

# Down Syndrome Disappearing

**Cheap diagnostic tests are on the way, but is Down syndrome the tip of the iceberg?**

The Margaret River burrowing crayfish, the orange-bellied parrot, the mountain mistfrog and the bare-rumped sheath-tail bat are among 36 species listed as “critically endangered” by the Australian government. If Dr Brian Skotko of Children’s Hospital Boston has his way, Down syndrome children should be added to the list. In a recent issue of the journal *Archives of Diseases in Childhood* he points out that the number of Down syndrome children born is declining year by year, at least in developed nations.

Because women are waiting longer before they have children and older women have a higher chance of having a Down syndrome child, the birth incidence should climb. In fact, it has actually decreased. Current studies show that 92% of women who receive a definitive prenatal diagnosis of Down syndrome choose to terminate their pregnancies. As a consequence, Down syndrome children are vanishing.

For instance, in the US there should have been a 34% increase in Down syndrome births without prenatal testing, largely because of older mothers. Instead there has been a 15% decrease – or a 49% gap. In the UK there is a 48% gap. No doubt the statistics are similar in Australia.

And Skotko says that there will be even fewer of these children in the future because a non-invasive blood test will soon be available that will provide a definitive diagnosis in the first trimester. Two companies in the US have announced that they will market such a test later this year.

Because it is uncomplicated, nearly all pregnant women will use it. Because it gives



an early diagnosis, a woman will be able to terminate in the first trimester, so there will be less risk to her health. And because it is non-invasive, it poses no risk to the foetus before the diagnosis.

Current tests entail a small risk of miscarriage, which means that sometimes a normal child dies in the course of testing whether it has Down syndrome. In fact, a UK study last year claimed that for every 660 Down syndrome foetuses that are detected and terminated each year, 400 normal children perish as well.

There will also be a financial incentive. Because the new tests are relatively cheap, health insurance plans will probably cover them, making their uptake even more widespread. Down syndrome children often have complicated health problems, and the insurers could see this as a cheaper option than paying for the medical care for the rest of their lives.

A plethora of knotty ethical problems are contained in this situation, even for those who accept a woman’s right to choose an abortion. The obvious one is: what is really the big deal?

“Parents who have children with Down syndrome have already found much richness in life with an extra chromosome,” writes Dr Skotko. Admittedly, Down syndrome children have impaired intellectual skills (although some have

made it through university) but they are often extraordinarily loving, cheerful and affectionate. Parents often remark that they have special gifts that other children lack.

What worries Skotko is that most doctors know very little about the positive side of life with Down syndrome and misrepresent the burden of raising a Down syndrome child. They often give inaccurate, incomplete and sometimes insensitive advice to women. He feels that this effectively makes it impossible for women to give informed consent.

The new non-invasive tests will make the diagnosis of hundreds of genetic conditions possible before birth. Down syndrome is just the first of them. So, in the not-too-distant future, we are sure to debate what kinds of genetic variability we will tolerate in our society.

An opinion piece in the leading journal *Nature* recently argued that “genetic diversity, from within or among groups, should be embraced and celebrated as one of humanity’s chief assets”. However, having drawn the line at Down syndrome, what other conditions will women be encouraged to terminate?

Prenatal diagnosis is a mixed blessing: it gives knowledge but not necessarily the ethical insight to use it wisely.

Michael Cook is editor of the bioethics newsletter, BioEdge.