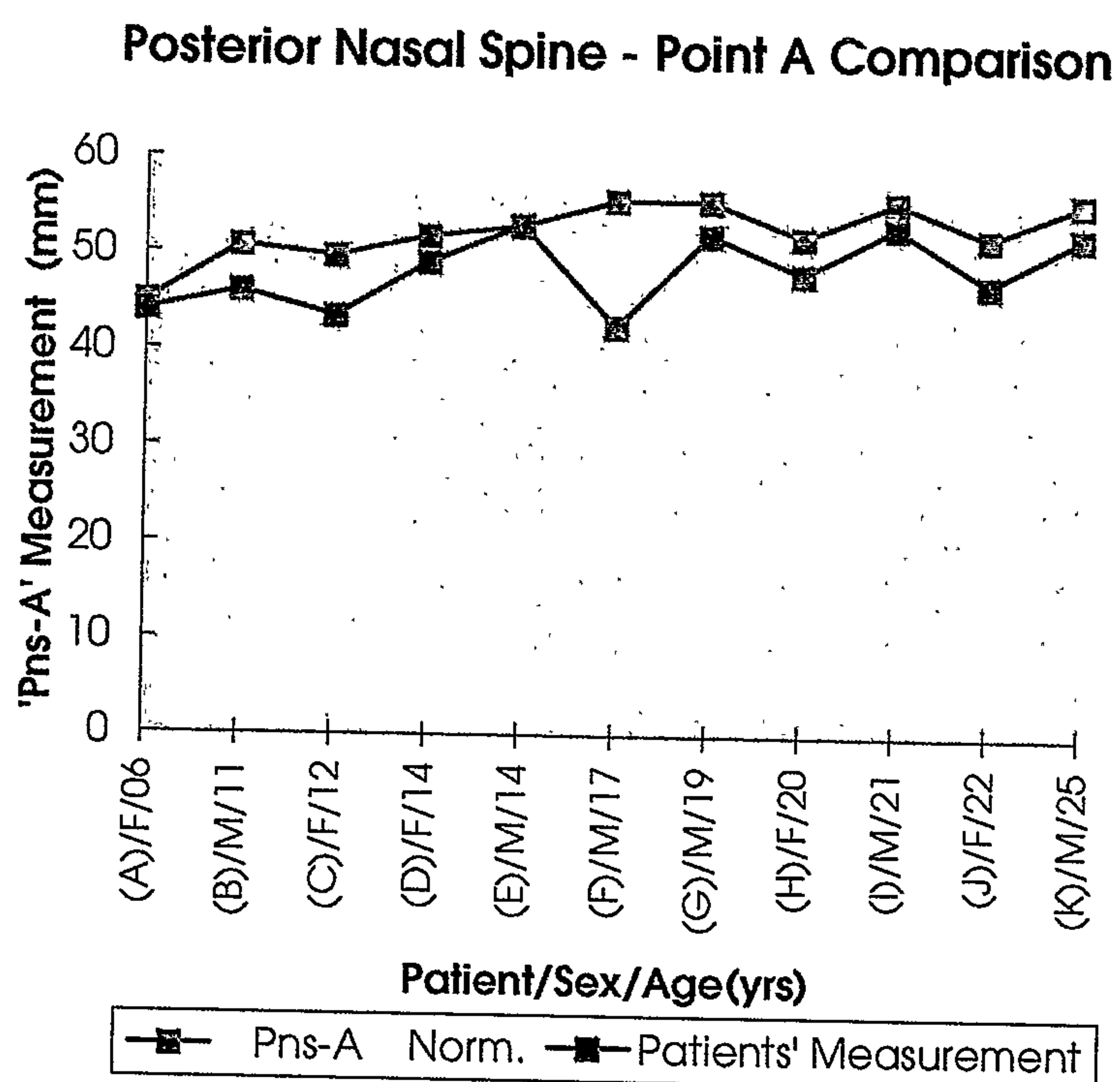


Fig. 6.2: Graph showing comparison between the patient sample and the norms for maxillary length (PNS-A).

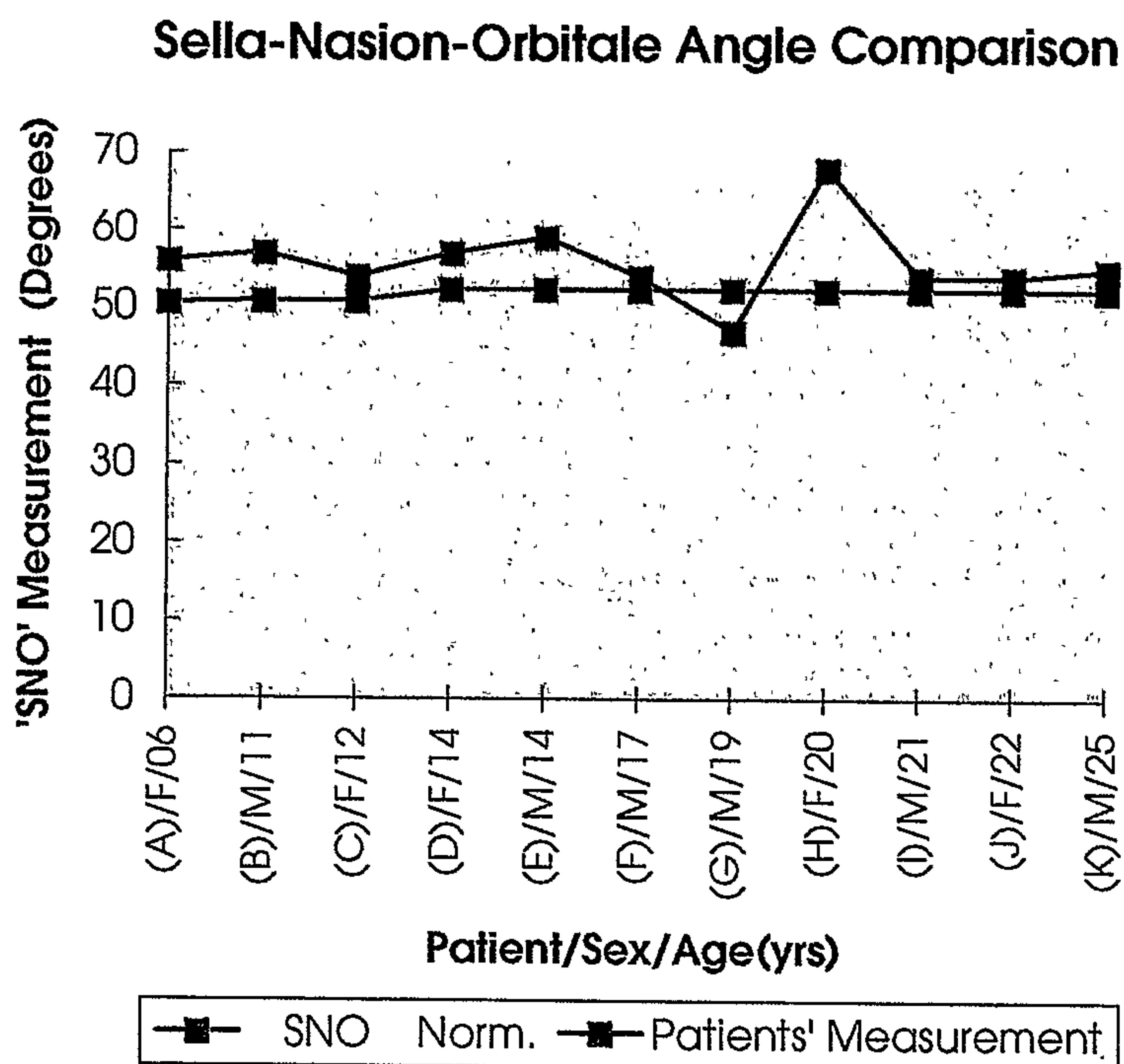


Fig. 6.3: Graph showing comparison between the patient sample and the norms for orbitale position relative to the anterior cranial base (angle SNO).

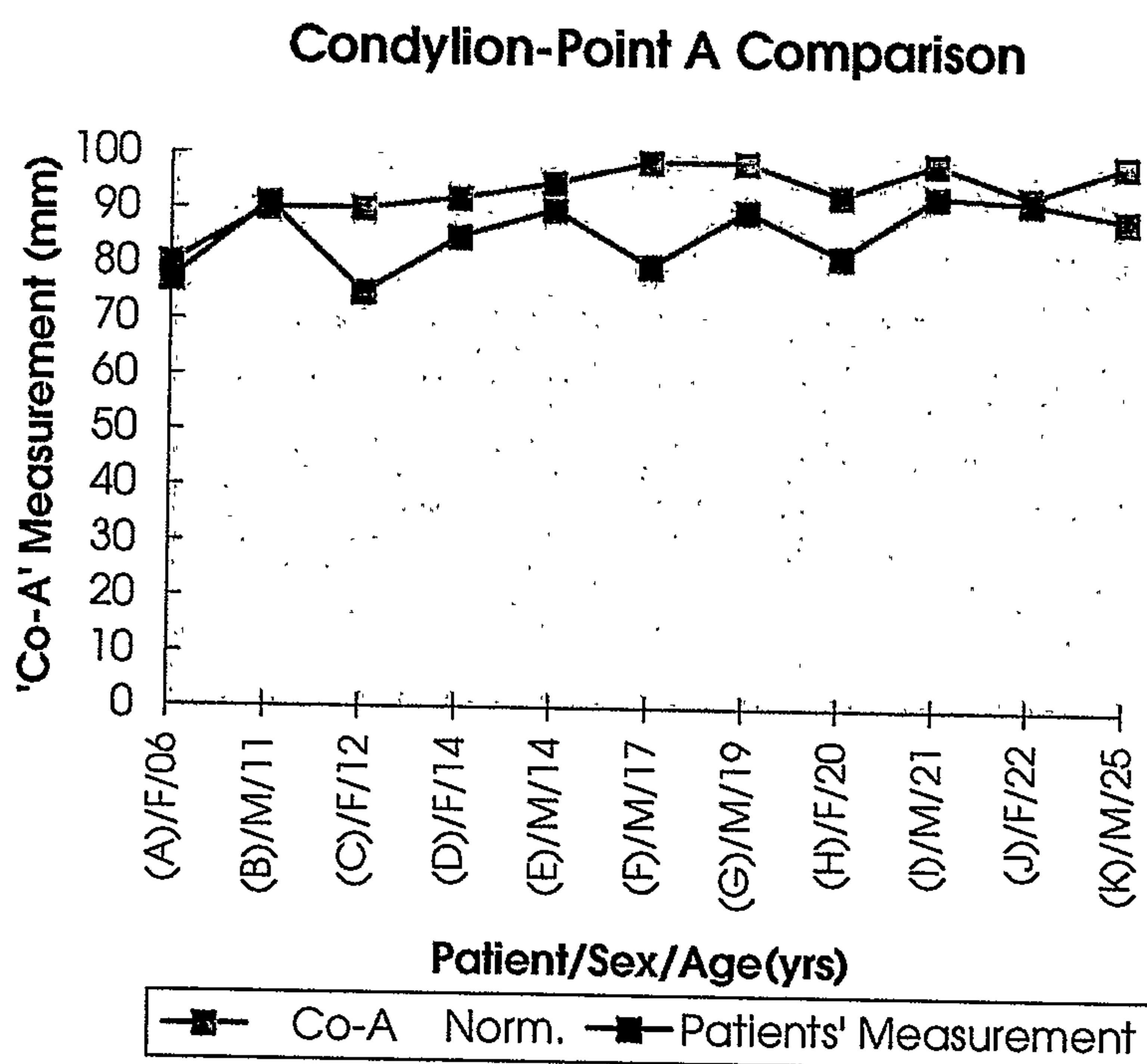


Fig. 6.4: Graph showing comparison between the patient sample and the norms for the effective maxillary length (Co-A).

