The challenge of overdiagnosis begins with its definition

Overdiagnosis means different things to different people. S M Carter and colleagues argue that we should use a broad term such as too much medicine for advocacy and develop precise, case by case definitions of overdiagnosis for research and clinical purposes.

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The implicit social contract underpinning healthcare is that it will reduce illness and preventable death and improve quality of life. But sometimes these promises are not delivered. Sometimes health services take people who don’t need intervention, subject them to tests, label them as sick or at risk, provide unnecessary treatments, tell them to live differently, or insist on monitoring them regularly. These interventions don’t improve things for people; they produce complications or illness, reduce quality of life, or even cause premature death. Active health intervention is not always a good thing: it can be “too much medicine,” or produce what is often called overdiagnosis.

Although the concept of overdiagnosis has been described in the literature for nearly 50 years in relation to cancer screening, it was Welch and colleagues’ 2011 book, Overdiagnosed: Making People Sick in the Pursuit of Health, that popularised the term. Overdiagnosis is now an acknowledged problem for patients, clinicians, researchers, and policymakers; it is discussed in journals and at specialist conferences and addressed through policy and practice initiatives.

There is, however, no formal, agreed definition of overdiagnosis. Rather, the word has become a banner under which disparate people with similar general concerns can unite. This vagueness and breadth allows the appearance of unity but does not serve the more exacting demands of research and healthcare. Here we examine the meanings of overdiagnosis more closely and discuss related challenges for healthcare professionals, patients, and researchers. If overdiagnosis is to be understood and mitigated, the broad concept should be subdivided into different problems and its ethical dimensions better acknowledged.

Towards a definition

Overlapping concepts

Aspects of overdiagnosis overlap with existing movements in health policy and practice such as evidence based medicine, patient centred care, strategies for disinvestment, and quality and safety in healthcare, especially preventing iatrogenic illness and low value healthcare. A careful comparison with these better defined problems will allow those concerned about overdiagnosis to learn from related work, avoid redundant work, and better identify what is unique about overdiagnosis.

Social and ethical dimensions of overdiagnosis

A deeper understanding of overdiagnosis requires moral as well as technical analysis. It is tempting to seek a purely technical definition of overdiagnosis that excludes context, values, and ethics. But the much debated balance of benefit and harm central to the concept of overdiagnosis is also central to healthcare ethics. Technical definitions of overdiagnosis quickly confront moral considerations, such as what types of benefit or harm should matter; how different benefits and harms, or benefits and harms to different people, should be weighted; whether benefits and harms should be measured in individuals or systems and society; and who should judge which benefits and harms matter. For example, the harms of overdiagnosis are often side effects of treatment. But which side effects are important enough to include in any measurement of harm? Are some side effects more important than others? Who should decide—patients, clinicians, or researchers? And what if they disagree?

To understand, define, and respond to overdiagnosis, we also need to understand complex healthcare systems and the people who use and serve them. Hoffman and Kanzaria argue that
Multiple related concepts

The table sets out an inclusive map of concepts related to the problems of overdiagnosis or too much medicine. Each concept is contestable; we provide rough rather than authoritative definitions. As illustrated, these concepts are interrelated. For example, overdetection and overdiagnosis are drivers of overtreatment and overutilisation. Disease mongering probably leads to overdiagnosis. Overmedicalisation permits or encourages disease mongering, overtreatment, and overutilisation.

The concepts in the table range from broad to narrow. The broadest is arguably overmedicalisation—for example, defaulting to biotechnological responses rather than existential wisdom to deal with our fear of death or defining disruptive behaviour in children as a medical problem requiring drug treatment when social or behavioural interventions may be equally effective with a lower probability of harm. These are cultural problems, connected to profound questions about what constitutes a good human life. Other overdiagnosis problems are far narrower and more instrumental—for example, how mammography services can minimise the number of women experiencing both late stage breast cancer and unnecessary treatment of early stage breast cancer. The narrowest concepts in the table are arguably overdetection and overdiagnosis. The narrower the concern, the more concrete and individually focused it is, and the more amenable to quantitative measurement.

