THE CHALLENGE OF DEFINING OVERDIAGNOSIS

A broad, inclusive conception of overdiagnosis is useful for advocacy, but other tasks require more precise thinking

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‘Overdiagnosis’ is now an acknowledged research and policy problem (Figure 1) with serious implications for clinical practice and patient welfare.\(^1\),\(^2\) There is, however, no formal, agreed definition of the term. In the absence of a definition, we suggest the following as a description of the broad domain referred to as ‘overdiagnosis’.

The implicit social contract underpinning health care is that health professionals’ interventions will, to the extent possible, reduce illness and preventable death in society, and improve quality of life. But sometimes these promised benefits are not delivered; sometimes health professionals and programs over-reach. They take people who don’t need intervention, label them as sick or ‘at risk’, and give them unnecessary treatments.\(^3\) This can reduce quality of life, or even cause people to die prematurely. Active health intervention is not always a good thing: it can be too much medicine.

Overdiagnosis in this broad sense has a revelatory, unsettling quality. It transforms familiar, helpful healthcare into strange, potentially dangerous healthcare. The generality of the concept, and the lack of an agreed definition, also create a conundrum. Diverse groups can discuss ‘overdiagnosis’ in this broad sense. They can share concern, feel unified, and work together for change.\(^1\),\(^4\),\(^5\) But this can occur with no explicit agreement on exactly what overdiagnosis is.

We aim to examine the meanings of overdiagnosis more closely. What lies under the surface of the broad concept? What are its ethical and social dimensions? What more specific concepts might it relate to? Who is invested in defining it, and why? And what challenges will be faced by anyone seeking to achieve a broadly-agreed definition?

We first argue that overdiagnosis is inseparable from values, ethics and social context. We then make our central point: that the word ‘overdiagnosis’ is used to label many different, interrelated concepts.
For advocacy purposes, this may allow the appearance of unity and so be useful. But it will not serve the more exacting challenges of research and service provision. If the problem is to be understood and mitigated, the broad concept may need to be subdivided into very different problems, and more precise conceptualisation developed for each one.

**Overdiagnosis cannot be separated from social context, values or ethics**

Overdiagnosis occurs in complex social and interpersonal systems: general practitioners attempting to help their patients in a system loaded with incentives and penalties, citizens trying to comply with health advice, companies required to deliver a profit, defensive medico-legal systems, and bureaucratic key performance indicators. To understand and respond to overdiagnosis, we need to understand and respond to this social dimension.

Overdiagnosis is a moral concern. It is taken to be a bad thing that should be prevented where possible. Importantly, overdiagnosis occurs only when interventions have *not benefited* or have *harmed*. Benefit and harm, central to healthcare ethics, are thus built into the concept of overdiagnosis. It may be tempting to seek a purely technical definition of overdiagnosis that excludes values and ethics. But any such attempt will 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; the unit of analysis (benefits and harms in individuals and/or systems and society); and who should judge which benefits and harms matter. These are not simple pragmatic or technical problems: they are contested ethical questions. The answers require moral, not just technical, justification.

‘**Overdiagnosis’ is similar to, and overlaps with, other problems**

Aspects of ‘overdiagnosis’ are similar to other, existing concerns in health policy and practice. There are overlaps with evidence based medicine, strategies for disinvestment, quality and safety in health care (especially preventing iatrogenesis and identifying low-value health care), and patient-centred care. Defining overdiagnosis will require consideration of its relationship to these pre-existing areas of concern.
The word ‘overdiagnosis’ is being used to refer to related but quite different problems

There are also complications inside the field of overdiagnosis. As we have argued elsewhere,\textsuperscript{7, 10} the term is used to refer to quite different, although related, concerns. A meeting about ‘overdiagnosis’ is likely to attract people interested in some or all of the following:

1. Expert panels expanding the parameters of conditions, so more people are diagnosed with disease or ‘pre-disease’ (e.g. with mental illnesses, diabetes, cardiovascular disease);\textsuperscript{11}
2. Pharmaceutical companies giving new diagnostic labels to normal physiological processes, encouraging healthy people to think of themselves as ‘at risk’, or refocusing clinical outcomes on surrogates (e.g. bone density instead of fractures);\textsuperscript{12}
3. Screening for risk factors and early disease in well people, thus increasing the incidence (of diagnosis) of non-communicable diseases (e.g. cancers, cardiovascular disease);\textsuperscript{13}
4. Detecting incidental abnormalities of uncertain significance via medical imaging (e.g. detection of thyroid nodules when investigating neck pain, or mild aortic aneurysm when investigating back pain);\textsuperscript{14}
5. Aggressive treatment of conditions detected via screening when this, on probabilities, is likely to be more harmful than watchful waiting (e.g. radical prostatectomy following Prostate Specific Antigen screening in asymptomatic men);\textsuperscript{15} and
6. Reallocation of resources from general and primary care for ill people towards resource-intensive testing and screening services for well people, decreasing health service utility.\textsuperscript{16}

Table 1 sets out a range of concepts related to the problems of ‘overdiagnosis’ or ‘too much medicine’. Everything in this table is highly contestable: we do not claim that each of these items should be included in the concept ‘overdiagnosis’. Rather, Table 1 maps the concepts currently connected, in some way, to the problem. There are several observations to be made across this set.