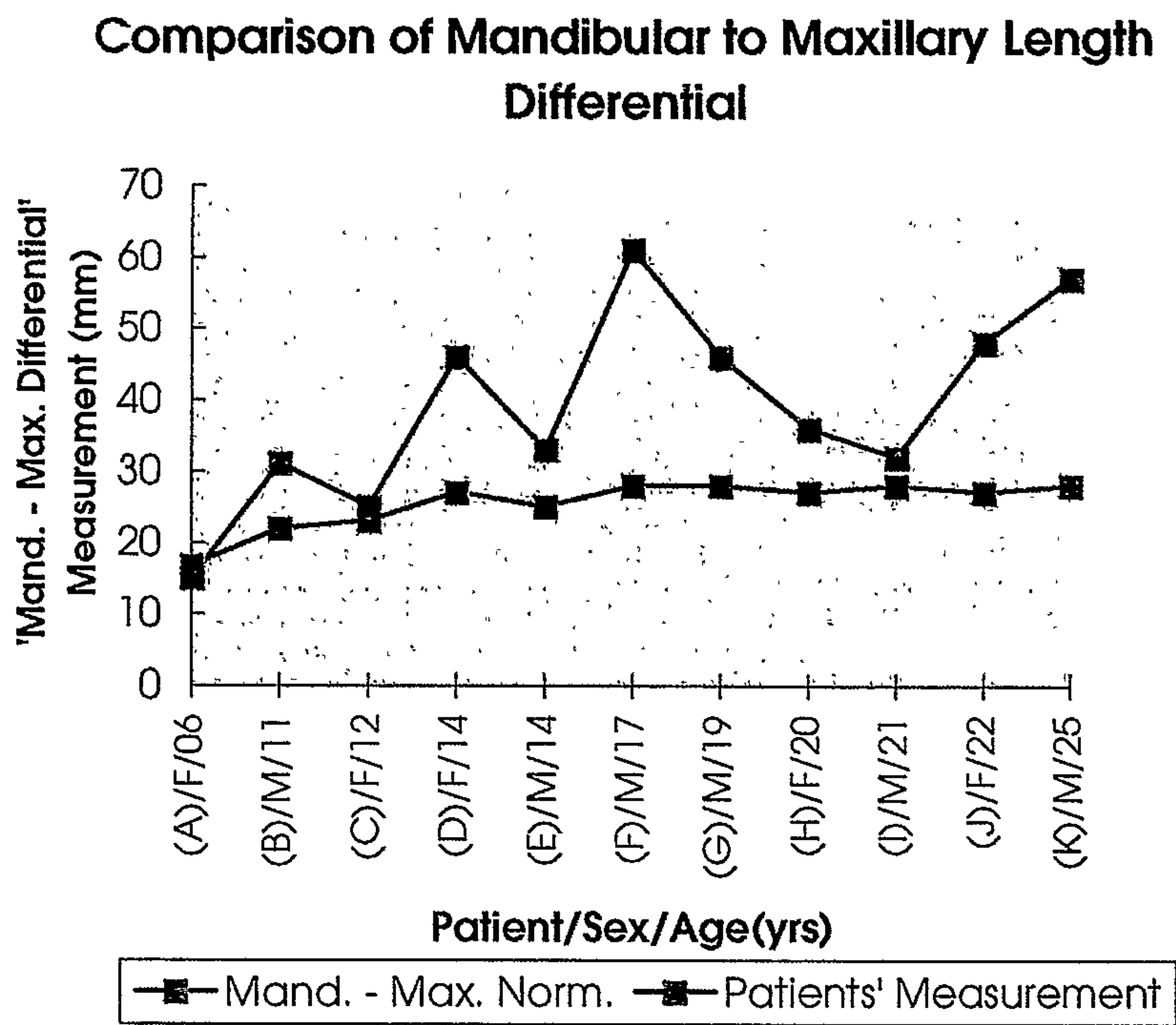


Fig. 6.5: Graph showing comparison between the patient sample and the norms for mandibular to maxillary differential.

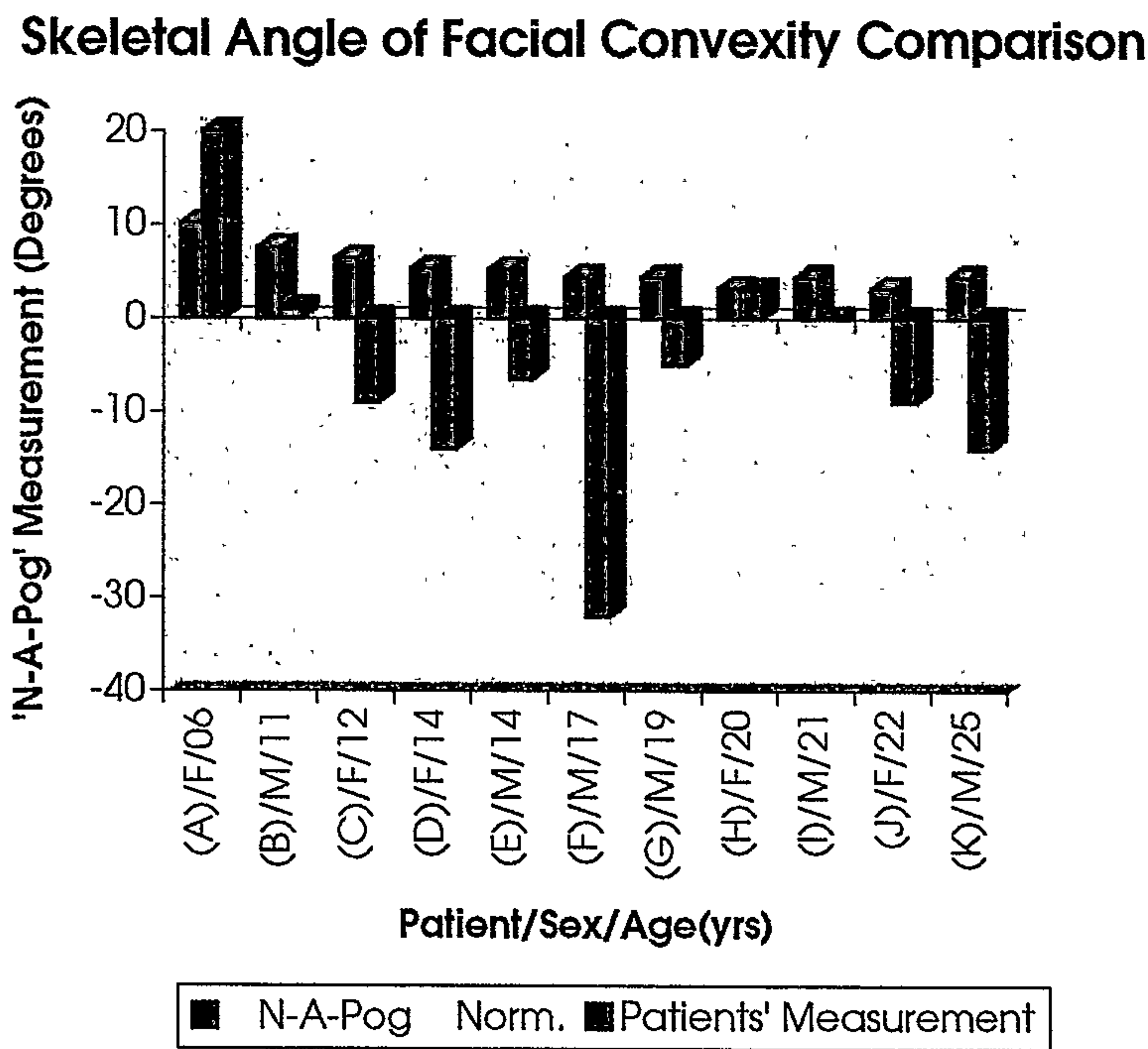


Fig. 6.6: Graph showing comparison between the patient sample and the norms for the skeletal angle of facial convexity (N-A-Pog).

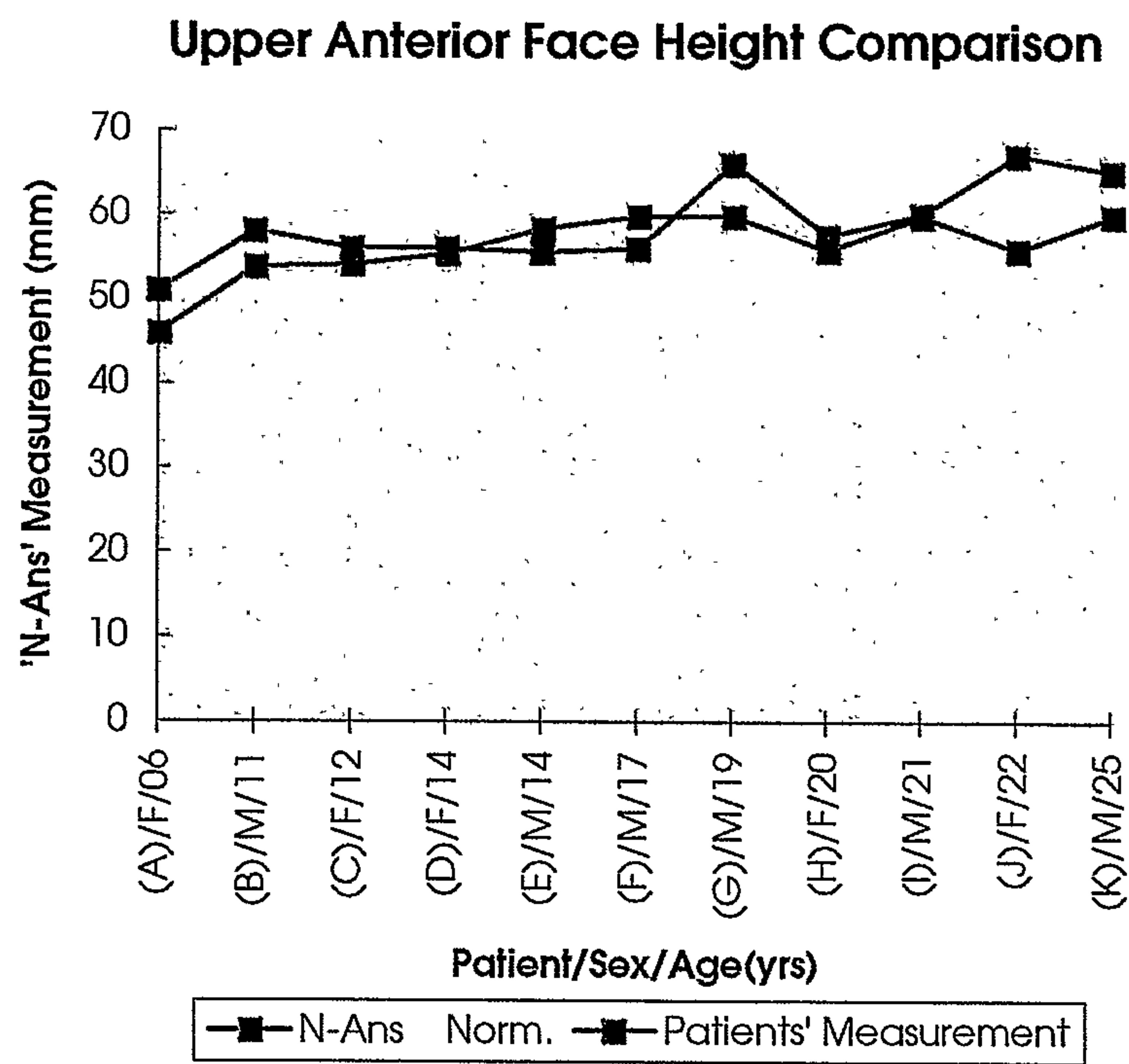


Fig. 6.7: Graph showing comparison between the patient sample and the norms for upper anterior face height (N-Ans).

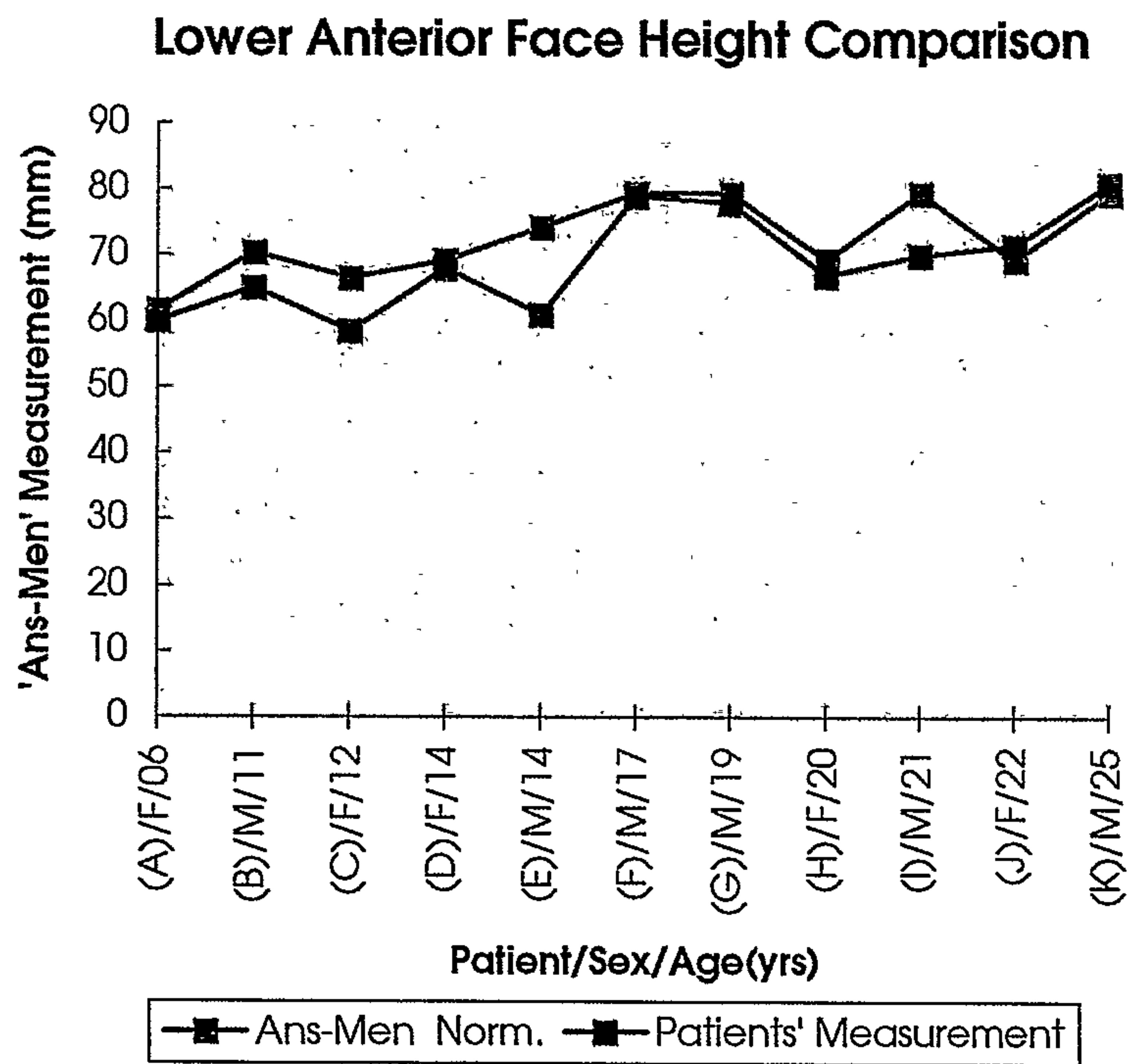


Fig. 6.8: Graph showing comparison between the patient sample and the norms for lower anterior face height (Ans-Men).

