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Pre-natal genetic testing for deafness – do families want it?

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Introduction
Advances in genetic testing, and in the understanding of the genetic causes of deafness, mean that prenatal diagnosis for deafness has become a possibility for some families and is likely to become more widely available in the future. Although there is some anecdotal evidence that families might wish to use prenatal testing to avoid the birth of a child with deafness, there is limited objective information about families’ views. A study was carried out asking hearing parents of deaf children about their views on prenatal testing in general, and on prenatal testing specifically for deafness. The families were also asked for their views on a new method of prenatal diagnosis, called preimplantation genetic diagnosis (PGD).

Until recently there were two methods of prenatal testing available; amniocentesis and chorionic villus sampling (CVS). Amniocentesis cannot be carried out until around 16 weeks of pregnancy. CVS can be done sooner at around 11 weeks, but carries a greater risk of causing miscarriage.

Prenatal diagnosis with amniocentesis has been available for over thirty years, and CVS has been in use for about twenty years. The number of conditions which could be detected using these methods was limited at first. Initially prenatal testing was used to detect conditions caused by a chromosomal abnormality, for example Down Syndrome. As technology improved, more conditions could be
identified, but the real expansion in testing has come in the last decade, since we began to understand the underlying genetic causes of different conditions, and to identify the gene changes responsible.

Both amniocentesis and CVS require a pregnancy to have started before testing can take place. Parents who find their baby is affected have to decide whether to continue or to terminate the pregnancy.

More recently a different method of prenatal diagnosis has been developed. Preimplantation genetic diagnosis (PGD) combines in-vitro fertilisation (IVF) techniques with genetic diagnosis. Embryos are created outside the womb, and tested for the genetic condition before they are implanted. In this way parents can choose only to have unaffected embryos implanted, avoiding the need for invasive testing on the pregnancy later on. This method of diagnosis is theoretically possible for some families, but it is not yet widely available and success rates are low.

A study was carried out to gauge the level of interest in PGD, and to determine the views of hearing parents of deaf children about prenatal testing in general, and on prenatal testing specifically for deafness.

**The study**

Thirty-two families were approached to take part in the study. They were identified from the records of patients who had attended a department of clinical genetics for genetic counselling. The criteria for inclusion were that the parents should be hearing; that they should have at least one deaf child, and that they should have undergone genetic testing for a common genetic cause of isolated non-syndromic deafness. The families were sent an initial information sheet, and an opt-out form to return if they did not wish to be contacted further. Families who did not return the opt-out form received fuller information about the different methods of prenatal diagnosis, and a questionnaire. One questionnaire was sent for
each parent so that both had a chance to give their view. Seventeen completed questionnaires were returned, representing a 30% response rate.

The questionnaire

The questionnaire consisted of a number of questions requiring a Yes/No answer, and respondents were given space to explain why they answered as they did. A series of questions was asked about attitudes to prenatal testing specifically for deafness, and also for other conditions. The same questions were then asked in relation to preimplantation genetic diagnosis. Finally parents were asked which of a number of options they would choose if they were planning another pregnancy.

Attitudes to prenatal testing

Parents were asked if they would consider prenatal diagnosis for deafness, with termination of an affected pregnancy. They were also asked whether they thought this should be available to others, even if they would not choose it for themselves. Most respondents (15/17) felt this was an unacceptable option for them, and 13/17 said it should not be available to anyone. Comments from these parents included

“Deafness is not a reason to abort the pregnancy. It is not life-threatening”

“Deafness in a baby is certainly not a reason for termination. A deaf child has as much right to life as a hearing one and can have an equally fulfilling life”

“Having a deaf child does put extra pressures on a family but that would never be a justifiable reason for termination.”

One parent said she would consider prenatal diagnosis without termination of pregnancy in order to be prepared for her child’s needs, should it be deaf.
Of the two respondents, both female, who said they would consider prenatal diagnosis for themselves, with termination of an affected pregnancy, one commented

_I would not want the struggles in our lives, the heartache we have experienced, and the worry._

Those who felt prenatal diagnosis should be available made comments suggesting that it should be an individual choice and recognised that some people might feel they could not cope with a deaf child

_“If a person felt that having a deaf child would be an insurmountable problem for them, I would not wish to prevent them from having prenatal diagnosis”_

Most parents (16/17) felt that prenatal diagnosis should be available for some conditions. Parents were asked to specify what sort of conditions they thought testing should be available for. The replies indicated that these parents felt it should only be available for “serious” conditions.

_“Severe brain damage or very severe physical disability”_  
_“For babies who would die young or be in constant pain…needing medical support 24 hours a day”_

Some parents took a broader view, recognising that _“the answer is not black and white. It depends on the severity of the disability and then it’s a very personal choice and depends on many circumstances”_

Fourteen parents said that if they were at risk of having a baby affected with a serious genetic condition, they would consider prenatal testing with termination of an affected pregnancy. Parents who made further comments were generally concerned with the quality of life of the affected child, and also the effect on the rest of the family.

_“To risk having a child with severe disabilities would be of great concern as it would affect the time I need to give to my current family”_  
_“You must consider the quality of life the baby would have.”_
Attitudes to preimplantation genetic diagnosis

Parents were generally much more positive about this option, with 11/17 saying that they would consider it for themselves, and 15/17 saying they felt it should be available for others. PGD was clearly regarded as ethically more acceptable than prenatal diagnosis and termination of pregnancy.

“The moral dilemma of whether to terminate an affected pregnancy is much greater and more complex than the decision whether to implant an embryo”

“This appears a good solution – ethically acceptable”

Another parent (who would not choose prenatal diagnosis) stated: “If I could eliminate the risk (of having another deaf child) I would”

Without exception, parents felt that PGD should be available for other conditions. A wider range of conditions were seen as being suitable for PGD than for prenatal diagnosis, although generally parents gave examples of conditions which would be regarded as serious. When asked if they would choose PGD themselves to avoid the birth of a child with one of these conditions, 16/17 parents felt this was an acceptable option, with the one parent who disagreed stating her unwillingness to undergo IVF to achieve a pregnancy as her reason.

Options for managing a future pregnancy

Parents were asked to give an order of preference to different options if they were planning a further pregnancy assuming all options were available to them. The choices were:- no testing in pregnancy; prenatal testing and termination of an affected pregnancy; or preimplantation genetic diagnosis. PGD was the most frequent first choice, followed by no testing in pregnancy.

Impact of deafness on the family
To try to determine whether parents’ decisions about prenatal diagnosis were affected by their experience of having a deaf child, they were asked whether they felt deafness had had an impact on the family. Their answers were categorised as being “positive” or “negative”. Six parents felt that there was a significant impact which was generally negative

“incredible strain on the whole family.”
“increase in general stress and anxiety. Less time for other children”
“sadly I feel it has had a very negative effect”

Three parents felt the impact had been positive

“It has totally changed the way I think about deafness. It has greatly enriched our lives”
“Our deaf child is so brilliant. We as a family are closer and support each other more”

Three parents felt that the impact on the family had been both positive and negative.

Summary

In this small study of a selected group of hearing parents of deaf children, there was low interest in prenatal diagnosis for deafness, with termination of an affected pregnancy. Although a significant proportion of parents felt that having a deaf child had affected their lives in a negative way, they did not feel that prenatal diagnosis and termination of pregnancy is an acceptable means of avoiding the birth of further affected children. Some parents felt the choice should be available for others. There was considerable interest in preventing the birth of further deaf children without undergoing termination of pregnancy. Although it is difficult to draw any firm conclusions from this small sample it does provide some indication of families views on the issues of prenatal testing for deafness.