First, there are different types of relationship between these concepts. For example: overdetection and overdiagnosis probably cause overtreatment and overutilisation. Disease mongering probably drives overdiagnosis. Disease mongering may be easier to achieve in the context of overmedicalisation. The concepts are overlapping and connected: they are not mutually exclusive.
Second, Table 1 contains both broad and narrow concerns. Broad concerns include, for example, the uninterpretable clouds of health-related genomic data that will soon be attached to each of us, or the use of biotechnological fixes instead of existential wisdom to address our fear of death. These are cultural problems, connected to profound questions about what constitutes a good human life. The broadest concepts in Table 1 are arguably overmedicalisation and disease mongering. Other ‘overdiagnosis’ problems are far narrower and more instrumental: how clinicians can prescribe statins to avoid both cardiac arrest and dizziness or falling, for example, or how mammography services can minimise both late-stage breast cancer and unnecessary treatment of early-stage breast cancer. The narrowest concepts in Table 1 are arguably overdetection and overdiagnosis. The narrower a concern becomes, the more concrete and the more individually-focused it is, and so the more amenable it is to quantitative measurement.

This leads us to our central observation. The word ‘overdiagnosis’ is being used in two ways. ‘Overdiagnosis’ is used as the umbrella term for at least seven concepts (Table 1), some broad, some narrow, and existing in complex relationships. ‘Overdiagnosis’ is also used to label one narrow concept among the set of seven (notice the problematic circularity here). The narrow concept is something like this: Overdiagnosis occurs when a person (some would say: an asymptomatic person) is diagnosed with a condition that would not have shortened the person’s life or caused any symptoms. While some make a hard distinction between overdiagnosis and false positives, others emphasise that there is often an irreducible and indeterminate zone between normal and abnormal tissue or function, and that overdiagnosis frequently occurs in this zone. One example of overdiagnosis in the narrow sense—overdiagnosis of thyroid cancer—is presented in Box 1.

Using the word overdiagnosis in both the broad and the narrow sense is confusing: it may be time to stop. 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. Referring to that broad problem as ‘too much medicine’ 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, overutilization, over-medicalisation and disease mongering cannot be easily shoehorned into a narrow definition of overdiagnosis, but fit easily into the broad set of ‘too much medicine’. Debate over the narrow conception of overdiagnosis could then be restricted to a few central concerns (e.g. can it occur in symptomatic individuals? what is the conceptual boundary between an overdiagnosis and a false positive?).

The definition of “overdiagnosis” needs to be made more precise for specific purposes

Broader and narrower conceptions of overdiagnosis seem better suited to different purposes.

One such purpose is advocacy at a community and policy level. 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.

Other purposes, however, require greater precision. Epidemiologists, for example, need to measure overdiagnosis. 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. Overdiagnosis in ADHD, for example, may be measured, and thus implicitly defined, differently to overdiagnosis in screening mammography, or in cardiovascular disease, or in pre-diabetes. This suggests that there may never be a unified definition of overdiagnosis (narrowly understood) for epidemiological use. Instead, there may be precise but contestable methods for measuring overdiagnosis in particular conditions.

Other academic disciplines will have different purposes. Social scientists will use empirical methods to explain how ‘overdiagnosis’ and ‘too much medicine’ are understood, created, used and addressed by different stakeholders, informing interventions. Scholars in the philosophy of medicine will develop precise, formal, coherent definitions or typologies problem-solving the logical structure of overdiagnosis, and potentially informing epidemiological measurements.

Meanwhile, on the front line of medicine and public health, clinicians and policymakers need to change practice to maximise benefits and minimise harms to patients and populations. Different
groups—general practitioners, clinical specialists, or policymakers—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 ostensibly good 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 particular overdiagnosis problems. Professional organisations will have an important role in translating this research and setting new practice standards.4, 23-25

CONCLUSION

‘Overdiagnosis’ is a social and moral problem: it matters because it harms, or at the very least, wastes resources. But there is little agreement on how it should be defined. We have suggested that this is, in part, because different interested parties are working with different implicit understandings of the concept (Table 1). They are focused on different conditions and problems (e.g. mental health, cancer screening, pharmaceutical promotions), and have different purposes: unpacking the logical structure of overdiagnosis, explaining how it occurs, measuring it, advocating for political change, and/or developing practical tools to change clinical or policy practice.