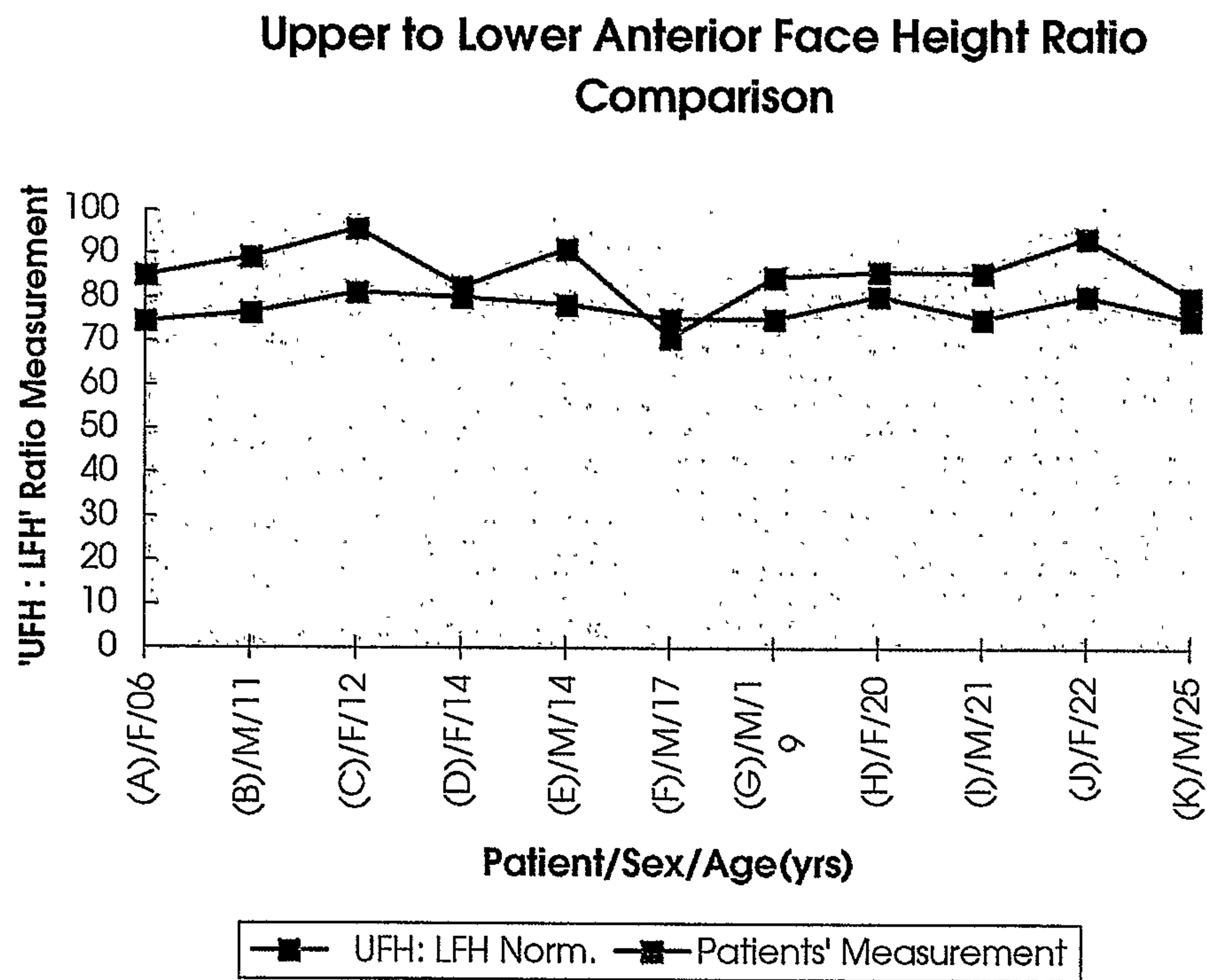


Fig. 6.9: Graph showing comparison between the patient sample and the norms for upper to lower anterior face height ratio.

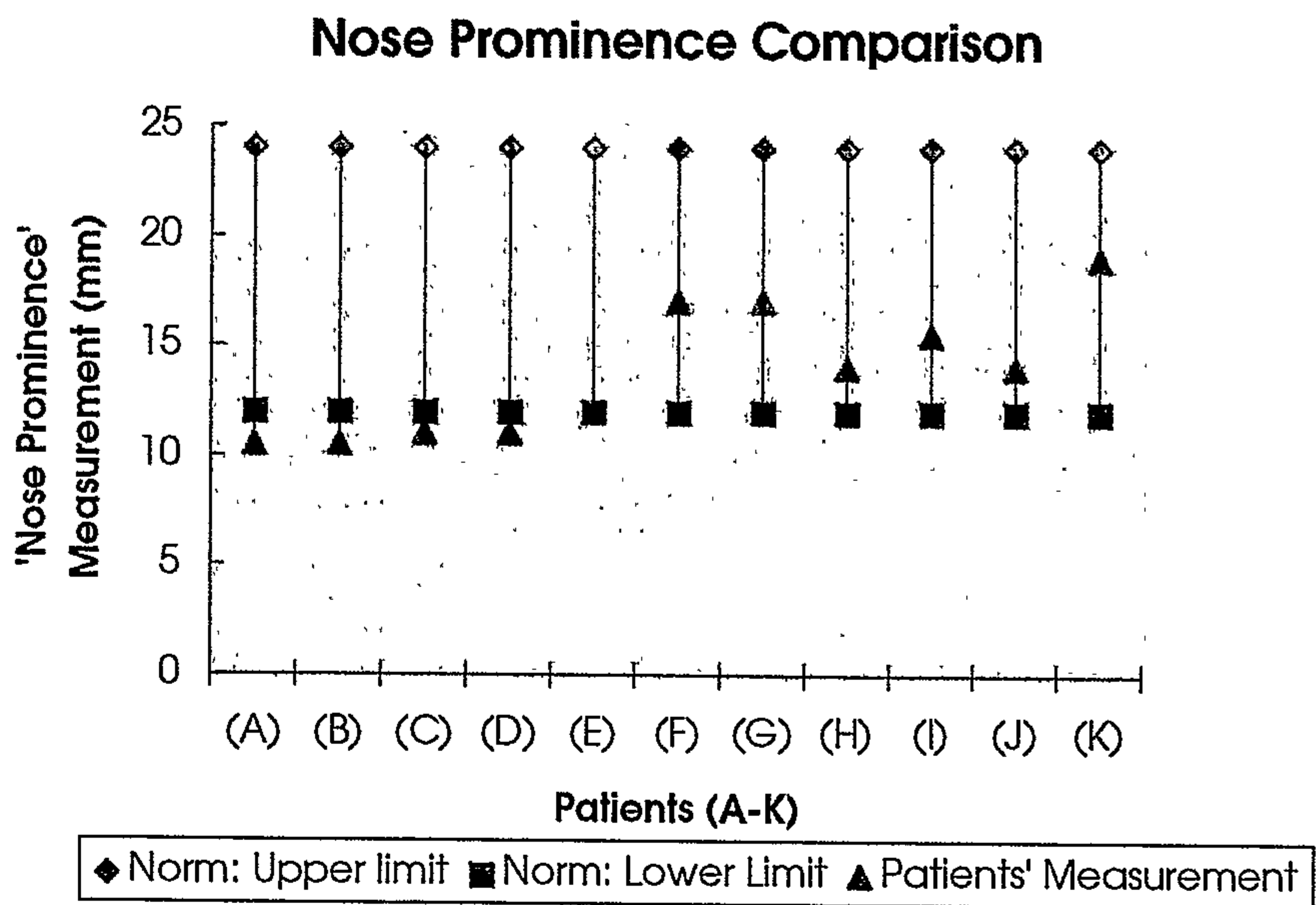


Fig. 6.10: Graph showing comparison between the patient sample and the norms for nose prominence.

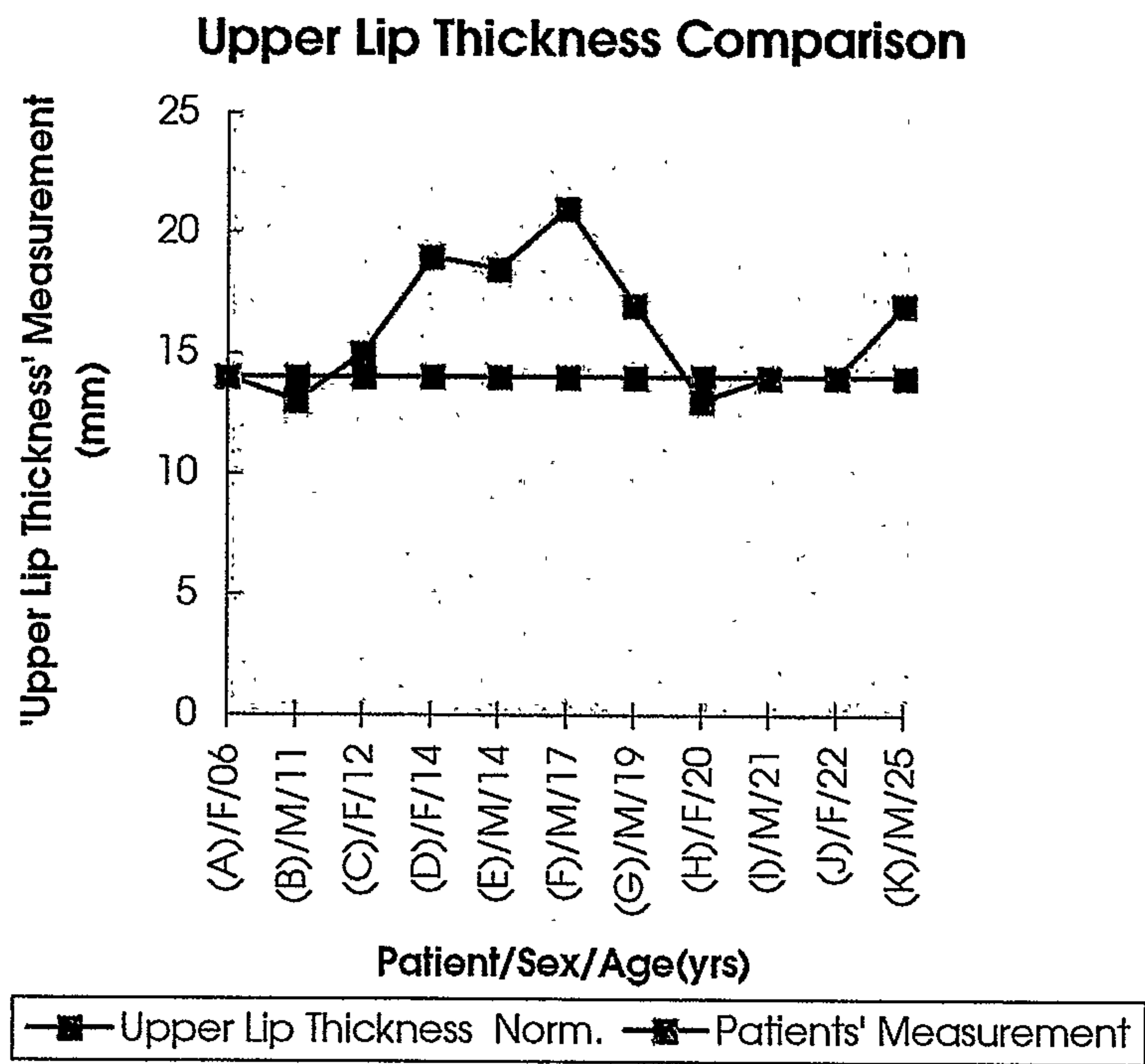


Fig. 6.11: Graph showing comparison between the patient sample and the norms for upper lip thickness.

