LEADER 8 April 2015

We must take baby steps into newborn genome sequencing

Genome sequencing at birth could be the start of a medical revolution that could save lives and slash healthcare costs. But we need to proceed with caution

Could baby genome sequencing become the norm in maternity wards?
(Image: ERproductions Ltd/Blend Images/Corbis)

SOME time in the next few days a new era in healthcare will be born. Doctors in Boston will sequence the whole genome of a newborn, the first of 240 babies to be fully sequenced. Afterwards, the infants will be monitored for at least five years (see ""Baby genes to be mapped at birth in medical first"")

The experiment anticipates a time in the near future when whole genome sequencing is a standard part of neonatal care. The potential benefits are huge: parents and doctors will be forewarned about a range of genetic diseases over and above the 30 or so that are picked up by today's screens. Sequencing could save lives and cut healthcare costs. But the wider implications are unknown; hence the experiment.

Neonatal sequencing certainly raises some difficult questions. Although the Boston trial will only look for serious childhood illnesses, neonatal sequencing can also uncover information about more uncertain and long-term risks, such as genes that increase susceptibility for diseases that won't appear until later in life, if at all.

What are parents expected to do with such information? Routine neonatal sequencing risks overwhelming them – and their doctors – with information that is both hard to interpret and difficult to act upon.

“Neonatal sequencing risks overwhelming parents with information that is hard to interpret or act on”

There are other issues too. The Boston team are motivated, in part, by the fact that private companies are champing at the bit to begin commercial newborn sequencing. It is easy to see how this could become a lucrative business. Companies already make a living out of banking umbilical cord blood from newborns as a source of stem cells that may prove useful if the child develops a blood disease or immune disorder later in life. According to the American Academy of Pediatrics, the possibility of actually needing the blood is about 1 in 200,000, but that does not stop many parents from banking some, just in case.

You might argue that selling newborn sequencing is little different from existing personalised DNA services offered by companies such as 23andMe. But there is a clear distinction: newborns cannot give informed consent. Parents are rightly empowered to make medical decisions for their children, but sequencing is not like agreeing to a course of treatment. The child will eventually become an adult who may not want to know their genetic destiny.

The complexities will only deepen as it becomes easier to sequence the genomes of children before they are born. This has already been proved possible in early pregnancy simply by taking a blood sample from the mother. Once it becomes routine, the choices could be agonising: the discovery of a genetic flaw may prompt parents to abort fetuses that would otherwise live largely healthy and productive lives.

And sequencing is just the start of a genetic revolution in prenatal and neonatal care. Advances in biotechnology also raise the possibility of fixing faulty genes in human embryos. Reports have emerged that groups in the US and China are already experimenting with this, prompting calls for a moratorium on such research.

That is an overreaction. Like sequencing, gene editing has enormous potential to alleviate human suffering, and looks unstoppable. But we owe it to ourselves and future generations to proceed with caution. The Boston trial is a vital first step on the road to what could truly be a brave new world.

This article appeared in print under the headline “Brave new world beckons”

A shorter version of this article was published in *New Scientist* magazine on 5 May 2018

Magazine issue 3016, published 11 April 2015