Statutory Approvals Committee – minutes

Centre 0102 (Guy's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Non Syndromic Congenital Deafness (DFNB29), OMIM #614035

Thursday, 28 September 2017

Church House Westminster, Dean's Yard, Westminster SW1P 3NZ

Committee members	Margaret Gilmore (Chair) Anne Lampe Ruth Wilde Bobbie Farsides	
Members of the Executive	Dee Knoyle	Committee Secretary
External adviser	Dr Alan Fryer	
Legal Adviser	Tom Rider	Fieldfisher LLP
Observers		

Declarations of interest

- Members of the committee declared that they had no conflicts of interest in relation to this item.

The committee had before it:

- 8th edition of the HFEA Code of Practice
- Standard licensing and approvals pack for committee members.

The following papers were considered by the committee:

- Executive Summary
- PGD Application Form
- Redacted peer review
- Genetic Alliance opinion
- One additional paper submitted with the application – Bashir *et al.*

1. Consideration of application

1.1. The committee welcomed the advice of its Specialist Adviser, Dr Alan Fryer, who confirmed that the condition was as described in the papers with the exception of a typing error, stating that the gene reference should read DFNB29.

1.2. The committee noted that the description in the application for Non Syndromic Congenital Deafness (DFNB29), OMIM #614035 is consistent with the Peer Review.

1.3. The committee noted that Genetic Alliance supported the application and provided some information gathered in relation to the types of symptoms seen in autosomal recessive non syndromic deafness and the impact of autosomal recessive conditions.

1.4. The committee had regard to its decision tree. The committee noted that the centre is licensed to carry out PGD. The committee was also satisfied that the centre has experience of carrying out PGD and that generic patient information about its PGD programme and associated consent forms had previously been received by the HFEA.

1.5. The committee noted that the condition being applied for is not on the list of approved PGD conditions.

1.6. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of Schedule 2 of the Act, i.e. 'where there is a particular risk that the embryo may have any gene, chromosome or mitochondrion abnormality, establishing whether it has that abnormality or any other gene, chromosome or mitochondrion abnormality'.

1.7. The committee noted that the condition is inherited in an autosomal recessive pattern and there is a 25% chance of an embryo being affected with the condition if each parent has a relevant mutation.

1.8. DFNB29 is a rare condition causing profound to severe hearing loss affecting all hearing frequencies. In the earliest reports it was not thought to cause a progressive hearing impairment but a recent report of a family from Newfoundland demonstrated progressive loss could occur with some mutations. Whilst variation in severity can occur, most affected children have severe to profound loss. Onset is in early infancy and bilateral sensorineural deafness results. The hearing loss is of pre-lingual onset meaning that children have hearing loss before they learn to speak. The condition is non-syndromic meaning that hearing impairment is the sole consequence of the condition.

1.9. No curative treatment is available and children with the condition are managed as with any other child with severe deafness. They require special help with education and are likely to attend schools for children with severe hearing loss. Some families may opt for the use of technology such as cochlear implant. Most will adapt to their hearing loss and manage their lives accordingly, but many will adapt their life choices to meet the challenges of their hearing loss.

1.10. The committee noted the inspectorate’s request to consider whether Non Syndromic Congenital Deafness (DFNB29), OMIM #614035 should be approved for inclusion on the PGD List. The committee agreed to consider the application on this basis.

1.11. The committee noted that a licence had been issued in 2001 for a family with non-syndromic congenital hearing loss and that licences had been issued for non-syndromic sensorineural hearing loss (OMIM #600965) and for Pendred syndrome. In these cases the major disability is sensorineural hearing loss of similar severity.

2. Decision

2.1. The committee had regard to its explanatory note. On the basis of the information presented, the committee was satisfied that there is a particular risk, a 25% chance, of having a child with Non Syndromic Congenital Deafness, if each parent has a relevant mutation. However, on the basis of the information presented and in the absence of further evidence, the committee was not satisfied that there is a significant risk that a person with profound hearing loss will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition which would severely impact on their quality of life.

2.2. The committee had reviewed the information provided in this application which included information from Genetic Alliance with some extracts from publications by the National Deaf Children's Society relating to:

- family support;
- communication with deaf children;
- speech and language development/therapy;
- hearing aids and cochlear implants;

2.3. The committee considered that although the onset of the condition is during infancy, at a pre-lingual stage, early diagnosis of this condition and intervention such as speech and language development/therapy and the use of hearing aids or cochlear implants could improve an affected individual's ability to adapt to profound hearing loss and learn good communication skills. Children with cochlear implants would have the ability to take part in most activities and may even attend mainstream school if well supported from an early age. Whilst the committee did acknowledge there may be an element of risk associated with fitting cochlear implants it agreed that this is an effective treatment which is available in the UK. The committee considered the applicant's submission that many individuals with profound deafness will not consider this a disability and will manage all aspects of their lives alongside people with standard hearing. The committee further noted the Peer Reviewer's submission that many individuals with severe or profound deafness will not consider this a disability or an impairment.

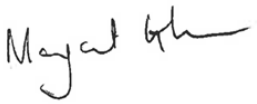
2.4. The committee considered the condition in its worst case scenario. It considered the potential impact on quality of life. It noted this particular gene mutation DFNB29 which can result in profound pre-lingual hearing loss from infancy, shows no symptoms other than profound to severe hearing loss. The committee recognised this condition could be hugely challenging to those affected.

2.5. In taking into account the condition at its worst, and without further evidence other than that submitted in the papers and by the Specialist Adviser, the Committee decided by majority vote, that it was not satisfied that the condition of Non Syndromic Congenital Deafness (DFNB29), OMIM #614035 meets the criteria for testing under paragraph 1ZA(2) of Schedule 2 of the Act.

3. Chair's signature

3.1. I confirm this is a true and accurate record of the meeting,

Signature



Name

Margaret Gilmore

Date

12 October 2017