Each of these purposes is important, but they occur at very different levels of generality. Advocates, for example, can employ the broadest ‘too much medicine’ conception, while epidemiological measurement requires precise rules, encoding multiple assumptions, specific to each condition studied.

We may never agree on a single definition of overdiagnosis. But we can—and we would argue, should—be more explicit in each instance: about breadth or precision, relevant conditions, assumptions regarding benefit and harm, and purpose. Clarifying these dimensions would serve our ultimate goal: to get a better grasp on the important problem of ‘too much medicine’.

KEY MESSAGES

- The term ‘overdiagnosis’ is used to refer to a range of concepts: there is no agreed definition of overdiagnosis.

- Any definition of overdiagnosis must incorporate the social and moral dimensions of the problem.

- There are broader and narrower senses of ‘overdiagnosis’, each relevant for particular purposes: there may never be a single unifying definition of overdiagnosis.

- We can and should commit to greater explicitness about crucial dimensions of the ‘too much medicine’ problem: the purpose of the discussion, whether we are speaking broadly or precisely, which conditions we are discussing and what assumptions have been made about benefit and harm.
REFERENCES


3. Heath I. Role of fear in overdiagnosis and overtreatment—an essay by Iona Heath. BMJ. 2014;349:g6123


5. Otte JA. Less is more medicine. [Cited 2014 Nov 25]; Available from: www.lessismoremedicine.com/


Table 1: Mapping concepts related to ‘overdiagnosis’, ‘too much medicine’ or ‘less is more medicine’

<table>
<thead>
<tr>
<th>RELATED CONCEPT</th>
<th>MEANING</th>
<th>OVERLAP WITH OTHER CONCEPTS</th>
<th>POSSIBLE DRIVERS</th>
<th>POSSIBLE REASONS FOR FAILURE TO BENEFIT</th>
<th>POSSIBLE EXAMPLES (NB assume ‘with no benefit’)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Overdiagnosis (in the narrow sense)</td>
<td>Some argue that this should be further limited to asymptomatic people only, others disagree.</td>
<td>Some argue that this should be further limited to asymptomatic people only, others disagree.</td>
<td>Overdiagnosis often occurs in an indeterminate zone between normal and abnormal tissue or function.</td>
<td>Disease was indolent or would have regressed</td>
<td>Non-beneficial diagnosis of non-progressive breast cancer through population mammographic screening</td>
</tr>
<tr>
<td></td>
<td>* Most people stipulate that the diagnosis should not be a false positive. Others argue that overdiagnosis often occurs in an indeterminate zone between normal and abnormal tissue or function.</td>
<td>Overdiagnosing interventions will often benefit some individuals while failing to benefit or harming others.</td>
<td>Early detection programs (screening)</td>
<td>Non-medical care would be more effective/beneficial</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>There is disagreement over the relationship between overdiagnosis and false positives.</td>
<td>Clinicians practicing defensively</td>
<td>Treatment produces no benefit or more harm than benefit (e.g. side effects)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>[Items in square brackets are contested]</td>
<td>An [asymptomatic] person is diagnosed with a condition that would not have shortened the person’s life or caused any symptoms.</td>
<td>Clinicians’ employers enforcing guidelines that encourage testing</td>
<td>Harmful labelling (e.g. causing anxiety, disrupting self-concept)</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Overdiagnosing interventions will often benefit some individuals while failing to benefit or harming others.</td>
<td></td>
<td>Intergenerational effects of harmful labelling (parental diagnosis leads to children being considered ‘at risk’)</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Overdiagnosis often leads to overtreatment and overutilization</td>
<td></td>
<td>‘At risk’ label promotes later overdiagnosis of disease or condition</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Expanded definitions and disease mongering likely to increase overdiagnosis.</td>
<td></td>
<td>‘Incidentalomas’ detected on MRIs conducted for other purposes</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Overdetection is likely to increase overdetection.</td>
<td></td>
<td>Detection of risk factors in routine general practice care or via commercial ‘health check’ services (e.g. blood pressure, cholesterol, blood glucose)</td>
<td></td>
</tr>
<tr>
<td>Overdetection</td>
<td>* controversial – some people see no distinction between overdiagnosis and overdetection</td>
<td>A health risk is detected in an [asymptomatic] person, probably via testing technology. That risk would not have shortened the person’s life or caused any symptoms.</td>
<td>Expansion of ‘at risk’ categories is likely to increase overdetection.</td>
<td>Person would not have experienced illness if risk factor remained undiscovered</td>
<td>‘Incidentalomas’ detected on MRIs conducted for other purposes</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Risk detection will often benefit some individuals while failing to benefit or harming others.</td>
<td>Overutilisation of testing technologies will drive overdetection</td>
<td>Harmful labelling (as above)</td>
<td>Detection of risk factors in routine general practice care or via commercial ‘health check’ services (e.g. blood pressure, cholesterol, blood glucose)</td>
</tr>
<tr>
<td></td>
<td>[Items in square brackets are contested]</td>
<td></td>
<td>Overdetection may lead to overdiagnosis, overtreatment and overutilisation</td>
<td>Intergenerational effects of harmful labelling (‘at risk’ parent leads to children being considered ‘at risk’)</td>
<td></td>
</tr>
<tr>
<td>Misdiagnosis</td>
<td>* Controversial: some argue that overdiagnosis should refer only to correct diagnosis; others</td>
<td>Incorrect diagnosis of a symptomatic person with a condition that they do not have (with corresponding</td>
<td>Encouraging well people to be tested</td>
<td>Incorrect diagnosis therefore treatment not directed to cause of illness</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Expansion of ‘at risk’ categories is likely to increase overdetection.</td>
<td>Purchase of expensive testing technologies, which services then must use to justify the expense</td>
<td></td>
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<tr>
<td></td>
<td>Incorrect diagnosis of a symptomatic person with a condition that they do not have (with corresponding</td>
<td>Overdetection is likely to increase overdetection.</td>
<td>Clinicians practicing defensively</td>
<td>Intergenerational effects of harmful labelling (‘at risk’ parent leads to children being considered ‘at risk’)</td>
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</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Overutilisation of testing technologies will drive overdetection</td>
<td>Clinicians’ employers enforcing guidelines that encourage testing.</td>
<td>‘At risk’ label promotes later overdiagnosis of disease or condition</td>
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<td></td>
<td></td>
<td>Results in over-estimates of the prevalence/incidence of overdiagnosed conditions and sometimes</td>
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<td></td>
<td></td>
<td>May be due to fear of missing most serious diagnosis, lack of diagnostic specificity.</td>
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</table>