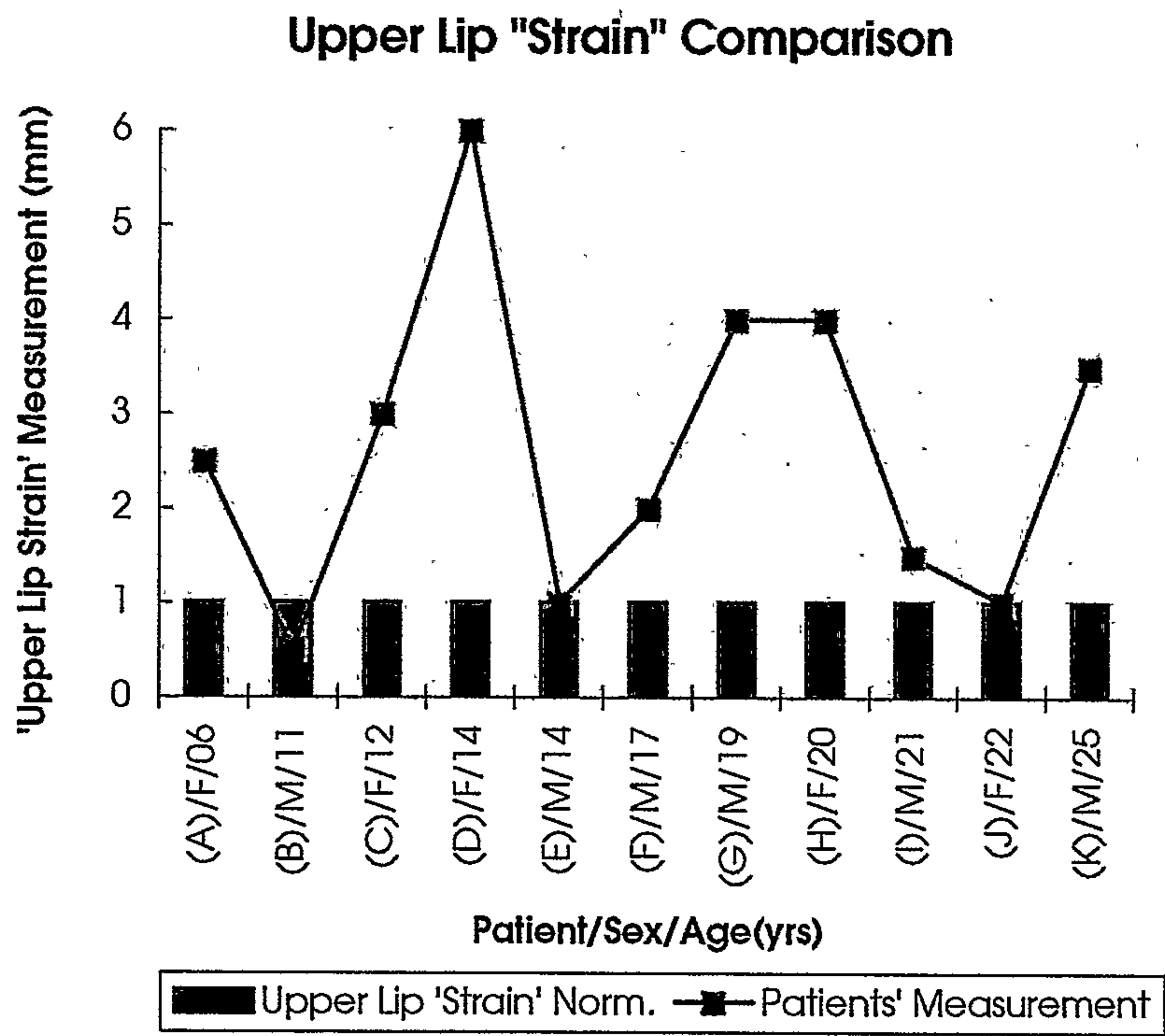


Fig. 6.12: Graph showing comparison between the patient sample and the norms for upper lip strain.

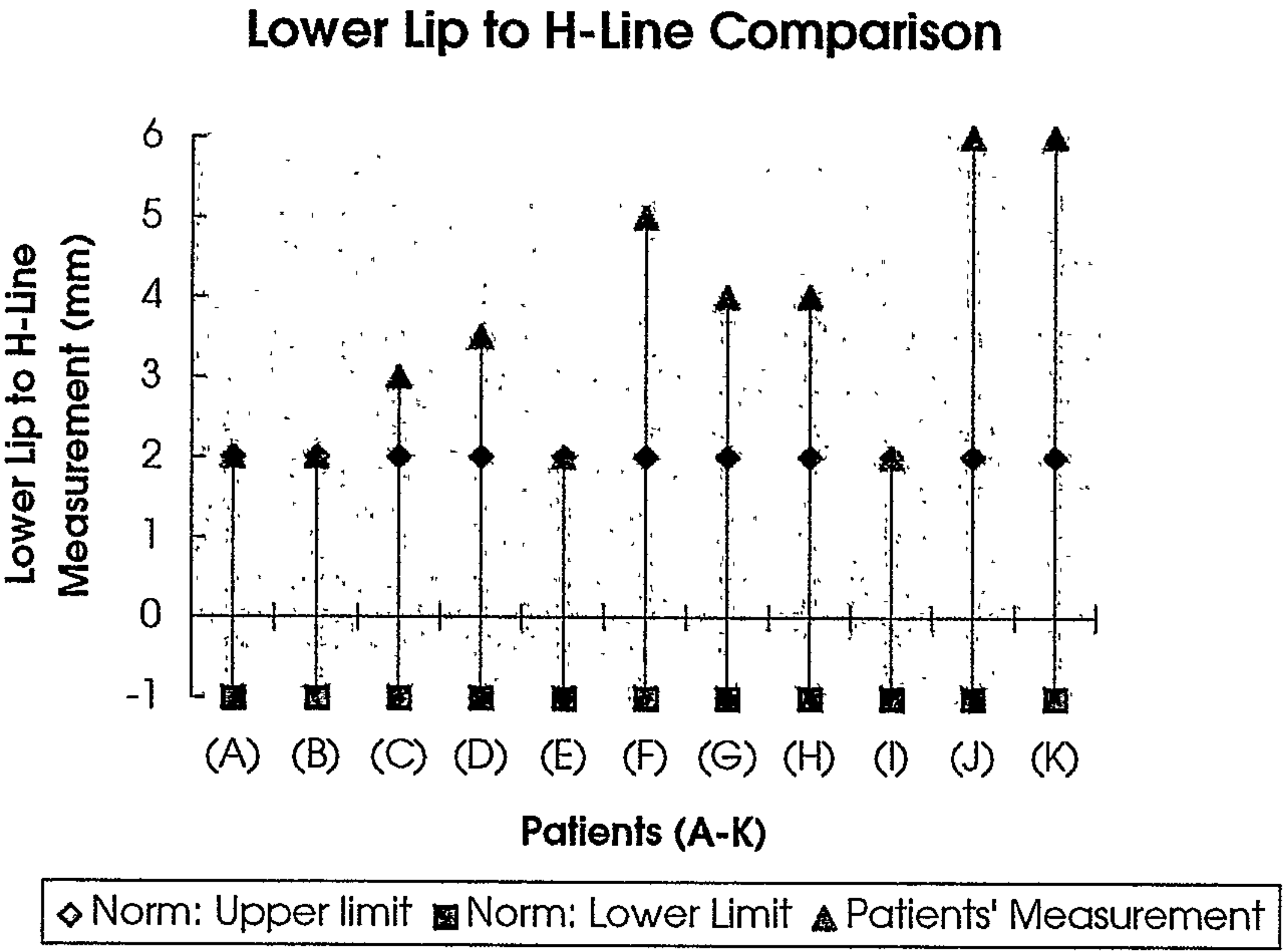


Fig. 6.13: Graph showing comparison between the patient sample and the norms for lower lip to H-line.

CHAPTER 7.

PART II: PHOTOGRAPHIC STUDY

7.1 Aims of the Present Study

This cross-sectional, retrospective study investigated facial profile characteristics of BS and CDP patients compared to their parents, using black and white life-sized profile photographs.

The objectives of this investigation were:

- 1) to investigate the presence of any measurable difference in facial profile between patients with a positive diagnosis for CDP or those diagnosed as having BS and healthy individuals matched for age and gender.
- 2) to investigate the presence of any measurable similarities of facial features between patients and their parents.

The following factors were used to evaluate the profile characteristics:

- 1) Family relationship - Patients, both BS and CDP, or parents
- 2) Gender - Male or female; mothers or fathers.

Thus, the four subgroups studied were as follows:

- 1) Mothers: Group One
- 2) Female patients : Group Two
- 3) Fathers : Group Three
- 4) Male patients : Group Four

7.2 Materials and Methods

Life size profile photographs of patients diagnosed with either CDP or BS (Tables 7.1 and 7.2), and who had attended either the Murdoch Institute of Birth Defects or the Royal Children's Hospital in Melbourne, were measured. There were altogether 80 life size photographs (40 frontal and 40 profile views). The 40 profile views were used for analysis, as these gave the most reliable measurements according to Farkas (1981). Of these 40 cases, there were 21 females and 19 males. Of the females, there were 11 mothers and 10 female patients. Of the males, there were six fathers and 13 male patients.

The photographs were sent to Sydney from the Murdoch Institute of Birth Defects in Melbourne. Initially, the only information provided with the photographs were the names, age and gender of each patient. Photographs of appropriate quality were randomised and coded prior to measurement, to avoid comparing cases with similar surnames. Measurements were carried out 'blind' without the knowledge of

the diagnosis to reduce bias. Once measurements were complete, details of each subject, including the diagnosis and family relationship, were obtained.

Table 7.1: Diagnoses of female patients in photographic study.

FEMALE PATIENT	AGE	DIAGNOSIS
A	38 YEARS 5 MONTHS	CDP (AS PER MALE "I" TWIN)
B	12 YEARS 3 MONTHS	CDP
C	11 YEARS 5 MONTHS	BS
D	21 YEARS 3 MONTHS	BS
E	6 YEARS	CDP
F	12 YEARS	CDP
G	12 YEARS 5 MONTHS	CDP
H	22 YEARS 7 MONTHS	CDP (s)*
I	11 YEARS 5 MONTHS	CDP
J	14 YEARS 2 MONTHS	BS

Table 7.2: Diagnoses of male patients in photographic study.

MALE PATIENT	AGE	DIAGNOSIS
A	8 YEARS 5 MONTHS	CDP
B	4 YEARS 1 MONTHS	CDP
C	9 YEARS 5 MONTHS	CDP (s)#
D	13 YEARS 4 MONTHS	CDP (s)#
E	8 YEARS	CDP
F	15 YEARS 3 MONTHS	BS
G	3 YEARS 6 MONTHS	SIBLING TO FEMALE G
H	18 YEARS 5 MONTHS	BS
I	ADULT	CDP (TWIN OF FEMALE "I")
J	16 YEARS 5 MONTHS	CDP + AMYOPLASIA
K	ADULT	BS
L	11 YEARS 2 MONTHS	CDP (s)#
M	15 YEARS	CDP (s)#

CDP (s)=Chondrodysplasia Punctata with 's' signifying a 'severe type with symmetrical lesions'

7.2.1 Standardised Facial Profile Photography

Many authors, (Burstone, 1958, Morrello, *et al.* 1977, Farkas, 1981, Larrabee *et al.* 1985 and Crawford, 1987) emphasised the necessity of standardised facial photography to achieve consistency of subject position and make valid comparisons of patients, especially for longitudinal studies.

Photographic profilometric analyses also require standardisation to focus on important structural characteristics of the aesthetically pleasing profile (Stoner, 1955; Neger, 1959; Peck and Peck, 1970; Larrabee *et al.* 1985; and Yuen and Hiranaka, 1989).

Farkas (1981) compiled extensive direct anthropometric measurements for an atlas of normal values. Social opinion of aesthetics vary markedly with classical 'canons' of beauty and it was, therefore, necessary to obtain measurements from normal, healthy individuals from a general population so that a range of values were obtained rather than a single, absolute "ideal".

7.2.2 Selection of Subjects for Photographs

The criteria in selecting the sample for this study were:

- 1) Patients at the Murdoch Institute of Birth Defects or the Royal Children's Hospital in Melbourne who had been diagnosed with either CDP or BS.
- 2) Parents of the aforementioned patients.
- 3) Photographs used in the study have patient consent and were taken from ongoing studies of BS and CDP (Horswell *et al.* 1987).
- 4) The patients had not undergone orthodontic treatment, orthognathic surgery or rhinoplasty prior to the photographs.
- 5) The photographs had the subject's head in natural head position, were life-sized, and were of appropriate quality to enable landmark identification.

