HFEA Statutory Approvals Committee

28 August 2014

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

Minutes – Item 3

Centre 0035 (Oxford Fertility Unit) – PGD application for Hyperphosphatasia with Mental Retardation OMIM #615716

Members of the Committee: David Archard (lay) Chair Sue Price (professional) Rebekah Dundas (lay) Jane Dibblin (lay) Advisor: Dr Peter Turnpenny	Committee Secretary: Lauren Crawford Legal Adviser: Shelley Edwards, Fieldfisher

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
- Scientific literature regarding the condition

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 hyperphosphatasia with mental retardation syndrome 4 (HPMRS4) (OMIM #615716) 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 that Characteristics of HPMRS include microcephaly (small head), seizures and severe developmental delay. HPMRS is caused by mutations in various genes (of which there are around 30) in the GPI anchor pathway involved in protein function, and mutations in the PGAP3 gene lead to HPMRS4. Symptoms of HPMRS4 include those listed as characteristics of HPMRS, as well as intellectual disability and sometimes a cleft palate. Affected children are weak and floppy from birth and often their motor milestones do not progress to being able to walk.

5. The Committee noted that Children can also develop seizures during infancy, involving muscle rigidity, convulsions, and loss of consciousness (tonic-clonic seizures). Seizures can also be associated with embarrassment and frustration. Due to the risk of seizures affected children will require 24-hour care.

6. The Committee noted that there is no treatment available for this condition.

7. The Committee noted that the application is consistent with the Peer Review.

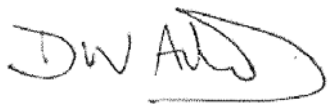
8. The Committee welcomed the advice of its Advisor, Peter Turnpenny, who confirmed that the condition was as described in the papers and is variable.

9. The Committee considered that the condition is serious as it will have a serious impact on the quality of life of the affected person who would require 24 hour care. The Committee was reminded by the Legal Adviser that it is for the Committee to consider applications to perform PGD for an abnormality without reference to the particular circumstances of any individual or family.

10. 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.

11. The Committee agreed to authorise the testing of embryos for hyperphosphatasia with mental retardation syndrome 4 (HPMRS4) (OMIM #615716).

Signed: Date: 17/09/2014



David Archard (Chair)