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SPECIAL ISSUE

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Live up to the Hype?**

**Are We Prepared for
Our Test Results?**

**Should Genomes Be
Screened at Birth?**

**Should You Share Your
Genetic Data Online?**

**Will a Genome Test Lead
to Healthy Behaviours?**

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Genomic Testing as a Lifetime Health Resource?

AINSLEY NEWSON & DAVID AMOR

If lives could be saved by being "forewarned" by a genomic test, should we perform genomic testing of all babies at birth?

Rapid developments in genomic testing methods have made the sequencing of a person's DNA faster and cheaper than ever before. The latest gene sequencing machines can sequence all 20,000 human genes in less than 3 days at a cost of less than \$2000 per person. This is comparable to the cost of testing just one gene using slightly older sequencing machines.

But what are the scientific and ethical issues involved in the use of genomic information as a "lifetime health resource"? Are we ready for the wide application of genome testing in people who are otherwise well?

Genomic Testing

Genomic testing has proven to be a powerful research tool, and already has facilitated many major research breakthroughs,

particularly in the fields of cancer medicine and rare genetic diseases. More recently, clinical genomic testing has emerged as a component of individual health care, and a range of genomic tests are now offered by public and commercial laboratories.

Clinical genomic tests are already highly effective and cost-efficient in the diagnosis of patients with rare or unusual presentations. A recent study published in *Genetics in Medicine* by the Melbourne Genomics Health Alliance (<http://tinyurl.com/z94bw7z>) demonstrated that genomic testing of previously undiagnosed children could identify a diagnosis in approximately half of children tested, at a much higher diagnosis rate and lower cost than traditional diagnostic tests. In many cases the diagnosis led to significant changes to the child's treatment – such as a child experiencing seizures being able to be treated with a vitamin. It also provided fami-



lies with valuable information and an ending to what for many had been a prolonged “diagnostic odyssey”.

The success of genomic testing for the diagnosis of rare disorders has in turn raised the question of whether these tests should be undertaken in healthy people before they get sick. The potential benefits of early diagnosis are obvious, particularly for conditions for which prevention is possible. Further, if lives could be saved by being forewarned by a genomic test, why not perform genomic testing in all babies at birth?

Newborn Screening

Newborn screening is, in fact, a well-established public health endeavour in Australia and around the world. However, these programs do not yet utilise genomic technology. In existing newborn screening programs, parents give permission for some blood to be taken from their baby’s heel soon after birth. This sample is used to check for a selection of about 30 diseases for which early identification or treatment is beneficial.

One of the best examples of diseases screened is a metabolic condition called phenylketonuria (PKU). Diagnosis of PKU will enable parents to provide a tailored diet that prevents their child from developing an intellectual disability. However, PKU and other disorders tested by current newborn screening tests represent just a fraction of the thousands of rare genetic disorders that can affect children. Genomic sequencing provides the opportunity to test for all of this with a single test.

The potential benefits of genomic newborn screening are clear. A baby identified as being at risk of sudden cardiac death could be placed on protective medication that may save her life. A baby born with a high risk of cancer could have extra

cancer screening tests to make sure that his cancer was detected at an early stage. A baby born with a predisposition to a fatal drug reaction could carry a warning bracelet to make sure she was never given the drug. And in all of these situations there may also be a health benefit to the baby’s parents and other relatives who may unknowingly carry the same genetic risk.

Moreover, there is no need for the health benefits of genomic sequencing to end in childhood. Genomic information can provide a blueprint for individual health that is relevant through all ages and stages of life. Therefore a test performed in a newborn represents a potential lifetime health resource.

Should We Use Genomic Screening in All Newborns?

The above individual examples make a strong case for genomic testing of newborns. However, the huge resources required for such a program also need to be considered.

First and foremost, we must consider the financial cost. Even if costs could be reduced to \$1000 per genome sequence for each baby tested, to test all 300,000 Australian babies born each year would cost \$300 million for the testing alone. When the costs of counselling and interpretation of the huge amount of information are added, the cost would be considerably higher. In contrast, current newborn screening using biochemical techniques costs just \$20 per baby screened.

