Statutory Approvals Committee – minutes

Centre 0044 (The Centre for Reproductive and Genetic Health)

Pre-implantation Genetic Diagnosis (PGD) application for Deafness Autosomal Dominant 28, OMIM #608641

Thursday, 28 November 2019

HFEA, Spey Meeting Room, Level 2, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Anne Lampe Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr Alan Fryer	
Legal Adviser	Eve Piffaretti	Blake Morgan LLP

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:

- 9th 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 UK statement
- Supporting evidence 1
- Supporting evidence 2
- 2018-06-28 Statutory Approvals Committee Minutes - TPRN-associated autosomal recessive non-syndromic deafness, OMIM #613307
- 2017-09-28 Statutory Approvals Committee Minutes – PGD Non-Syndromic Congenital Deafness (DFNB29), OMIM #614035
- 2017-09-28 Statutory Approvals Committee Minutes – PGD Congenital Deafness with inner ear agenesis, microtia and microdontia, OMIM #610706
- 2017-07-27 Statutory Approvals Committee Minutes – PGD Non syndromic sensorineural hearing loss, OMIM #600965.

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.

1.2. The committee noted that the description in the PGD application for Deafness Autosomal Dominant 28, OMIM #608641 is consistent with the peer review.

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

1.4. The committee noted that the Genetic Alliance UK statement provided a perspective on the impact of the condition on patients, their families and carers.

1.5. 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.

1.6. 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 Deafness Autosomal Dominant 28, OMIM #608641 is inherited in an autosomal dominant manner which means there is 50% chance of having an affected child in each pregnancy, if either parent has a relevant mutation.

1.8. The committee noted the penetrance of the condition is hard to determine due to there being few reports. The penetrance is age-dependant and appears to be low before 20 years of age in the families reported in the literature but high by 50 years of age and above.

1.9. Deafness Autosomal Dominant 28, OMIM #608641 is a form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNA28 is characterized by mild to moderate hearing loss across most frequencies that progresses to severe loss in the higher frequencies by the fifth decade.

1.10. There is no cure for this condition and treatments include hearing aids, vibrotactile devices and cochlear implantation to improve hearing.

1.11. The committee noted the executive's request to consider Deafness Autosomal Dominant 28, OMIM #608641 for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that, in the worst-case scenario Deafness Autosomal Dominant 28, OMIM #608641 is a rare and serious condition, given the possibility that in certain cases an early onset of hearing loss may occur. There is no cure for the condition and treatment has a limited effect on those affected by profound or total deafness. The committee noted the possible physical and psychological effects on those affected with the condition – indeed, hearing loss may affect speech and language development if the condition manifests early in life.

2.2. The committee noted that this particular application describes a family where the patient presented at birth with hearing loss. The committee received advice from its specialist adviser and was sited to a letter from a leading expert in the clinical and molecular genetics of hearing loss who confirmed that in certain circumstances, there are individuals with a GRHL2 gene

mutation who have childhood-onset hearing loss and who have a 50:50 chance of passing on the gene to their children.

2.3. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormalities in question and that there is a significant risk, that a person with such an abnormality will, given the conditions' worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the following condition does meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

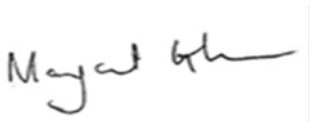
2.4. The committee agreed to authorise PGD testing for Deafness Autosomal 28, OMIM #608641, for the specific family in this application. The committee agreed that this condition is not authorised for general testing in the future and will not be added to the approved HFEA PGD list of conditions.

2.5. The committee also confirmed that it will continue to consider these types of applications on a case by case basis to allow families to have the opportunity to request PGD when individual cases reflect a worst-case scenario.

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
24 December 2019