Bulletin of Medical Ethics, 1999, 147:13-17.
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The ethical implications of antenatal screening for Down's syndrome

Antenatal screening for Down's syndrome is growing fast and has important ethical dimensions. The medical press scarcely considers these but concentrates on how to make screening more sensitive and specific, cost-effective, and supportive to prospective parents. Emphases in bioethics on cost-benefit analysis, respect for the rational autonomous person, on freedom of choice, and non-maleficence, which now includes the notion of preventing a "wrongful life", can all be used to condone screening for Down's syndrome. These emphases can also divert attention from the prior questions: Why screen for Down's syndrome? And on what evidence and logic are the arguments which support screening based? Three researchers say how their discipline addresses these ethical questions.

Socratic enquiry

Chris Goodey, philosophy

What can philosophy contribute to research? One of philosophy's tasks is to verify whether we have a genuine claim to know something, another is to distinguish facts from values. Both are relevant to antenatal testing for Down's. Implicit in testing are claims to know, 1) that it prevents harm and suffering, 2) what it is like to have Down's (since without this knowledge the first claim could not be true), and 3) that Down's is a "mental" or learning disability. According to Socratic method, ignorance is more than just absence of knowledge: it may mean claiming to know something when we do not, in which case it will actually cause harm. Thus the method involves a discussion of values. Like the Cochrane collaboration it exposes falsehoods; rather than offering positive evidence of its own, it acts as a clearing-ground for research from other disciplines.

Can we truly claim to know that Down's involves suffering? Most physical conditions associated with Down's can be treated, as for other individuals; if we mean solely physical suffering, then, this cannot be used as cover for unjustified claims to know that people with Down's suffer in any other sense. If we claim that people with Down's suffer from other people's contempt, the very practice of antenatal testing would endorse the negative attitudes which constitute this harm. If suffering lies within the specifically mental life of people with Down's, people without Down's clearly cannot know this life by direct personal experience. If, finally, it is families who are said to suffer, doctors and counsellors do not usually have any empirical knowledge of living and being with those who have the condition. Do people with Down's experience the "mental" component of their disability as suffering, and does it cause their families to suffer? Until we genuinely know these things, how can prospective parents be counselled?

One answer might be that at least we know what a learning disability is. Psychology claims to know it as a scientific datum with exactly the same sureness that molecular biology knows the extra 21-chromosome. However, history shows that learning disability in any form recognizable to us only arrived on medical and psychological agendas in the eighteenth century. It was not a "discovery" like circulation of the blood; it came into our awareness only as part of a modern way of life that has become more difficult for certain kinds of people. Much of our knowledge about the mind has been short-lived and socially contingent. It is simply not of the same type as knowledge about the body; this implicit but false parallel confuses our picture of Down's and indeed most cognitive and behavioural genetics.

Realising that things were different in the past helps to highlight current social shifts: for example the disparity in the rate of live births for different conditions detected by the same initial test, or the finding that the more information individuals obtain, the less they take up services.(1) These tendencies counteract the original motive behind testing, which presumably was to eliminate or reduce the condition as far as possible (consent only became "informed" once prospective parents began asking questions).(2) Clearly different people have different values. But lay people's sophisticated distinctions about the nature of suffering cannot be just "values", by contrast with the "facts" implied in the three knowledge claims mentioned at the beginning.

Where does this leave bioethics? Philosophical ethics asks what is the good life. Bioethics, however, asks what is the quality or value of life.(3) These are not necessarily the same thing. First, we should consider how far the quality of a life in contemporary social conditions which make people with Down's stand out, matches up to a good life which might transcend any era. Stereo typically, they do badly at maths and well at interpersonal skills. Which of these contributes more to the good life? Secondly, bioethics tends to be applied only after the biotechnology is in place; philosophy, by contrast, tends to see ethics as a starting-point, as habits of thought which lie behind the actions of everyone. Doctors and biologists increasingly look to bioethics as an expertise alongside and separate from their own, but this could impede the exercise of their own ethical habits.

In summary, it is not clear that prenatal testing has or can have a consistent rationale. Research into it should proceed logically from clear evidence and ethically considered knowledge about the reality of a life with Down's, which we either cannot claim to possess or have not so far considered in any depth. The ground-clearing activities of philosophy may prepare us for such research.

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The good life?