Second, we must acknowledge that our ability to interpret genomic information is in its infancy. Even for those whom we know, with certainty, have a genetic condition, it can be challenging to identify a specific disease-causing gene change. A particular problem is that we all carry literally millions of

these genetic variants. Of these, perhaps a handful might cause disease but the remainder are harmless. Another problem is that a particular variant may not always lead to disease in everyone who has it.

Sorting out which variants we need to be worried about is far from straightforward, particularly when the people being tested are healthy newborns. Unfortunately, genomic testing is not yet the precise tool that some might imagine it to be.

Therefore genomic testing of all newborns would lead inevitably to some healthy babies being incorrectly diagnosed with a serious disorder, while other babies who did have a genetic condition could have their diagnosis missed. We would also have to work out how to manage and use this complex information as the child grows up.

We therefore have two good reasons, huge financial costs and difficulty interpreting results, that go some way to explaining why genomic sequencing is not yet ready for use as a tool for newborn screening. But what if these two obstacles could be removed, as may possibly be the case in the future? Would there still be other reasons to avoid genomic screening in newborns?

Ethics and the Lifetime Health Resource

Common to debates about genetics are issues surrounding the privacy and disclosure of genetic information, and the storage or future use of test samples and data. However, Australia has good legal and regulatory controls in place. So if it's affordable, easier to interpret and well-protected, do other ethical concerns arise?

First, the way that consent to this test is obtained needs further consideration. We know that parents already have poor recall of newborn screening. It's also widely recognised that the standard ways of gaining consent to a health test are not suitable for genomic testing. If the test is going to become more complex we'll need to engage parents in a way that promotes the significance of the test and its future importance without negatively impacting acceptance of existing screening methods.

Second, in newborn testing it will be the child's parents or guardians who are providing this consent, but not all of the information is going to be relevant in childhood. A well-established principle in genetics is that genetic information not relevant to childhood should not, in general, be disclosed until the child in question is old enough to be part of the decision.

We think this principle still has a strong foundation and should not be routinely overridden for the sake of it being convenient to obtain this information. This may mean that certain information is not disclosed to parents, but is left for the child to decide about later in life.

If we are to uphold this principle in genomic newborn screening, it will add to the resource costs. But if we don't uphold this principle, we risk undermining the future autonomy of the child who is tested.

Third, genetic information is shared within families, so information obtained in genomic newborn screening may have relevance for other family members too. While the prospect of these benefits cascading out is important, this will also give rise to issues over appropriate communication and possible further counselling in these additional people.

Finally, alongside the significant hype about genomics, a quieter discussion is taking place around the appropriate place and use of genomic testing in medicine. Unfettered use of genomic testing will likely lead to false positive and false negative results, or to overdiagnosis. Overdiagnosis occurs when a diagnosis is "correct" but leads to harmful interventions or labelling. As an example, for each baby who benefits from increased cancer screening there will undoubtedly be many others who undergo regular screening when they would never have developed cancer. Because screening itself can carry risks, these harms also need to be considered.

If we are to take seriously the point about appropriate use of genomic technologies, we need to commit to using them when they are likely to be effective and when the majority of ethical concerns have been addressed. For this reason, mass population screening programs of any kind tend to be led by a specified and important health care need, rather than a desire to use a particular technology. Criteria for screening acceptability from the World Health Organisation emphasise this alongside other considerations such as the need for an intervention, population acceptability and economic balance.

We suggest that any decision about creating lifetime health resources should only be done when we have a clear idea about the benefits this may give rise to, with appropriate protections in place for the custodianship and use of the information gained.

Engagement with Health

While genomic testing has important potential to optimise human health, we are not yet ready – medically or ethically – to obtain the genomes of all newborns. If a lifetime health resource is to come to fruition, we need to think more about cost-effectiveness, custodianship of the data and engagement with families over time.

We have suggested some reasons why it's still premature to routinely sequence the genome of all newborns. This conclusion was also reached by the Global Alliance for Genomics and Health, which will soon release a set of recommendations on genomic technologies in newborn screening. We need to think more about the knowledge we'd gain and how it could be used in childhood and beyond. In so doing, we should not ignore the interests of the adults whom the children will eventually become.

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