7.2.3 Method Used to Standardise Facial Photography

The photographs were taken over a period of three to four years, approximately between 1987 and 1990. Although there was a protocol for the taking of these records, it cannot be confirmed that the Melbourne researchers strictly adhered to it, nor that the records were of the highest level of standardisation that would enable definitive data collection. For this reason, the present study sample size was reduced, and to reduce the degree of error, the analysis was limited to proportions or ratios rather than linear data.

To minimise variability in skin tone and colour, black and white photographs were taken. The following procedures were used in the taking of photographs for the present study:

- 1) Head position was standardised using the Natural Head Position (NHP) technique. This technique allows reproducible head position results, without the need of a head-holding apparatus, which would interfere with the visualisation of the face (Larrabee *et al.* 1985),
- 2) Subjects were instructed to stand at ease, relax their jaw, and look into the distance,
- 3) Subjects had their face levelled with the plane of the camera lens and the focal point was at the alar base (Horswell *et al.* 1988),
- 4) Lip posture was standardised by asking subjects to relax their lips, allow them to lightly touch together with minimal strain,
- 5) A clearly marked ruler with a white background was either fixed above the heads of the subjects or a measuring tape was affixed to the forehead. These were used to obtain the best approximation to 1:1 lifesize photographic prints.

7.2.4 Selection of Soft Tissue Reference Points

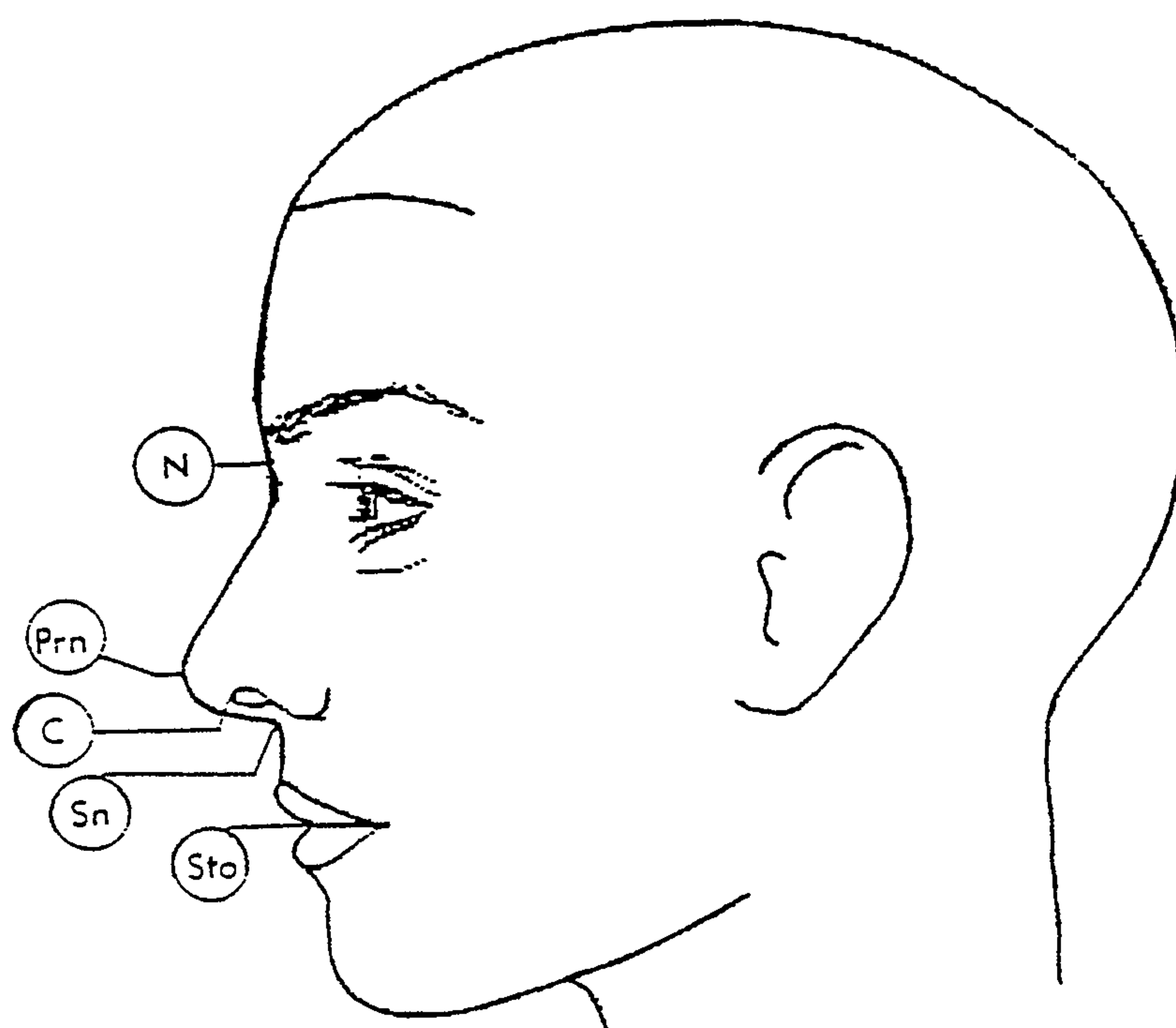


Fig. 7.1: Diagram of soft tissue points measured on photographs.
(Adapted from Farkas, 1981)

According to the method proposed by Farkas (1981), the following landmarks are reliable measures for photogrammetry: (Fig. 7.1)

- 1) **N** nasion
- 2) **Sn** subnasale
- 3) **Sto** stomion
- 4) **Prn** pronasale
- 5) **C** columellar crest

7.2.5 Definitions of Soft Tissue Distances and Landmarks:

- a. *N - Sto* (nasion to stomion): the length of the nose and upper lip.

Soft -tissue nasion (*N*): is the point in the midline of both the nasal root and the nasofrontal suture. In the present study, which is indirect anthropometry, the deepest point on the curvature of the root of the soft tissue nose above the level of the inner canthus of the eye was used (Farkas, 1981).

- b. **Stomion (*Sto*):** is the point at the crossing of the vertical facial midline, as defined below, and the horizontal labial fissure between gently closed lips with the teeth in intercuspal position (Farkas, 1981).

- c. *N - Sn* (nasion to subnasale): the length of the vertical nose.

Subnasale (*Sn*): is the midpoint of the columella base at the apex of the angle where the lower border of the nasal septum and the surface of the upper lip meet.

- d. *Sn - Sto* (subnasale to stomion): the length of the upper lip.

- e. *Sn - Prn* (subnasale to pronasale): the length of nasal projection.

Pronasale (*Prn*): the most protruded point of the apex nasi. This point is particularly difficult to determine if the nasal tip is flat (Farkas, 1981).

In this sample of BS and CDP patients, the problem of landmark location due to the flattening of the nasal tip was more pronounced.

- f. *Sn - C* (subnasale to columellar crest): the length of the columella.

Highest point of the columella (*C*): is the point on each columella crest, level with the top of the corresponding nostril (Farkas, 1981).

7.2.6 Method Used to Calculate Ratios from the Profile Measurements

Due to the lack of standardisation of the available data from Melbourne, the linear measurements were converted into ratios to reduce the degree of error that would otherwise be introduced. The proportionate photographic analyses used in this study will assess how the subjects varied from the statistical means as derived from anthropometric data reported by Farkas (1984).

The general formula is:

Ratio 1 = N-Sn : N-Sto

Calculated as: N - Sn (nasion to subnasale): the length of the vertical nose.

N - Sto (nasion to stomion): the length of the nose and upper lip.

Ratio 2 = Sn-Sto : N-Sn

Calculated as: Sn - Sto (subnasale to stomion): the length of the upper lip.

N - Sn (nasion to subnasale): the length of the vertical nose.

Ratio 3 = Sn-C : Sn-Prn

Calculated as: Sn - C (subnasale to columellar crest): the length of the columella.

Sn - Prn (subnasale to pronasale): the length of nasal projection.

Guidelines to the interpretation of these ratios were given by Farkas (1987) and Farkas and Munro (1987).

7.2.7 The Normative Anthropometric Data Used

The ratio norms derived are from anthropometric data published by Farkas (1981) who measured 1312 healthy, normal Canadian Caucasian subjects from 1973 to 1976.

Farkas established 26 groups, each comprising 50 subjects of one sex at a specific age from six to 18 years, containing children from British, Latin, French, Germanic, Slavic and other Caucasian origins. In each group, Farkas aimed to have 20 children of British origin, 16 of Latin origin (10 French and six other Latin), six of Germanic origin, four of Slavic origin, and four "other Caucasians".

However, the majority of fourth or earlier generation subjects was British (51.9 percent).

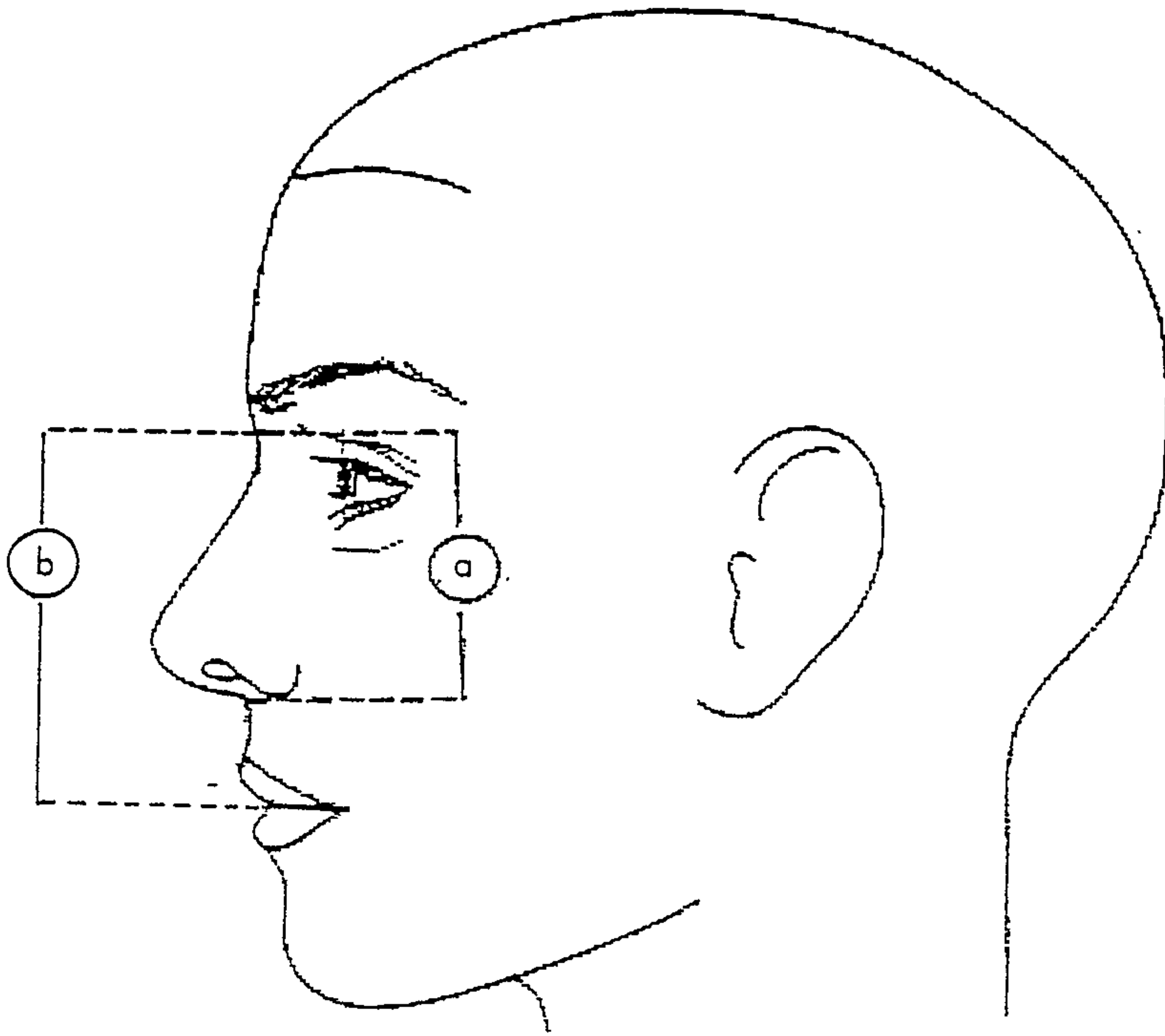


Fig. 7.2: Diagram of the profile measurements that comprise ratio 1.

