Researcher looks at public perceptions around newborn testing

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While 94 per cent of Canadians surveyed said they would participate in public health programs that screen newborns for a specific number of genetic conditions, only 80 per cent said they would be willing to participate in screening that would sequence their newborns' genomes. Most newborns in North America have a "heel prick test" in their first day or two of life in which a tiny amount of blood is taken from their heels and tested for about five to 54 conditions, depending on the state or province. Some conditions commonly tested for include cystic fibrosis, the enzyme deficiency phenylketonuria or PKU, and hypothyroidism, a thyroid hormone deficiency.

The idea is to identify and treat diseases before irreversible damage has occurred to prevent or reduce developmental, neurological or other health issues. These public health measures are not mandatory, but parental consent is typically implied because newborn screening is considered the standard of care.

Dr. Yvonne Bombard, a genomics and health services researcher in the Li Ka Shing Knowledge Institute of St. Michael's Hospital and an assistant professor at the University of Toronto's Institute of Health Policy, Management and Evaluation, said technology has evolved to the point where we could now scan a baby's entire DNA sequence – not just conditions that can be treated in infancy, but such things as whether the baby carries the BRCA1 or BRCA2 genes associated with breast and ovarian cancer or has a genetic predisposition to Alzheimer's.

"Where do we draw the line on what we screen for?" she asked. "Public opinion should matter when we make these decisions. Newborn screening programs require almost 100 per cent participation to be effective and if we lose people's trust, people may opt out."

Dr. Bombard explores those issues in a paper published in the *European Journal of Human Genetics*.

She and other researchers conducted an online public survey that found 94 per cent of respondents would be willing to participate in newborn screening using existing technologies the screen for specific genetic conditions that can be treated in childhood versus 80 per cent who would participate in screening that would sequence their newborns' genomes for any and all forms of disease. Much smaller numbers thought it was a parent's responsibility to participate in newborn screening programs (48 per cent for existing technology and 30 per cent for whole genome sequencing).

Dr. Bombard said some of the issues around sequencing newborns' genomes include freedom of choice and the danger of over diagnosis.

"Does whole genome sequencing take away a child's choice to remain unaware of his or her future health risks?" she asked. "It could also generate a lot of genetic information that we don't understand. For example, there are genetic variants in the cystic fibrosis gene that are not known to be associated with cystic fibrosis. What do we do about that information? Monitor the child? Overtreat the child? Are these added harms that we are introducing into this screening program that may challenge its authority to continue to identify and treat the affected infants?"

"We can filter out what we look at in a genome. That is, we can type the whole book but don't have to read every sentence. But as a public health

program we don't have the capacity to give parents a menu of options and ask them what type of health information they want to select to learn about in their children, so it's important that the public understand this new technology and its potential limitations so we can draw an appropriate line on what we screen for in these newborns."

"It's essential that we respond to technological developments with a full understanding of the needs of newborns, children and their families," said Fiona A. Miller, the study's principal investigator and associate professor in U of T's Institute of Health Policy, Management and Evaluation , noting that the study highlights the need for newborn screening policies that are informed by a range of stakeholders, not special interest groups exclusively.

"When we decide that it makes sense to screen for a particular condition in a newborn, we need to ensure that the whole system works to maximize benefits and minimize harms. As well, we need to invest in the care that can be of greatest value to children and their families."

Provided by St. Michael's Hospital

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