Parents who pass on genetic conditions are likely to opt for prenatal testing in future

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Having a child with a severe genetic condition will heavily impact on a parents' decision to have future prenatal testing, according to a new study co-conducted by Plymouth University.

Professor Heather Skirton, Professor in Health Genetics, was part of an academic team that interviewed 17 parents who were carriers of a serious autosomal recessive condition – spinal muscular atrophy, cystic

fibrosis or thalassemia – and had a child, living or deceased, with the condition.

The conditions are such that both parents must carry a harmful variant in one of their copies of the relevant gene and an affected child inherits the varied copy from both parents, and has no normal copy. All humans carry some variants for recessive conditions, but if they also have a normal copy of the gene they are usually healthy. Most parents are therefore not aware they are carriers. Prior to the affected child's diagnosis, the parents entered the pregnancy with no belief that there would be any cause for concern.

But the study showed that their child's diagnosis had impacted or would heavily impact on their decision to have non-invasive prenatal testing in future, as they described feelings of reproductive vulnerability and consequent realisation of risks to future children.

The study began when Professor Skirton's team began to examine a new non-invasive prenatal testing; a blood test which could be carried out nine weeks into the pregnancy.

Having the non-invasive procedure would mean that parents could find out about the condition earlier without threatening the unborn child. It would also mean that they could decide whether to terminate the pregnancy at an earlier stage.

Professor Skirton explains that the findings are important for midwives to consider, saying that when parents consider prenatal testing, they cannot control the vulnerability brought about by previous experience.

"The study showed that the impact of losing a child to one of these conditions, or knowing their child is living with it, is huge," she said. "The parents can't protect themselves from that previous experience and

the fear that goes with it, so midwives should be sensitive to parents' reproductive vulnerability and ensure they are supported to consider the option of non-invasive prenatal testing if appropriate.

"The parents saw the use of non-invasive prenatal testing as a way to mitigate threats to foetal health, while allowing them to prepare for an affected child or consider termination of pregnancy. By being carried out at nine weeks, the non-invasive prenatal test means that they could delay disclosing the pregnancy, if desired. The study showed that they also viewed it as a way to reduce threats to an unborn child.

"The overall findings were very interesting, as the decision whether or not to have the test would have been challenging for the participants, given that the resultant question – whether or not to terminate the pregnancy – would also be hard to face."
The full study, entitled Impact of fetal or child loss on parents' perceptions of non-invasive prenatal diagnosis for autosomal recessive conditions, is available to view in the journal Midwifery.

More information: Laura Pisnoli et al. Impact of fetal or child loss on parents' perceptions of non-invasive prenatal diagnosis for autosomal recessive conditions, *Midwifery* (2016). DOI: 10.1016/j.midw.2015.12.009

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