Ratio 1 (N-Sn : N-Sto)

a) N-Sn (Nasion to subnasale)

b) N-Sto (Nasion to stomion)

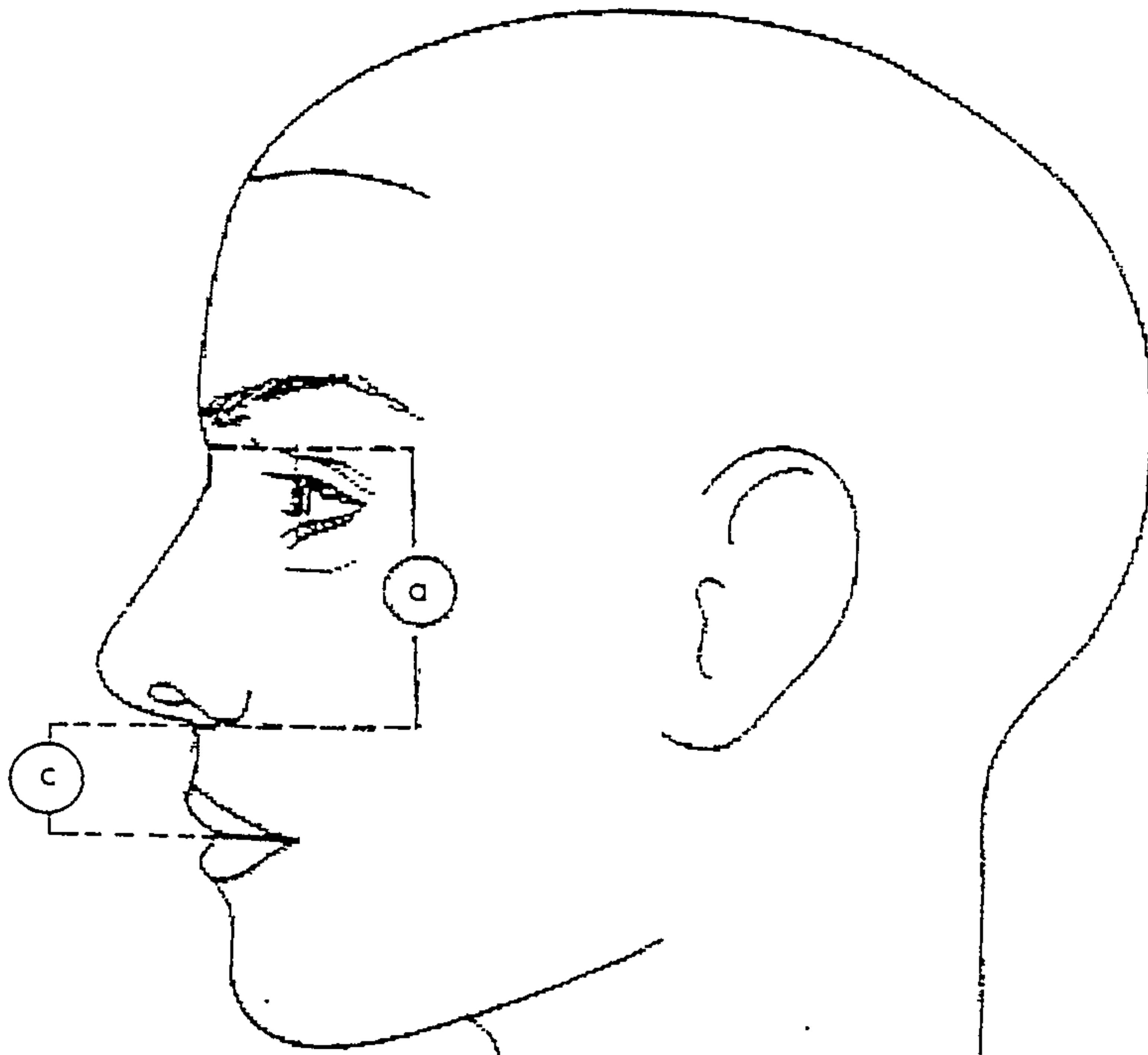


Fig. 7.3: Diagram of the profile measurements that comprise ratio 2.

Ratio 2 (Sn-Sto : N-Sn)

a) N - Sn (nasion to subnasale)

c) Sn - Sto (subnasale to stomion)

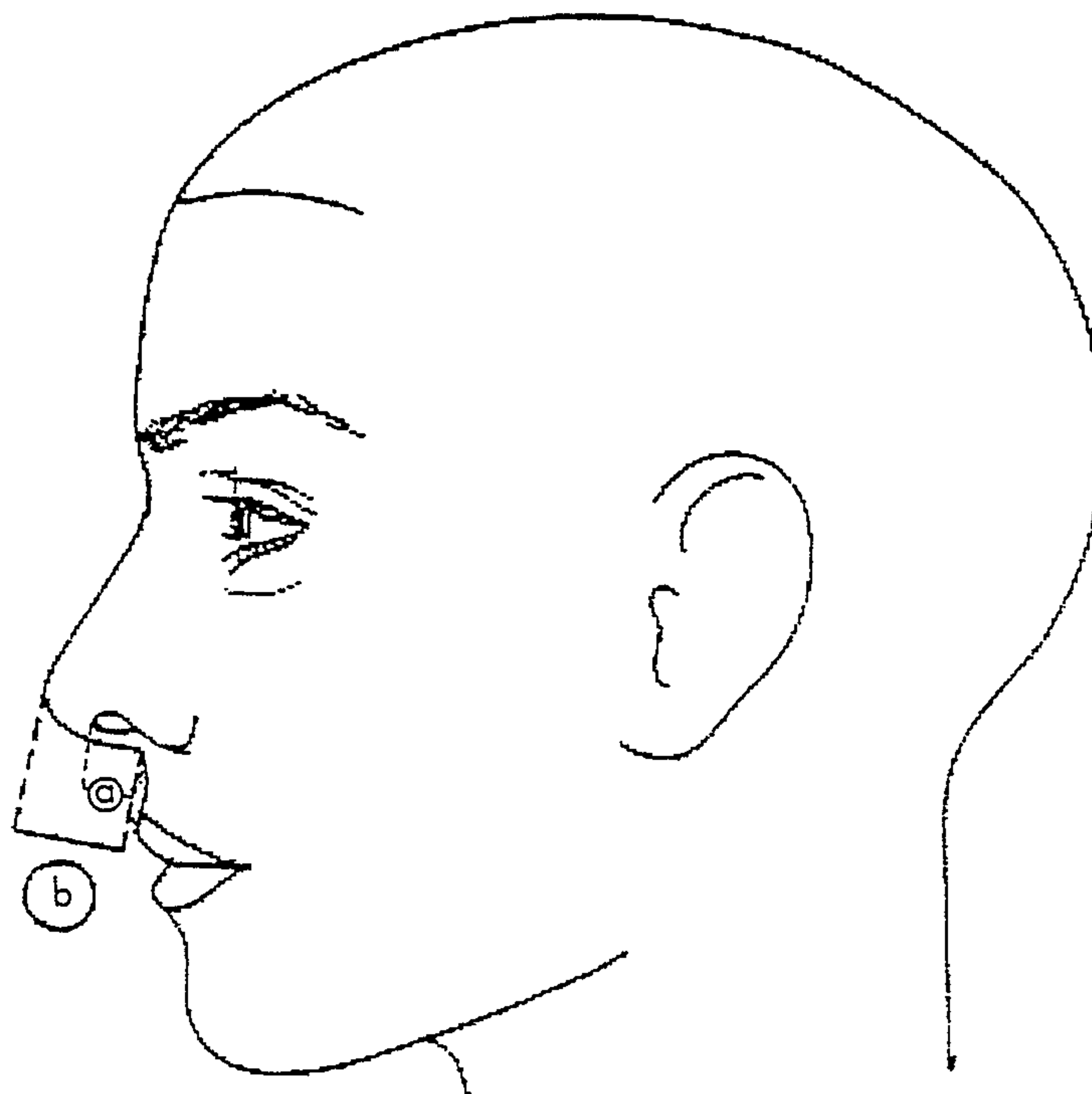


Fig. 7.4: Diagram of the profile measurements that comprise ratio 3.

Ratio 3 (Sn-C : Sn-Prn)

- a) Sn - C (subnasale to columellar crest):
- b) Sn - Prn (subnasale to pronasale)

7.2.8 Method Used for Measurement

The black and white lifesize photographs were judged for correct positioning of the head, the face leveled to the plane of the lens and the focal point at the ala base (Fig. 5.5 and 5.6). Many photographs did not meet these criteria and had to be excluded.

Several important drawbacks exist in the analysis of these photographs:

- 1) Only a number of photos had a centimetre ruler affixed to the forehead or held above the head by the patient on the frontal and lateral views, to aid in calculating enlargement.
- 2) The quality of the photographs varied, as some had distracting backgrounds, inconsistent lighting or were incorrectly exposed, which complicated landmark location.
- 3) Not all subjects were positioned in the standardised way, and no weighted plumb line was used to assess the gravity-determined vertical plane.
- 4) Detailed information, such as the type of camera, lens settings, or whether there was a fixed subject to camera distance was unavailable.

Because these patients represent a relatively small group with still much to be studied regarding the aetiology of their condition, these records were of great value

in the preliminary stages, and thus considered worthy for providing preliminary observations.

The proportionate photographic analysis used in this study assessed the subject variation from the statistical means as derived from anthropometric data (Farkas, 1981, 1984).

In this sample of 21 females, comprising of 11 mothers and 10 female patients, and 19 males, comprising of six fathers and 13 male patients ratios of five soft tissue variables were measured photogrammetrically as described.

All measurements were carried out using randomised original life-size profile photographs with no identification other than a letter of the alphabet, in order to reduce bias. All the photographs were traced with a 0.3 mm 3B mechanical pencil onto 0.003" matte acetate orthodontic tracing paper. All tracings within a particular study were completed within the same day, so that systematic errors were minimised (Houston 1983). All measurements were double checked against the photograph, and were recorded to the nearest millimetre.

After measurements were completed, the sample in the present photographic study was allocated into four main groups according to gender and whether they belonged to the "patient" or "parent" groups, and then re-labelled alphabetically: The ratio measurements were plotted on individual graphs, the data were analysed. The results of the photographic study appear in chapter 8.

7.2.9 Reliability of Measurements

An error study was carried out on the photographs for all the soft tissue measurements, which were re-measured on a separate occasion by the author. Paired t-tests were performed on double determinations for each subgroup of the sample. This was carried out for the five linear measurements used to create the profile ratios in the photographic study. The results, tabulated below in Table 7.3, showed that these measurements were reliable except for N-Sto and Sn-Sto.

Table 7.3 Results of double determinations on 40 photographs for 5 soft tissue variables.

Variable	Sub Group	N	Mean	St Dev	SE Mean	t	P-Value
N-Sto	Female pts	10	-0.500	0.624	0.197	-2.54	0.032*
	Mothers	11	-0.023	0.932	0.281	-0.97	0.35
	Male pts.	13	-0.385	0.618	0.171	-2.25	0.044*
	Fathers	6	-0.333	0.753	0.307	-1.08	0.33
N-Sn	Female pts	10	-0.200	0.422	0.133	-1.50	0.17
	Mothers	11	-0.045	0.350	0.106	-0.43	0.68
	Male pts.	13	-0.154	0.658	0.182	-0.84	0.42
	Fathers	6	-0.167	0.516	0.211	-0.79	0.47
Sn-Sto	Female pts	10	-0.100	0.394	0.125	-0.80	0.44
	Mothers	11	0.136	0.234	0.070	1.94	0.082
	Male pts.	13	-0.346	0.516	0.143	-2.42	0.032*
	Fathers	6	-0.167	0.516	0.211	-0.79	0.47
Sn-Prn	Female pts	10	0.050	0.284	0.090	0.56	0.59
	Mothers	11	0.045	0.270	0.081	0.56	0.59
	Male pts.	13	0.000	0.408	0.113	0.00	1.00
	Fathers	6	0.083	0.376	0.154	0.54	0.61
Sn-C	Female pts	10	-0.100	0.316	0.100	-1.00	0.34
	Mothers	11	0.136	0.234	0.070	1.94	0.082
	Male pts.	13	0.031	0.250	0.069	0.44	0.66
	Fathers	6	0.000	0.316	0.129	0.00	1.00

* = Statistically significant difference at p < 0.05



Fig. 7.5: An example of one of the photographs used: frontal view.



Fig. 7.6: An example of one of the photographs measured: profile view.

CHAPTER 8.

FINDINGS OF THE PHOTOGRAPHIC STUDY

8.1 Comparison of the Study Sample Groups with Normal Ratios Matched for Age and Sex

8.1.1 Group One: Mothers

There were 11 individuals in this group.

Paired t-tests were performed in order to compare the mothers with ratio norms matched for age and sex. For all three ratios, the mean values for mothers did not significantly differ from the mean normal values. However, two individuals in the parent group had a clinically noticeable appearance of a shortened columella or reduced nasal protrusion, or a combination of both. The graphs of these mothers appear to show that ratio 3 (Sn-C : Sn-Prn) was reduced markedly (Mothers "H" and "K").

It is impossible to assume any heritable link in this finding, as there is inadequate information on other family members, especially that of the older generations. In addition, these findings may be affected by factors including photographic distortion or measurement error.

8.1.2 Group Two: Female Patients

There were 10 individuals in this group. Their ages ranged from six years to 38 years and five months, with a mean age of 15 years and nine months at the time the photographs were taken. The majority were younger than 14 years of age. Six patients in this group were diagnosed with CDP (60 percent), the rest were diagnosed with BS.

