Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital)

Preimplantation Genetic Diagnosis (PGD) application for Autosomal Recessive Deafness 77 (DFNB77), OMIM #613079

Date:	28 January 2021
Venue:	Microsoft Teams Meeting
Committee Members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde
Specialist Adviser	Peter Turnpenny
Legal Adviser	Gerard Hanratty - Browne Jacobson LLP
Members of the Executive	Moya Berry - Committee officer Catherine Burwood - Licensing Manager
Apologies:	No apologies were received for the meeting
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
- Standard Licencing and Approvals Pack

The following papers were considered by the committee:

- Executive Summary
- PGD Application form
- Redacted Peer review
- Genetic Alliance (UK) Statement
- 2018-06-28 Statutory Approvals Committee Minutes, PGD for TPRN-associated autosomal recessive non-syndromic deafness (DFNB79), OMIM #613307

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Professor Peter Turnpenny, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Autosomal Recessive Deafness 77 (DFNB77), OMIM #613079 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 that Autosomal Recessive Deafness 77 (DFNB77), OMIM #613079, is inherited in an autosomal recessive manner, which means there is a 25% chance of an embryo being affected by the condition in each pregnancy if each parent has a relevant mutation.

1.8. The committee noted that the penetrance of the condition is 100%.

1.9. Autosomal Recessive Deafness 77 (DFNB77), OMIM #613079 is a form of deafness that is very variable both within and between families. The hearing loss may be progressive in some cases and non-progressive in others, with the level of hearing loss most often being profound either from birth or progressing to profound deafness during childhood and into the teenage years.

1.10. There is no cure for this condition and treatments include the use of hearing aids and/or cochlear implant. Suitable educational programmes can also be offered to support those affected.

1.11. The committee noted the executive's request to consider Autosomal Recessive Deafness 77 (DFNB77), OMIM #613079, 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, Autosomal Recessive Deafness 77 (DFNB77), OMIM #613079, is a serious condition with the level of hearing loss most often being profound from birth, or progressing to severe/profound deafness during childhood and into the teenage years. Hearing loss may occur before language skills are developed. Cochlear implant surgery, which is not without serious risk and complications, can help to improve the quality of sound, but may not be an option for everyone. So, while some people will benefit from the treatment not everyone will. The committee considered the possible serious physical and

psychological impact on those with the condition, particularly where hearing loss may also affect speech and language development if the condition manifests early in life.

2.2. The committee noted that a similar condition, TPRN-associated autosomal recessive non-syndromic deafness (DFNB79), was licensed for PGD in 2018, and resembles the worst case of Autosomal Recessive Deafness 77 (DFNB77), OMIM #613079.

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 abnormality in question and that there is a significant risk that a person with such an abnormality will, given the condition's worst symptoms, have or develop a serious physical disability, a serious illness, or any other serious medical condition.

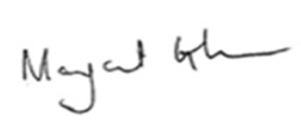
2.4. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Autosomal Recessive Deafness 77 (DFNB77), OMIM #613079

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

23 February 2021