HFEA Statutory Approvals Committee

25 September 2014

Finsbury Tower, 103-105 Bunhill Row, London, EC1Y 8HF

Minutes – Item 4

Centre 0044 (Centre for Reproductive and Genetic Health) – PGD application for Pendred Syndrome, OMIM #274600

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Sam Hartley, Head of Governance and Licensing
Debbie Barber (professional)	
Rebekah Dundas (lay)	
Jane Dibblin (lay)	Legal Adviser:
Hossam Abdalla (professional)	Dawn Brathwaite, Mills & Reeve
Advisor: Dr Edward Blair	

Declarations of Interest: The Members declared no conflicts in relation to this item.

The following papers were considered by the Committee:

- Executive summary
- PGD application form
- Redacted Peer Review form
- Genetic Alliance Opinion

The Committee also had before it:

- HFEA Protocol for the Conduct of Licence Committee Meetings and Hearings
- 8th edition of the HFEA Code of Practice
- Human Fertilisation and Embryology Act 1990 (as amended) ("the Act")
- Decision trees for granting and renewing licences and considering requests to vary a licence (including the PGD decision tree); and
- Guidance for members of Authority and Committees on the handling of conflicts of interest approved by the Authority on 21 January 2009.
- Guidance on periods for which new or renewed licences should be granted
- Standing Orders and Instrument of Delegation
- Indicative Sanctions Guidance

- HFEA Directions 0000 – 0012
- Guide to Licensing
- Compliance and Enforcement Policy
- Policy on Publication of Authority and Committee Papers
- HFEA Pre-Implantation Diagnostic Testing ("PGD") Explanatory Note For Licence Committee

Discussion

1. 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.

2. 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'.

3. The Committee noted that Pendred Syndrome, OMIM #274600 is inherited in an autosomal recessive pattern and there is a 1 in 4 chance of an embryo being affected with the condition where both parents are carriers.

4. The Committee noted the Executive's papers that stated that the symptoms of Pendred syndrome can vary from hearing loss to total deafness with thyroid enlargement, and can include thyroid gland abnormalities with mental compromise (mental retardation). The progression of Pendred syndrome can be rapid in early childhood and may be associated with head injury or infection. Vertigo can precede or accompany fluctuations in hearing, and approximately 30% of children with Pendred syndrome have symptomatic vestibular problems (problems with balance), with dizziness/vertigo. Goitre (enlargement of the thyroid gland) can become apparent after puberty, which can lead to hypothyroidism.

5. The Committee noted that symptoms are likely to develop at birth or in early childhood for affected individuals.

6. The Committee welcomed the advice of its advisor, Dr Edward Blair. Dr Blair advised that this condition is essentially profound deafness. He advised that thyroid gland abnormalities and complications were treatable, and that thyroid function was checked in all new-borns. He advised the Committee to focus on

the descriptions of worst-case scenario of profound deafness and resultant complications. The Committee accepted the advice.

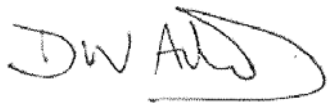
7. The Committee noted that the application is consistent with the Peer Review and is also supported by the Genetic Alliance.

8. The Committee considered that the condition is serious because it is an incurable condition that, taken in its worst form of severe deafness, would have a profound effect on the patient's quality of life.

9. The Committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The Committee was therefore satisfied that the condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 to the Act.

10. The Committee agreed to authorise the testing of embryos for Pendred Syndrome OMIM #274600.

Signed: Date: 07/10/2014



David Archard (Chair)