This leads us to our central observation. The word overdiagnosis is being used in two ways. It is used as the umbrella term for most of the concepts in the table, but it is also used to label one narrow concept in the set. Using the word overdiagnosis in both the broad and the narrow sense is imprecise for researchers and clinicians and potentially confusing for the public and decision makers. We suggest it should stop. If concerned parties continue to use the word overdiagnosis to mean several different things at different levels of generality, they are likely to talk past one another or waste energy on unnecessary disagreement. Instead, a new umbrella term (such as too much medicine or less is more medicine) could be used for the broad conception summarised at the beginning of this article. Adopting a new umbrella term would spare the word overdiagnosis for the narrower, more precise, meaning.

Such a change could help resolve some disagreement over the extent and scope of the concept. Issues such as overtreatment, overutilisation, overmedicalisation, and disease mongering cannot be readily shoehorned into a narrow definition of overdiagnosis but fit easily into the broad set of too much medicine. Debate over the narrow concept of overdiagnosis could then be restricted to a few central concerns (box 1), such as whether it occurs in symptomatic, or only asymptomatic, people and the relations between overdiagnosis, risk factors, and false positive findings.

Precise definition for specific purposes

Broader and narrower conceptions of overdiagnosis are suited to different purposes. Different interested parties are working with different implicit understandings of the concept. They are focused on different conditions and problems (mental health, cancer screening, drug promotions, etc) and have different purposes: unpacking the logical structure of overdiagnosis, explaining how it occurs, measuring it, advocating for political change, or developing practical tools to change clinical or policy practice.

Each of these purposes is important, but they occur at different levels of generality. Advocacy, for example, can employ the broadest conception. It is here that a new umbrella term such as too much medicine may be most helpful because it is arresting, inclusive, and easy to understand. In contrast, epidemiological measurement of overdiagnosis requires greater precision and encodes multiple assumptions specific to each condition studied. Methods for measuring overdiagnosis in a particular condition, along with the assumptions underpinning these methods, implicitly define overdiagnosis in that condition. These methods and assumptions will change disease by disease, test by test.

For all conditions, measuring overdiagnosis requires good quality, large scale data collection over time, and an international effort to agree on appropriate analysis methods for that condition. This agreement has proved difficult. More work has been done on methods for estimating overdiagnosis in cancer screening than in any other condition, but still there is deep disagreement about appropriate methods, and all current methods carry considerable risk of bias. This process needs to be repeated for other conditions, because in each condition the drivers of overdiagnosis and potential sources of bias differ. For example, overdiagnosis of breast, prostate, thyroid, and lung cancer is driven largely by screening and early detection programmes, so potential biases include lead time; confounders include population trends in cancer risk factors such as use of hormone therapy, bodyweight, and smoking. In contrast, the drivers of overdiagnosis in high blood pressure, high cholesterol, and diabetes include more frequent testing and regular changes in the thresholds for what is considered abnormal (the threshold for high blood pressure, for example, has fallen from 160/100 mm Hg to 140/90 mm Hg). Rather than responding to lead time, methods for estimating overdiagnosis in these conditions must respond to threshold changes, but it is not yet clear how this should be done. For each condition the pattern repeats: different drivers, different biases, and so different methods.

Beyond advocacy and epidemiology, others are examining overdiagnosis for different purposes. Social scientists are studying how clinicians, decision makers, patients, and citizens make sense of overdiagnosis and too much medicine. For example, some of us are studying general practitioners’ understanding and management of overdiagnosis in prostate cancer in Australia and the UK and Australian decision makers’ understanding of overdiagnosis in mammography; we are mapping the diversity of what overdiagnosis means to different stakeholders and the deeply held values that support their understandings. These are conceptualisations based in everyday practice and will inform the policy response to the problem. At a more abstract level, scholars in the philosophy of medicine are studying the logical structure of overdiagnosis, developing precise formal definitions and typologies and sound arguments on questions such as whether overdiagnoses, false positive results, and misdiagnoses intersect or are mutually exclusive. For each of these purposes, the word overdiagnosis is being used in two ways. It is used as the umbrella term for most of the concepts in the table, but it is also used to label one narrow concept in the set. Using the word overdiagnosis in both the broad and the narrow sense is imprecise for researchers and clinicians and potentially confusing for the public and decision makers. We suggest it should stop. If concerned parties continue to use the word overdiagnosis to mean several different things at different levels of generality, they are likely to talk past one another or waste energy on unnecessary disagreement. Instead, a new umbrella term (such as too much medicine or less is more medicine) could be used for the broad conception summarised at the beginning of this article. Adopting a new umbrella term would spare the word overdiagnosis for the narrower, more precise, meaning.

