Genomic screening for newborns

Soon after birth, every newborn baby in Australia is offered a test to identify rare but serious health conditions. A nurse usually takes a blood sample by pricking the baby's heel, and a few drops of blood are collected on a paper card, hence the common name of 'heel prick test'. The test is optional, and parents need to agree to the test in writing, but it is unusual for someone to decline (99.9% of children are tested). This is because, although the conditions tested for are rare, if the baby has one of these conditions and it is not discovered and treated early on, the baby could become permanently disabled or even die.

Screening for these conditions is required because the baby may have one of them and still appear to be healthy. By the time symptoms show, irreversible damage may already have occurred. Screening through the heel prick test enables prompt diagnosis and treatment to reduce the negative impact of the condition. Most babies assessed will receive a normal result.

Newborn bloodspot screening is an extremely successful public health program internationally to prevent avoidable disability[1]. At the moment each state runs its own program, but a national policy to support newborn bloodspot screening programs is currently being developed in Australia.

At the moment only a few dozen diseases are included in the screen. They include genetic, metabolic and hormonal disorders that don't show symptoms at birth but can be dangerous if left untreated. They include the following:

Phenylketonuria (PKU): an inherited disease in which the body cannot metabolize a protein called phenylalanine. Without treatment, it can cause intellectual disability. Treatment involves dietary changes.

Hypothyroidism: this is a condition where the baby is born with too little of the thyroid hormone. Untreated low thyroid hormone levels can lead to problems with mental development and growth. It is treated with thyroid hormone medication.

[1] Therrell BL, Padilla CD, Loeber JG, Kneisser I, Saadallah A, Borrajo GJ, et al. Current status of newborn screening worldwide: 2015. Semin Perinatol. 2015;39:171–87. https://doi.org/10.1053/j.semperi.2015.03.002

Congenital adrenal hyperplasia (CAH): this is an inherited disease of the adrenal glands. Babies born with this condition cannot make enough of the hormone cortisol, which helps control energy, blood sugar levels, blood pressure, and how the body responds to the stress of injury or illness. It is treated with medication.

Cystic fibrosis (CF): this is an inherited disease that causes serious lung and digestive problems. Early diagnosis and treatment improve the outlook for babies with CF. Treatment is complex and needs to be assessed for each individual.

Galactosaemia: This is an inherited disorder in which the baby is unable to metabolise galactose, a sugar in milk. Without treatment (avoidance of milk), it can be life threatening.

Homocystinuria: This inherited disorder causes intellectual disability, bone disease, and blood clots. It's caused by a deficiency of an enzyme needed to digest an amino acid called methionine. Treatment consists of a special diet and supplements.

You can see that these diseases are serious conditions which have an impact early in life and are treatable. The screening program in its current form should be welcomed as an important contribution to the welfare of young children. But changes are afoot.

Since the mapping of the human genome in 2003, an enormous amount of research has explored the genetic basis of human disease. Technology now exists to reveal the complete genetic profile of a newborn from the heelprick test. Australians have been notified that routine genomic screening for newborns may not be far away. If genomic screening is rolled out, it's likely that, in the short term, tests will only look for genes that cause diseases that can be treated immediately, just as current screening does. This is ethically uncontroversial. But we all have around 22,000 genes, and currently thousands of conditions for which we can test.

Five major studies running across Australia are testing out applying genomic screening to newborns. As we widen the number of diseases we screen for, we start to move away from those that have an impact in childhood or are treatable. We also move away from most babies getting a normal result. A research study in the USA found single-gene disease risks in 11% of newborns and 88% of newborns had a harmful recessive gene that could be passed on to their children.[2]

The kind of information you can receive in a genomic test result can vary. For example, you can get information about:

a) Diseases for which there is prevention and/or treatment available
b) Diseases for which no prevention or treatment is available
c) Diseases that would not occur until adulthood
d) Gene changes which will not affect the individual but may be passed onto one's children
e) Gene changes which mean you have an increased risk of disease but will not definitely get the disease

[2] https://www.genomes2people.org/research/babyseq/

How much do you want to know?

There is nothing in the Bible that suggests that it is wrong to use technology to give us more information about our health. God gave us minds that are capable of exploring his creation. Technology itself is neither good nor evil. It is how we use it that makes the difference, and we have been appointed as stewards to take responsibility for the creation (Genesis 1:26; Psalm 8).

However, as our genetic knowledge increases, so does our need for both wisdom and humility. Wisdom in our decision-making. Humility to realise the limits of our understanding.

The bible teaches that all human beings have been individually created in the image of God, and as such have intrinsic worth, which is not dependent on our genetic makeup (Genesis 1:26, Genesis 9:6). The use of genetic screening to decide which human beings should not be born is not consistent with Christian belief. However, this is not the situation for the heelprick test.

Genetic testing of children is done with the consent of their parents or guardian. Testing children where the result could be of potential benefit them in childhood is ethically permissible, and consistent with a parent's responsibility to care for their child. Opting to receive results (a) some would say is obligatory, as this knowledge can benefit both the child and society. Whether parents want to know about results (b) or (e) is something they should carefully consider, as this knowledge may cause distress without providing health benefits.

A minor cannot give informed consent, so it can be argued that it is wrong for them to be tested for disorders that will neither affect his or her health until adulthood or be acted on before adulthood (results © and (d)). The reason for this is that the parents will not use this information – it will be used by the child in adulthood. Therefore, it is suggested that the child should be given the opportunity to decide whether or not they want to have this information, i.e., they should be given the opportunity to decide whether these conditions are tested for when they can give informed consent themselves. This is information that you cannot 'un-know', once you have been told. While some people find this kind of knowledge empowering, others regard knowledge about one's genetic risk as burdensome and a source of anxiety.

It's tempting to think genetics will give us certainty about what our future health will be. But genetics is not absolute. Sometimes genetic tests raise more questions than they answer. And there is no test for a 'healthy child'. As newborn screening evolves, we must be vigilant in asking our healthcare providers to be transparent in what is being tested, and why.

Megan Best

August 2023