When graphs of ratio measurements were examined, a definite trend emerged. In ratio 1 and 2 the females did not differ greatly from the norms (except Female "E", a six year old with CDP, who had a high ratio 2 reading). However, in ratio 3 (Sn-C : Sn-Prn) which is the ratio of the length of the columella to the degree of nasal tip protrusion, many of the female patient group stayed below the normal range :

- Female "B"-12 years 3 mths, diagnosed with CDP
- Female "G"-12 years 5 mths, diagnosed with CDP- sibling of Male "G"
- Female "H"-22 years 7 mths, diagnosed with CDP(s)
- Female "T"-11 years 5 mths, diagnosed CDP

Paired t-tests were carried out to compare the female patients with ratio norms matched for age and sex. For ratio 1 and 2, the female patients did not differ significantly from the norm.

However, for ratio 3, there was a statistically significant difference from the norm ($p < 0.0005$). For four individuals, ratio 3 was markedly reduced, in others this ratio was less affected. This finding appears to be borne out in the tremendous variation in the phenotype of CDP or BS with respect to the nasal deformity. As described by Holmström (1986), there are some cases where the length of the columella (Sn-C) is severely reduced and for others the columella is not as short, perhaps even near normal range, but the nasal tip protrusion (Sn-Prn) is flattened and the ala base is retruded.

8.1.3 Group Three: Fathers

There were six individuals in this sample. The ratios were plotted on graphs as in the previous groups, and compared to the norms from Farkas (1981). For all three ratios, the fathers followed the normal ratio values.

Paired t-tests were conducted on the sample to compare how they varied as a group from normal values. For all three ratios, there was no statistically significant difference from the norm. Speculation may be made, that the fathers represented a normal group, and did not show phenotypic traits of BS or CDP. The casual observer would find that the fathers' photographs appeared normal.

8.1.4 Group Four: Male Patients

There were 13 patients in this sample, which represents the largest number of all the four groups. Ten of these patients represented those diagnosed with CDP (76.9 percent) and three were diagnosed with BS (30 percent). The ages ranged from three years six months to 18 years five months, with a mean age of 12 years at the time the photographs were taken.

The graphs of this sample group showed a trend similar to that of the female patients, but ratios 2 and 3 were affected, whilst ratio 1 remained within normal limits.

Ratio 2 was reduced in the following:

- Male "E"- 8 years old with CDP, has a 12 year 3 month old sister - Female "B" also with CDP
- Male "F"- 15 years, three months old with BS
- Male "I"- CDP, adult twin of Female A who has CDP

Ratio 3 was reduced in the following:

- Male "C"- 9 years 5 months, with CD (s)
- Male "F"- 15 years 3 months, with BS
- Male "G"- 3 years 6 months, sibling of sister who was Female "G" (CDP)
- Male "I"- CDP, adult twin of Female A who has CDP
- Male "J"- 16 years 5 months, diagnosed with CDP and Amyoplasia
- Male "K"- adult with BS
- Male "L"-11 years, 2 months with CD (s)
- Male "M"-15 years, with CD (s)

Paired t-tests were performed to evaluate and compare normative versus patient data. Results showed that for ratio 1 the male patients were within the normal range.

However, for ratio 2, which is Sn-Sto : N-Sn, or the ratio of upper lip length to the length of the nose, the results approached significance ($p=0.057$).

For ratio 3, (Sn-C : Sn-Prn), which is the ratio of the length of the columella to the degree of nasal tip protrusion, the male patient group was significantly different to the norms ($p=0.0069$).

8.2. Comparison between Parents and Children in the Sample Groups for Each Ratio

The photographic study also evaluated the presence of measurable similarity of the facial profile between 21 pairs of patients and their parents for these three ratios. Pearson's Correlation Coefficients showed no correlation between parents and patients for any of the three ratio measurements evaluated: ratio 1 ($r= -0.260$); ratio 2 ($r= -0.161$); ratio 3 ($r= 0.178$).

8.3 Comparison between Individual Siblings

Due to the small number of observations, no statistical evaluation was carried out between the 4 pairs of siblings. However, graphs of ratio relationships between siblings showed no relationship emerging for any of the three profile ratios.

CHAPTER 9. DISCUSSION

9.1 Selection of Subjects

9.1.1 Subjects for the Cephalometric Study

The cephalometric study of 11 patients of Caucasian background comprising of five females and six males represented a cross-section of patients of various age groups with the majority diagnosed with CDP (Table 5.1). Only two patients of 11 were diagnosed with BS, who also had Phenytoin embryopathy. None of these patients had dental study-models taken or had accompanying photographs or radiographs of parents available.

9.1.2 Subjects for the Photographic Study

The subjects in the sample was of Caucasian background. Unlike the cephalometric study, there were relatives of the patients represented in the data for comparison. Of these 40 cases, there were 21 females and 19 males. Of the females, there were 11 mothers and 10 female patients. Of the males, there were six fathers and 13 male patients. A wide variety of profiles were demonstrated amongst the two parent groups. However, the patients had a marked similarity in the appearance of the nasomaxillary region that was characteristic of BS and CDP, whilst they varied in lower jaw morphology.

Five subjects were diagnosed with BS (two females and three males), 11 with CDP (six females and five males), and five with 'severe symmetrical CDP' or CDP (s) (one female and four males), (Tables 7.1 and 7.2). The terms 'CDP' and 'CDP (s)', with 's' signifying a 'severe type with symmetrical lesions' were an arbitrary classification, which is based on the presence of one or more sites of punctate calcification seen radiographically (Sheffield *et al.* 1976).

A comparison between each of the above subgroups according to diagnosis would have been of value, as the aetiology of BS is not clearcut and reflects the view held by some authors, that CDP and BS represent two ends of a broad spectrum of Chondrodysplasias. However, there were far less BS patients than those with CDP and CDP (s), making statistical analysis difficult. One reason for this may be the fact that the sample was referred to a paediatric craniofacial unit that recognised the features of CDP, and the patients presented initially when infants. This permits the observation of punctate calcifications that confirm the diagnosis of CDP.

9.2 Reliability of Measurements

Houston (1983) indicated that errors in measurement studies may be systematic or random. He believed that soft tissue studies are prone to random errors

due to the way the subject posed. These random errors are influenced by lip posture and facial muscular activities at the time of photograph or lateral headfilm taking. Whether the subjects were standardised by standing in 'Natural Head Position', with Frankfort Horizontal parallel to the floor, and the difficulty of some landmark identification may also have contributed to random errors. However, these are reduced if measurements are replicated and averaged. Houston maintained that apart from the measurements, tracings should also be replicated because the greatest errors may arise from point identification rather than in measurement. Systematic errors can be partly controlled by randomising the order in which the records are traced and measured to avoid bias, and by using a single observer.

Baumrind and Frantz (1971a, 1971b) found three sources of measurement errors:

- 1) Errors of projection
- 2) Errors of landmark location
- 3) Mechanical errors in drawing lines between points on tracings and in measuring with a ruler or protractor.

9.2.1 Errors of Projection

The X-ray source can cause distorted enlargements with head films, because the cephalogram is only a two dimensional shadow of a three dimensional object. Since the rays originate from a small source and are non-parallel, head films are always distorted enlargements, with the error varying with the plane at which the estimated point lies. Further distortions on headfilms result from the foreshortening of distances between points lying on different planes and by radial displacement of all points and structures not on the central ray. Baumrind and Frantz (1971b) stated that the introduction of correctional factors for the errors are cumbersome and impractical for clinical use and, therefore, not generally employed.

The photographic negatives were enlarged to live dimensions (1:1) and measured using a ruler scale. However, not all photographs were strictly standardised. The enlargement factor was difficult to calculate to exclude discrepancies, due to the variation in sharpness and the rulers not being in a fixed position on all photographs. However, angular measurements would not have been affected by enlargement, and should remain constant (Hautvast, 1971).

9.2.2 Errors of Landmark Location

Most references refer to cephalometric landmark location, which has limited relevance to measurement of photographs. Gravely and Benzies (1974) attributed the lack of clarity of cephalometric landmarks to superimposition of structures, blurring of the image caused by movement during exposure, lack of film contrast and emulsion grain. These problems were significant to photographs because the

midsagittal landmarks used were easily located on the external surface of the profile on the life-size print. However, the landmarks were not marked directly on the subject's skin prior to photography, as recommended by Farkas (1981), although absolute certainty of landmark location cannot be guaranteed even with this precaution.

Richardson (1966), Baumrind and Frantz (1971b) and Hillesund *et al.* (1978) found that some cephalometric landmarks can be located with greater reproducibility than others, with some more reproducible vertically than horizontally, and vice versa. These findings were considered when selecting the points, planes, or lines for this study. In the photographic study, for example, soft tissue nasion was used instead of soft tissue glabella to mark the upper limit of the face as it is more reproducible (Powell and Humphreys, 1984).

Burstone (1958), Wisth and Boe (1975) and Hillesund *et al.* (1978) reported that variation in facial expressions can make landmark location difficult. This finding highlights the importance of standardising facial expression and lip posture. However, because the present study is descriptive, reproducibility of the lip position is not as critical as in the use of serial photography in the evaluation of changes following treatment. A natural relaxed lip position with lips in light contact is preferred as it is a more accurate representation of soft tissue facial features.

Richardson (1966) found that intra-observer variability is less than inter-observer variability. A single observer could reproduce measurements with an acceptable degree of accuracy, provided all measurements were carried out using standardised measuring technique. Hence, in the present study, all tracing and landmark location were performed by the author to maximise measurement reproducibility and minimise systematic error.

9.2.3. Mechanical Errors in Drawing Lines between Points on Tracings and in Measuring with Ruler or Protractor

Gravely and Benzies (1974) found that measurement error, associated with thickness of pencil lines and perceptive limits of the human eye, contributed to tracing errors. Baumrind and Frantz (1971b) suggested that these errors can be eliminated by using machines to compute linear and angular relationships algebraically from landmark coordinates. Due to technical constraints in the present study, all measurements were carried out manually.

In addition, certain points are more difficult to reproduce accurately. Wisth and Boe (1975) and Hillesund *et al.* (1978) found that the vertical position of a point situated on a wide curvature difficult to define.

9.3 Data from the Cephalometric Study

9.3.1 Hard Tissue Comparison between the Sample Group in the Study and Normal Values Matched for Age and Sex

The patients in the sample were pooled, and individuals compared with normal values matched for age and gender and variation from the norms were calculated.

Effective mandibular length, Co-Gn, also remained within normal range, confirming that the relative prominence of the lower jaw and Class III dental relationship was due to a hypoplastic maxilla rather than mandibular prognathism in CDP and BS.

Orbital position relative to S-N, (SNO), was found to be mildly recessive, although this location is likely to be an under-estimation, as the anterior cranial base length (S-N) is smaller than average, and nasion would be further back due to a recessed nasomaxillary complex. This finding is in keeping with that of Horswell *et al.* (1988), who used SNO in 19 BS cases. He found that projection of a normally positioned nasion (normal S-N length), would reveal a recessed orbitale position with as much as 2 standard deviations below the norm for SNO.

9.3.2 Hard Tissue Comparison between the Sexes in the Sample Group

For all variables except for the anterior cranial base length (S-N), no statistically significant difference existed between male and female groups in the way they differed from norms matched for age and sex. Thus, the data suggest that BS and CDP affected the two groups in a similar fashion. The male patients had larger values than the female patients for the anterior cranial base length (S-N), ($p=0.015$).

9.3.3 Soft Tissue Comparison between the Sample Group in the Study with Ideal Values

Holdaway's (1986) soft tissue cephalometric analysis was used to compare the patients with BS or CDP with ideal values. In addition, the nasolabial and nasofrontal angles were used, to evaluate nasal prominence without solely relying on linear measurements.

Significant differences were found in the sample group for four of the thirteen soft tissue variables measured. The BS and CDP patients, on average, had a reduced nose prominence, an increased upper lip thickness, an increased upper lip "strain", and a prominent lower lip to H-Line measurement.

Summary

The anterior cranial base length was reduced in a high proportion of the patients in this study. This finding may reflect the true diminution in the anterior cranial fossa and subnormal ethmomaxillary horizontal growth. Alternately, the glabella and frontal sinus region may have remained proportionately underdeveloped throughout growth, resulting in a retruded nasion, as described by Horswell *et al.* (1988).

The mean maxillary vertical length (SE-PNS) was within the normal range. However, the mean maxillary horizontal length, as measured by the two variables (PNS-A) and effective maxillary length (Co-A), was less than normal in most cases for both variables in this sample. For both horizontal maxillary length determinations, there appeared to be a parallel relationship to the anterior cranial base dimensions.