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Beyond advocacy and epidemiology, others are examining overdiagnosis for different purposes. Social scientists are studying how clinicians, decision makers, patients, and citizens make sense of overdiagnosis and too much medicine. For example, some of us are studying general practitioners’ understanding and management of overdiagnosis in prostate cancer in Australia and the UK and Australian decision makers’ understanding of overdiagnosis in mammography; we are mapping the diversity of what overdiagnosis means to different stakeholders and the deeply held values that support their understandings. These are conceptualisations based in everyday practice and will inform the policy response to the problem. At a more abstract level, scholars in the philosophy of medicine are studying the logical structure of overdiagnosis, developing precise formal definitions and typologies and sound arguments on questions such as whether overdiagnoses, false positive results, and misdiagnoses intersect or are mutually exclusive.
Meanwhile, on the front line of medicine and public health, strategies are being developed to decrease both too much medicine and specific cases of overdiagnosis. Different groups—general practitioners, clinical specialists, policy makers, citizens, and patients—face different challenges. For those on the front line, the most important step may be accepting the unsettling general sense of too much medicine: that medicine is an uncertain practice, that healthcare may be harmful, and that attending to harms is as important as attending to benefits. Such a cultural shift will make it easier to translate the precise work done by researchers on specific cases of overdiagnosis. Such translation has begun, facilitated by professional organisations and consumer groups including Consumer Reports, the Academy of Medical Royal Colleges, the American Board of Internal Medicine Foundation, the National Institute for Health and Care Excellence, and the Royal College of General Practitioners.

From here

We may never agree on a single definition of overdiagnosis. But we can and should be more explicit about what we mean when we use the term, including the breadth or precision, relevant conditions, assumptions regarding benefit and harm, and purpose (box 2). Clarifying these dimensions will serve our ultimate goal of getting a better grasp on the important problem of too much medicine.

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Box 1: Classic overdiagnosis, narrowly conceived, precise, and condition specific

Thyroid cancer provides a useful example of the narrow sense of overdiagnosis. 6 In the US, the rate of diagnosis of thyroid cancer has tripled over the past 30 years, from 3.6 cases/100,000 in 1973 to 11.5 cases/100,000 in 2005, with most of the extra diagnoses being of papillary cancer. This rise in thyroid cancer diagnosis has been linked to the increased use of portable ultrasound machines for screening asymptomatic people. Before ultrasonography was available lesions were identified by clinical examination, usually when patients presented with symptoms. Now lesions as small as 2 mm can be identified and biopsied. If malignant cells are found, patients are offered surgery. 7-9 By 2010, the rates of which in the US have increased by 60% over the past 10 years. Despite the rise in diagnoses and treatment, the death rate from thyroid cancer has remained stable. 10 This suggests that the extra diagnoses and treatments are not reducing morbidity or mortality.

To tackle this example of overdiagnosis we need to understand the natural course of these very small lesions, which may grow too slowly to become symptomatic during the person’s lifetime. Are they one end of a spectrum of tumour behaviour ranging from indolent to aggressive, and if so, can we identify which will remain indolent? Or are they a separate pathological phenomenon? What are the relevant histopathological and genomic features that might answer these questions?
Box 2: Actions to clarify overdiagnosis and too much medicine

- Recognise that both overdiagnosis (narrow) and too much medicine (broad) are social and ethical problems, not just technical and scientific problems.
- Stop using the word overdiagnosis to refer to the broad too much medicine problem.
- Recognise the need to respond to overdiagnosis specialty by specialty and condition by condition.
- Develop a clear definition of too much medicine for use in public and political communication.
- Engage with others working in closely related movements, such as low value care and patient centred care, and systematically study the similarities and differences.
- Recognise the potentially competing values at stake in defining and tackling overdiagnosis and too much medicine and develop inclusive strategies to take full account of these.
- Promote public debate on the inherent uncertainty and limitations of healthcare and their implications for overdiagnosis and too much medicine.