Priscilla Alderson, sociology

Social and psychological research about antenatal screening concentrate mainly on large scale surveys of public and professional attitudes and behaviours, such as reported anxiety of screened women, (1) and their decision making.(2) Another approach is to hold interviews and small group discussions which consider themes through informal discussion. These sessions yield some quantitative data, but mainly explore how people understand their richly diverse lives and circumstances.

For example, researchers could ask prospective parents about the nature of any anxiety they might have. How does general anxiety about becoming a good enough parent differ from worry about possible fetal abnormality? Are they worried about uncertainty itself and potential loss of control, or about feeling forced to know and to choose, to become responsible when they feel powerless? Do they feel dread, or pity, for themselves or for the baby? Are they mainly afraid of the prospect of extra hospital visits and tests, ominous news, or the possibility of an abortion? And are these partly iatrogenic fears mis-attributed by practitioners to fear of the fetus? Or are women afraid of giving birth to a monstrous child that will make them feel a failure, depressed and isolated, and unable to have a career or care for their whole family? Do they believe that a child with Down's might never speak or walk, or have any hope of a good life?

As phrased here, the questions sound harsh, intrusive and loaded. It is vital for researchers to be informal and respectful, (3) to ask open-ended and follow-up questions including positive ones, and to encourage people to feel free to say as much or as little as they choose. Then, many people will give long replies and introduce sensitive topics themselves.

Another important topic of enquiry is the source of knowledge about Down's. Some paediatric textbooks mention Down's only in reference to antenatal screening,(4) or simply as a set of physical anomalies and "severe mental retardation" that are "not treatable".(5) Professional case reports usually emphasise difficulty, inability and need, in order to obtain more resources and services for the child. Many parents and "self-help" organisations similarly (over)emphasise the frequency and severity of the problems, believing this benefits affected people by increasing professional support and public funding. All such reports serve specific purposes rather than aiming to state general truths.

Research is often skewed by negative emphases. Questions like "how does your problem affect your life?" can exaggerate the effect of the syndrome by excluding other strong influences like income, opportunities, friends, or attendance at a segregated or a mainstream school. Favoured terms like "patient, disease, suffering, coping" turn Down's into an illness. If, rarely, people with Down's are interviewed, researchers may confuse them with technical terms or talk down to them, ask closed questions, or interrupt detailed replies as irrelevant, or be too authoritative or pitying - styles which discourage any interviewee.

I interviewed five people with Down's syndrome among 40 pilot interviews with adults who have conditions which are tested for antenatally.(6) Open questions enquired about their family and friends, education and employment, interests and difficulties, hopes and aims, their views on being or becoming a partner and a parent, and on antenatal screening. The five had wide-ranging employment records: working in an office and a hospital kitchen, teaching doctors, teaching disabled people about independent living, lecturing and also acting in theatres in England and abroad, and co-writing plays about disability, stigma and genetics. Their detailed interviews were often dramatic and humorous, for example:

Interviewer: And your mum had a row?

Martha: It wasn't a row, it was a fight. She had to fight for my rights you see. [The head teacher had said] "What is this mongol person doing in my school?" I was there for five years, five years, and she said that!

Int: So what did your mum say?

Martha: Well, what did she say to her? My mum went to the high court straight away. Of course. The high court of justice! [The fight was to be able to stay in a mainstream school] Because the special school didn't do me any good at all basically. [Martha enjoys travelling but]
the trouble is getting the money together, we're on benefits, you can't afford it on benefits.

Int: No, but you do manage to live quite a - good life?

Martha: I do, yes.

I asked the above question because, like the other people with Down's, she described parts of her life with such enthusiasm.

Asked what she might say to a woman who had been told that her baby would have Down's syndrome, Martha replied, "I'd tell her to do everything, feed it, look after it, give it lots of love". Peter and Philip had eagerly described their current play about "Mongol boy" who was rejected by his family. They looked very sad at this question. Peter said, "That is actually what we are doing in this play, for heaven's sake. (A pause) I'm speechless. The productions we have done are just fantastic. (They look as if their reply is too profound to put into words.) Carry on [to the next question]. Pass."