This conclusion supports the contention of Munro *et al.* (1979) and Horswell *et al.* (1988) who studied BS, and concluded that a short maxilla was positioned on a short anterior cranial base in these subjects. According to Horswell *et al.* (1988), "... the interesting finding of an initially decreased maxillary height which 'catches up' in vertical growth and a horizontal maxilla which remains small brings into play the complex relationship of basicranium, ethmomaxillary complex and nasal septal apparatus during growth." However, it must be considered that Horswell *et al.* (1988) compared 19 BS patients in four broad age groups, with composite normative data in his retrospective cross-sectional material, and made statements about growth from it. Because he used cross-sectional data from a small sample and did not account for sexual dimorphism, Horswell *et al.* (1988), the growth analysis has severe limitations. However, Olow-Nordenram and Thilander (1987,1989) conducted investigations on the growth and development of children with BS in two longitudinal groups. One group consisted of 13 orthodontically untreated children (Olow-Nordenram and Thilander, 1987) and the other group consisted of 15 orthodontically treated patients at corresponding ages (Olow-Nordenram and Thilander, 1989). It was shown that all linear dimensions changed with age parallel with normal growth and approximately to the same extent, although on a reduced dimensional level for variables such as the anterior cranial base and maxillary lengths.

The finding of the present study, where the vertical height of the midface was unaffected whilst anteroposterior dimensions were reduced, is also supported by the midfacial growth literature on similar syndromes such as arhinencephaly (Latham and Burston, 1966). Thus, BS and CDP represent experiments of nature, where the premature and uncontrolled calcification of the foetal septal cartilage may restrain

the normal growth rate of the septum. This premature calcification of the septum reduces the forward progress of the midface that follows, and due to the reduced traction of the septopremaxillary ligament upon the anterior nasal spine to which it attaches, the anterior nasal spine may not develop fully.

Olow-Nordenram and Thilander (1989) also undertook a study to compare their two longitudinal samples to study the effects of orthodontics on Binder's cases. Unfortunately, no positive influence of the orthodontic treatment on craniofacial growth in BS subjects could be stated, which supported the findings of an implant study conducted earlier, using Delaire face mask therapy, by Rune *et al.* (1982).

Munro (1979) reported that the mandible was prognathic. However, the present sample group had effective mandibular lengths within the normal range. For this reason, the maxillomandibular differential probably reflects a retrognathic maxilla in the true BS and CDP cases.

Olow-Nordenram and Thilander (1989) found that the maxillary retrognathia of the more severe cases was aggravated by a craniofacial Class III tendency in the lower face. In such cases, correction was undertaken by combined orthodontics and orthognathic surgery, performed after the completion of growth.

The position of the orbitale was determined by the angular measurement SNO. The present study found that the orbitale in most cases tended to be positioned slightly posteriorly relative to the anterior cranial base (S-N). One theoretical possibility is that a true midfacial hypoplasia at the Le Fort II level may be masked in this measurement due to the short anterior cranial base influencing the antero-posterior position of nasion. Horswell *et al.* (1988) reported the same finding, but when SNO was superimposed on a normal/projected nasion, there was a recessed orbitale which was two standard deviations below the norm.

The skeletal angle of facial convexity (N-A-Pog) reflected the tendency toward flattening or concavity of the facial profile in the sample. If the chin point followed normal growth whilst maxillary and nasal growth lagged behind, this angle would reflect the worsening of profile concavity with time. This result supported the finding by Olow-Nordenram and Thilander (1989), as well as that of Horswell *et al.* (1988), who also carried out a photographic measurement of the soft tissue facial contour angle (FCA) which followed the hard tissue findings.

9.4 Data from the Photographic Study

9.4.1 Comparison Between Sample Groups and Normal Ratios Matched for Age and Sex

9.4.1.1 Group One: Mothers

There were 11 individuals in this group who were graphed against the normal ratio values derived from Farkas (1981). In general, for ratios 1 and 2, the mothers fell within normal range, with the exception of two who had a higher ratio. A small number of mothers showed a markedly reduced ratio. The small numbers in this sample and the fact that the study looked only at the previous generation, do not permit firm conclusions about the parents and relatives. Although some findings merit discussion.

Farkas (1987) gave certain guidelines as to the interpretation of proportional indices, or ratios: a reduction or increase in an index or ratio could be due to one of several factors. These include:

- 1) If one measurement was normal and the other was abnormal, either larger or smaller
- 2) If both were abnormal one larger and the other smaller, or both larger or smaller but greatly differing)
- 3) If both were normal but tending toward the opposite ends of the normal range.

Thus, a face with statistically disproportionate indices may have normal measurements (Farkas and Munro 1987). Because of these factors, one must exercise great caution in interpreting ratio/index differences. If the linear data cannot be checked and validated against direct anthropometric measurements, one can only speculate as to why in these few cases, the ratios were affected.

In this study, the mean values for all three profile ratios for mothers did not significantly differ from the mean normal values. Thus, observations of individuals were not statistically significant when pooled together. However, some parents may have presented with a clinically noticeable shortened columella, a reduced nasal protrusion, or both.

9.4.1.2 Group Two: Female Patients

A definite trend emerged: for ratio 1 and 2, female subjects did not differ greatly from the norm, except Female "E", a six year old with CDP, who had a high ratio 2 reading. However, in ratio 3, Sn-C : Sn-Prn, some in the female patient group stayed below the normal range, especially four females who were diagnosed with CDP, one of whom was classified as a 'severe symmetrical CDP'.

Paired t-tests were carried out to compare the female patients with ratio norms matched for age and sex. For ratio 1 and 2, the female patients did not differ significantly from the norm.

However, for ratio 3, ($p < 0.0005$), there was a statistically significant difference from the norm. This finding confirmed the observations from the graphs. For some individuals, ratio 3 was markedly reduced (Females B, G, H and I), whilst for others this ratio was less affected. This is borne out in the tremendous variation in the phenotype of BS or CDP with respect to nasal deformity. Holmström (1986), noted that some cases have a severely reduced columella (Sn-C) length, while in others the columella was longer, approaching even near normal range, but the nasal tip protrusion (Sn-Prn) was flattened and the ala base was retruded.

9.4.1.3 Group Three: Fathers

There were six individuals in this sample. For all three ratios, the fathers followed the normal ratio, with one exception, Father "E", whose ratio 2 and 3 were high. Paired t-tests were carried out on the sample to compare how they varied as a group from the normal values. For all three ratios, as was in the case of the mothers, there were no statistically significant differences from the norm. This finding confirmed the observations on the graphs. Speculation may be made, that the fathers represented a normal group, and did not show phenotypic traits of BS or CDP. The casual observer would find that the fathers' photographs appeared normal.

9.4.1.4 Group Four: Male Patients

There were 13 patients in this sample, which represent the largest number in all the four groups. Ten of these patients represented those diagnosed with CDP (76.9 percent) and three were diagnosed with BS (30 percent). The ages ranged from 3 years 6 months to 18 years 5 months, with a mean age of 12 years at the time the photographs were taken. The graphs of this sample group showed a trend similar to that of the female patients, but ratios 2 and 3 were affected, whilst ratio 1 remained within normal limits. Paired t-tests were performed to evaluate and compare normative versus patient data. Results showed that for ratio 1 the male patients were within the normal range, that is, there was no significant difference.

For ratio 3, : Sn-C : Sn-Prn; the ratio of the length of the columella to the degree of nasal tip protrusion, the male patient group was significantly different to the norms ($p=0.0069$).

9.4.2 Comparison between Parents and Children in the Sample Groups for Each Ratio

The photographic study assessed and compared the facial profiles between 21 pairs of patients and their parents for three ratios. Pearson's Correlation Coefficients showed no correlation between parents and patients for any of the three ratio measurements evaluated: ratio 1 ($r = -0.260$); ratio 2 ($r = -0.161$); ratio 3 ($r = 0.178$).

9.4.3 Comparison between Individual Siblings

Due to the small number of observations, no statistical evaluation was conducted between the four pairs of siblings. However, graphs of ratio relationships between siblings showed no relationship emerging for any of the three profile ratios.

Summary

The mean vertical length of the nose (N-Sn), and the mean length of the upper lip (Sn-Sto) was in the normal range for all but a few exceptions, indicating the existence of a fairly normal vertical dimension in the nasomaxillary skeleton. These findings agree with the Horswell *et al.* (1988) photogrammetric and anthropometric study. The length of the columella appeared shorter in the patient groups. Nasal projection, (Sn-Prn), was decreased in both male and female patient groups, mirroring the underlying maxillary hypoplasia. These findings supported the Holmström (1986) description of nasal deformity pertaining to shortened columellae or nasal tip retrusion.

The ratios from the linear data demonstrated the effect of the syndrome on the dimensions of the columella and nasal tip projection, as these readings were components of ratio 3 (Sn-C : Sn-Prn), which was reduced in many cases. Again, caution is required in interpreting the ratio measurements and the best way to validate these findings would be to follow up with direct anthropometric measurements (Farkas and Munro, 1987).

CHAPTER 10. CONCLUSIONS

10.1 General Conclusions

In the present multinormative cephalometric study of 11 patients, that is, one in which age and sex were taken into account, there were statistically significant findings for the following variables:

- 1) The anterior cranial base length (S-N) was reduced ($p=0.0001$); male patients had significantly larger measurements for S-N than female patients ($p=0.015$).
- 2) Total maxillary length (PNS-A) was reduced ($p=0.0002$).
- 3) The skeletal angle of facial convexity (N-A-Pog) was reduced, that is, more values were in the negative range, indicative of profile concavity ($p=0.0019$).
- 4) Effective maxillary length (Co-A) was reduced ($p=0.0005$).
- 5) Mandibular-maxillary differential (Co-Gn to Co-A) was increased ($p=0.0005$).
- 6) The orbitale position was recessive, as the SNO angle was reduced ($p=0.044$).
- 7) Lower anterior face height (ANS-Men) was increased ($p=0.046$).
- 8) Upper to lower face height ratio (UFH:LFH) was increased ($p=0.0009$).
- 9) Nose prominence was reduced ($p=0.001$).
- 10) Upper lip thickness was increased ($p=0.038$).
- 11) Upper lip "strain" was increased (with norm set at 1mm for strain, $p=0.0054$).
- 12) Lower lip to H-line was prominent ($p=0.00005$).

The photographic study suggests that there are statistically significant differences between parents and their children in the third ratio (Sn-C :Sn-Prn). These differences may indicate that either the columella in the patient groups was shorter; there was a reduced nasal tip protrusion, or various combinations of these factors occur.

The main difficulty in assessing photographic and cephalometric data in any syndrome is the small patient sample size. Syndrome identification is further complicated when the aetiology is unknown. Not only is there heterogeneity of the genetic defect plus incomplete penetrance, or one cause with multiple symptoms, but also the possibility of heterogeneity of aetiology, that is, one syndrome with multiple causes.

The hard tissue cephalometric observations supported findings in the current literature (Olow-Nordenram, Sjöberg and Thilander, 1986; Horswell *et al.* 1988), that the nasomaxillary complex was affected in BS and CDP. The soft tissue cephalometric analysis confirmed the presence of nasal retrusion and upper lip thickness due to its unique convex morphology. The lower lip prominence reflected

an underlying Class III skeletal relationship, which was most often due to maxillary retrusion in the present sample. Contrary to findings by Horswell *et al.* (1988), there was a normal nasolabial angle in 55 percent of cases. The photographic study showed no similarity between parents and their children or between siblings, for the three ratios measured. This finding did not support studies which stated that the unusual facies was genetically inherited (Olow-Nordenram *et al.* 1986), although the facial deformity may still appear in families due to its multifactorial aetiology, and suggested that the individuals studied had significantly different midfacial morphology from normal subjects and their relatives.

10.2 Recommendations for Future Research

It would be useful to conduct a similar study on a larger sample and using direct anthropometric measurements in order to verify the photographic findings of Part II of this study. Such a study would broaden the clinical use of anthropometry to obtain racial norms, study other syndromes, as well as to assess objectively the results of plastic surgery. Rigid standardisation of records is requisite.

An ideal situation would involve a database of all patients diagnosed with the various subcategories of CDP, with records of their family members placed on computer over a period of time. The establishment of an adequate database would greatly facilitate research into the incidence of these syndromes in Australia and be able to retrieve information and obtain hard copies of records for multi-centre analysis. Computer scanning of records of these patients is already possible with present technology. Such records could include strictly standardised photographs and cephalograms before and after treatment, with the full details of pregnancy history, associated malformations, latest biochemical tests, early childhood growth progress charts, and details of multidisciplinary treatment used.

Furthermore, patients diagnosed with BS could be compared with the subcategory of CDP that is mild, and called the Conradi-Hunermann Type, to help clarify the classification system. Patients requiring genetic counselling and health professionals treating cases severe enough to warrant orthodontics, as well as orthognathic and plastic surgery over time, would benefit greatly from this research database. These data would lead to improved preventive measures, diagnostic and treatment techniques and the cost-effective use of resources. Population trends for various syndromes would provide information and direction for government funding into education and prevention, and fully utilise already available technology.

To study the teratological effects of drugs, and the efficacy of vitamin K preventive regimes, prospective longitudinal studies on 'at risk' women and their offspring are indicated. New digital radiographic techniques would significantly reduce radiation dosage in the screening for the punctate calcification of bone seen

in CDP of 'at risk' newborns, of for example, mothers on anticonvulsants or anticoagulants.

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