Table 1 | Concepts related to too much medicine or “less is more medicine,” possible drivers, reasons for lack of net benefit, and examples

<table>
<thead>
<tr>
<th>Concept</th>
<th>Meaning</th>
<th>Drivers</th>
<th>Reasons for harm, or lack of net benefit</th>
<th>Examples (all assume no net benefit)</th>
<th>Inter-relation with other concepts</th>
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<tbody>
<tr>
<td>Overdiagnosis (in the narrow sense)</td>
<td>An (asymptomatic) person is diagnosed with a condition; that diagnosis does not produce a net benefit for that person</td>
<td>Disease mongering (see definition below); Expanding disease definitions by lowering thresholds for what is considered abnormal; Early detection programmes (screening); Defensive medicine; Guidelines or incentives that encourage testing</td>
<td>Non-medical care is more effective or beneficial; Disease is indolent, inconsequential, or will regress; Treatment produces no benefit or more harm than benefit (eg, side effects); Labelling causes psychological or social harms; Intergenerational effects of parental diagnosis lead to “at risk” offspring</td>
<td>Non-progressive breast cancer detected through population mammographic screening; High blood pressure diagnosed in asymptomatic people because of lowered thresholds for diagnosis</td>
<td>Overdiagnosis often leads to overdiagnosis, which in turn leads to overtreatment and overutilisation. Overdiagnosis can be difficult to distinguish from misdiagnosis and false positive results. Expanded definitions, disease mongering and overmedicalisation likely to increase overdiagnosis.</td>
</tr>
<tr>
<td>Overdetection</td>
<td>A health related finding is detected in an (asymptomatic) person, probably by testing technology. That finding does not produce a net benefit for that person</td>
<td>Disease mongering; Expanding disease definitions; Encouraging well people to be tested; Development of increasingly sensitive testing technologies (eg 3D digital mammography); Cultural norms about prevention (eg “an ounce of prevention is worth a pound of cure”); Overuse of expensive testing technologies to justify their expense; Defensive medicine; Guidelines or incentives that encourage testing; Direct to consumer testing (eg internet-enabled genetic testing)</td>
<td>Finding indicates something that is indolent, inconsequential, or would have regressed; Labelling causes psychological or social harms; Intergenerational effects of parental diagnosis leading to “at risk” offspring</td>
<td>Incidentalomas. * PSA testing in asymptomatic men; Detection of sub-segmental pulmonary embolism</td>
<td>Overdetection may lead to overdiagnosis, overtreatment, and overutilisation. Expanded definitions, disease mongering, and overmedicalisation likely to increase overdiagnosis. Overdetection can be difficult to distinguish from false positive result.</td>
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<tr>
<td>False positive</td>
<td>Classically: a test indicates that a condition is present, when in fact it is not. In practice: there is often a “grey zone” between normal and abnormal tissue or function, and in this zone it is not always possible to distinguish false positive</td>
<td>The rate of false positives is a characteristic of the test technology and dependent on our biological and technical knowledge. The boundary between a true positive, a false positive, and an overdiagnosis (in the narrow sense) is always set</td>
<td>Person wrongly informed that they do, or may, have the condition or risk factor. This can cause psychological or social harm and result in further unnecessary testing, especially if invasive</td>
<td>Recall after cancer screening, with negative result on retest</td>
<td>False positives can be difficult to distinguish from true positive and overdiagnosis. If it becomes a working diagnosis a false positive result can cause overtreatment and overutilisation (unnecessary follow-up tests).</td>
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**Table 1 (continued)**