The methods and findings of these conversational-style interviews deal with real-life ethics of respect and justice. They challenge the assumptions that life with Down's syndrome is inevitably limited and requires expensive support. The 40 interviewees said that discrimination is their worst problem in preventing them from contributing to society as they can and want to do. Ironically, antenatal screening officially endorses this costly discrimination which forces people into the segregated dependency that screening is supposed to reduce. Much more research into the lives which people with Down's do and could have is required before screening policies can be evidence based.

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Calculating the costs and the benefits

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Health economists are interested in tackling real-world dilemmas involving choice. Choice refers to the decisions we take implicitly and explicitly - as to how to spend our resources (time, labour, capital, and yes, money) to obtain the things we want knowing that more often than not we do not have enough resources to do all the things we would like (resources are scarce) [1].

For Down's syndrome screening there are many potential choices: how should a screening programme be organised; what tests are best [2]; and why screen at all. All these decisions involve costs and benefits. Investing in a national screening programme means not enjoying the benefits of some alternative way of spending health care pounds (or, if taxes are increased to cover the cost of the screening, of not enjoying the benefits of private spending). Every choice therefore involves a sacrifice. The central question is what level (and distribution) of benefits are we prepared to forgo in order to enjoy the benefits arising from using available resources in one way rather than another?

The guiding economic principle is therefore to maximise benefits and minimise costs (ie forgone benefits) that is, to be efficient. This also represents an ethical basis for choice; to be inefficient (to make a choice which involves more costs than benefits) is not just a waste of resources (usefully represented in monetary units) but, much more importantly, it is a waste of what those resources could produce lives saved, disability averted etc [3]. To ignore the resource implications of a decision involving a health care intervention is to risk increasing the avoidable burden and distress of ill health.

For a Down's syndrome screening programme, exactly which costs and benefits are to be weighed, measured and valued will depend on the choice or decision at hand and the information available (particularly the quality of information from clinical trials). Evaluative techniques, such as cost benefit or cost effectiveness analysis, have been developed specifically to address various classes of decision [4]. Should, for example, screening for Down's syndrome be done at all? Theoretically, a cost benefit analysis, in which all the costs (not just the financial costs to the health service, but all forgone benefits to society as a whole) and all the benefits are identified, measured, valued in monetary terms and then compared, could help answer this question. (The argument that it is impossible to value such things as a human life or the feelings of reassurance a negative screen result [cf 5] may provide, is contradicted daily by actual decisions taken by patients, health service managers and clinicians.) Other decisions may focus on the best (i.e. cheapest) way to screen to obtain an acceptable and adequate outcome to the screening process. Exactly what is acceptable and adequate is of course open to debate.

But why evaluate a Down's syndrome screening programme in the first place? The feeling that there does exist a decision to be taken (and hence an evaluation to be done) is a societal value judgement, and economic evaluations will reflect this. However, there can be some societal confusion or disagreement regarding the objectives of screening programmes for Down's which can complicate an economic evaluation. Commonly, the objectives of any screening programme are not simply to identify a condition, but to do something about it [cf 6]. But what parents want do about a fetus with Down's may not be what someone with the syndrome would want. Whose views about objectives should we take? If the real objectives of screening are to give parents the opportunity to make an informed choice about the continuation or termination of a pregnancy, then the viewpoint is the parents'. However, given the funding and structure of the NHS, exercising their choice is not just a private matter, but has implications for others. How to deal with such distributional issues (who enjoys the benefits, who bears the costs) in the essentially utilitarian framework of economic evaluation can be unclear, however.

Of course individuals' and society's values change and can be influenced, and the prior need for an economic evaluation, and further, the type of evaluation and the valuation of costs and benefits, will need to reflect such changes. For example, one element of the broad costs of having Down's syndrome might be the lost or lower than average earnings of people with Down's syndrome. Taking these into account in an economic evaluation is not intended to endorse any potential discrimination by employers towards those with Down's syndrome, but to reflect a potential societal view. What we see in the economic evaluation mirror may not be what we would like to see.

For pragmatic reasons, all economic evaluations make assumptions and adopt a particular viewpoint with regard to, say, whose values prevail in an evaluation, and how accurate and detailed the measurement and valuation of costs and benefits need to be. Good evaluations make this clear and explore how the results of an evaluation would change if alternative views, values or assumptions were adopted. Ultimately, it is not up to the economic researcher to make the decision, but, in the context in which the decision exists (for an individual, the NHS, society), to set out the costs and benefits implied by the choices being evaluated, for others to draw their own conclusions.

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