HFEA Statutory Approvals Committee

30 January 2014

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

Minutes – Item 4

Centre 0035 (Oxford Fertility Unit) – PGD application for Meckel-Gruber Syndrome Type 3 OMIM #607361

Members of the Committee: David Archard (lay) Chair Rebekah Dundas (lay) Sue Price (professional) Debbie Barber (professional) Advisor: Dr Peter Turnpenny	Committee Secretary: Lauren Crawford Legal Adviser: Graham Miles, Morgan Cole Also in Attendance: Sam Hartley, Head of Governance and Licensing, HFEA

Declarations of Interest: Debbie Barber works with the Oxford Fertility Unit and took no part in the discussion on this item.

The following papers were considered by the Committee:

- Executive summary
- Application form
- Redacted peer review
- Redacted Public Comment

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 Meckel-Gruber Syndrome Type 3 (OMIM #607361) 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 affected.

4. The Committee noted that this condition exhibits severe signs and symptoms that affect many parts of the body. The most common features are enlarged kidneys with numerous fluid-filled cysts; an occipital encephalocele, which is a sac-like protrusion of the brain through an opening at the back of the skull; and the presence of extra fingers and toes (polydactyly). Most affected individuals also have a build-up of scar tissue (fibrosis) in the liver. Other signs and symptoms of Meckel-Gruber Syndrome Type 3 vary widely among affected individuals. Numerous abnormalities of the brain and spinal cord (central nervous system) have been reported in people with this syndrome, including the group of birth defects known as neural tube defects. Meckel-Gruber Syndrome Type 3 can also cause problems with development of the eyes and other facial features, the heart, bones, urinary system and genitalia.

5. The Committee noted that the signs and symptoms of this syndrome typically develop at birth. Most individuals with Meckel-Gruber Syndrome Type 3 die before or shortly after birth due to their serious health problems.

6. The Committee noted that there is no curative treatment for Meckel-Gruber Syndrome Type 3.

7. The Committee noted that the application is supported by the Peer Reviewer.

8. The Committee noted that the application did not contain a Genetic Alliance Opinion but were satisfied that the information in paperwork was sufficient for them to consider the item in its absence

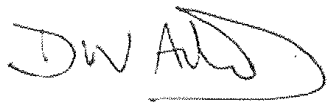
9. The Committee welcomed the advice of its Advisor, Dr Peter Turnpenny, who confirmed that the condition was as described in the papers.

10. The Committee considered that the condition is serious because most infants with Meckel-Gruber Syndrome Type 3 die of respiratory problems or kidney failure and the quality of life for affected children is severely affected.

11. 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) of Schedule 2 to the Act.

12. The Committee agreed to authorise the testing of embryos for Meckel-Gruber Syndrome Type 3 (OMIM #607361).

Signed: Date: 13/02/2014



David Archard (Chair)