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<tr>
<td>Misdiagnosis</td>
<td>Incorrect diagnosis of a symptomatic person with a condition they do not have</td>
<td>Fear of missing a serious diagnosis. Lack of diagnostic specificity. Limited diagnostic resources</td>
<td>Wrong treatment. Concurrent neglect or undertreatment of underlying disease</td>
<td>Patients with fever from other causes have malaria diagnosed. Patients with infective cough diagnosed with asthma</td>
<td>May lead to overtreatment or overutilisation. Disease mongering may increase misdiagnosis</td>
</tr>
<tr>
<td>Overdiagnosis</td>
<td>Provision of treatment with no net benefit by individual clinicians to their patients</td>
<td>Defensive practice. Guideline driven care. Tendency to treat rather than to watch and wait</td>
<td>Treatment produces no net benefit or more harm than benefit (eg, side effects)</td>
<td>Broad spectrum antibiotic use in viral infection. Antidepressants when non-drug therapies would be equally or more effective. Proposals for mass medication—for example, to treat all adults with a polypill or statins</td>
<td>Overdiagnosis, overdetection, misdiagnosis, expanded definitions, disease mongering, and overmedicalisation tend to lead to overtreatment. Overdiagnosis is a form of overutilisation</td>
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<tr>
<td>Overutilisation</td>
<td>Establishment of standard practice in health services or systems that do not provide net benefit to patients or citizens</td>
<td>Expanded definitions. Disease mongering. Guideline driven care. Expensive diagnostic equipment requiring high usage to justify expense</td>
<td>In individuals, harms of overdiagnosis, overtreatment, misdiagnosis. In systems, opportunity costs and economic costs</td>
<td>Routine MRI for lower back pain. Call-recall systems to encourage all patients to attend for an annual pelvic examination and cervical smear</td>
<td>All of the other concepts in this table are likely to produce overutilisation of certain services</td>
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<tr>
<td>Expanded definitions or disease mongering</td>
<td>Expansion of official disease or risk categories, or creating new conditions or promoting more frequent diagnosis of recognised conditions, without net benefit to patients or citizens. Creating “diseases” out of behaviour or feelings that are within normal human experience, and promoting those diseases to the public to encourage use of health services, especially tests and medicines</td>
<td>Overmedicalisation. Expert committees tend to expand disease categories. Profit motivated industries benefit economically as more people are diagnosed and treated</td>
<td>More people labelled as diseased, pre-diseased, or at risk—labelling psychologically or socially harmful. Treatment of newly diagnosed people produces no benefit or more harm than benefit (eg, side effects)</td>
<td>Expanding pre-diabetic so previously normal people are labelled prediabetic. Labelling low libido in women as female sexual dysfunction</td>
<td>May encourage overmedicalisation. Likely to increase overdiagnosis, overdetection, overtreatment, and overutilisation</td>
</tr>
<tr>
<td>Overmedicalisation</td>
<td>Altering the meaning or understanding of experiences, so that human problems are re-interpreted as medical problems requiring medical treatment, without net benefit to patients or citizens</td>
<td>All other concepts in table will drive overmedicalisation; the converse also seems likely</td>
<td>Provides an environment conducive to expanded definitions, disease mongering, overtreatment, overdiagnosis, overtreatment, and overuse</td>
<td>Fear of death treated as something that can be fixed with biotechnology rather than something requiring existential wisdom. Disruptive children treated with drugs</td>
<td>Overlaps with all other concepts in table. Extremely broad: occurs well beyond overdiagnosis in the narrow sense</td>
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Readers are likely to experience at least one of the following reactions. “But that’s a driver of overdiagnosis” (overdetection, expanded definitions, disease mongering, overmedicalisation). “But that’s a consequence of overdiagnosis” ( overtreatment, overutilisation, overmedicalisation). “But that’s caused by so many things other than overdiagnosis” ( overtreatment, overutilisation). “But that is, by definition, not overdiagnosis” (misdiagnosis, false positives). This is precisely our point. A term such as “too much medicine” or “less is more medicine” should be adopted as the umbrella term. This would readily accommodate everything in this table. Then disagreement would be limited to the much more fruitful question of how to define overdiagnosis in the narrow sense (that is, how to determine what belongs in the first row of this table).