NB: Non-be
argue that misdiagnosis should be included as a form of overdiagnosis.

<table>
<thead>
<tr>
<th>Overtreatment</th>
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<tbody>
<tr>
<td>* Likely consequence of (narrow) overdiagnosis</td>
</tr>
<tr>
<td>* Controversial: some wish to exclude because it can occur for reasons other than overdetection or overdiagnosis.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Overutilisation</th>
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<tr>
<td>* Controversial: some wish to exclude because it occurs for many reasons, not just 'overdiagnosis'</td>
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</table>

<table>
<thead>
<tr>
<th>Expanded definitions and/or disease mongering'</th>
</tr>
</thead>
<tbody>
<tr>
<td>Expansion of official disease or risk categories, or creating new conditions, or promoting more frequent diagnosis of recognised conditions, without benefit to patients or citizens</td>
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<table>
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<tr>
<th>Over-medicalisation</th>
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<tr>
<td>Altering the meaning or understanding of experiences, so that</td>
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| Human problems previously considered within a normal range, or as social problems, are re-interpreted as medical problems requiring medical treatment, without benefiting citizens | Disease mongering, overdetection, overdiagnosis, overtreatment and overutilisation. This category is extremely broad; overmedicalisation occurs well beyond 'overdiagnosis' narrowly defined | Overdiagnosis and overtreatment are likely to encourage thinking of human experiences as medical problems; the converse also seems likely. | Expanded definitions, disease mongering, overdetection, overdiagnosis, overtreatment and overutilisation | Childhood behaviour being seen as ADHD requiring drug treatment; reactive sadness in older people being seen as depression requiring anti-depressants; equating female breasts with being 'at risk' of breast cancer and needing regular mammography. |
Figure 1: Number of peer-reviewed papers indexed in PubMed with the word ‘overdiagnosis’ in title, abstract or keywords 1970-October 2014.
### Box 1: The example of overdiagnosis of thyroid cancer

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 11.6 cases/100,000 in 2009, 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. Prior to the advent of ultrasound, lesions were identified by clinical examination, usually when patients presented with symptoms. But now with ultrasound, lesions as small as 2mm in size can be identified and biopsied. If malignant cells are found, patients are offered thyroidectomy, the US rates of which have increased by 60% over the last 10 years. Despite the rise in diagnoses and treatment, the death rate from thyroid cancer has remained stable. This suggests, based on epidemiological and observational evidence, that the extra diagnoses and treatments are not reducing morbidity or mortality.

The challenge for addressing overdiagnosis in thyroid cancer is to understand the natural history of these very small tumours, which may grow too slowly to become symptomatic during the person’s life time. Are they one end of a spectrum of tumour behavior ranging from indolent to aggressive? Or are they a separate phenomenon, pseudo-disease rather than thyroid cancer? What are the relevant histopathological and genomic features that might answer these questions?