HFEA Licence Committee Meeting

1 November 2012

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

Minutes – Item 2

Centre 0044 (Centre for Reproductive and Genetic Health) – PGD for Bardet-Biedl syndrome (OMIM #209900)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Sue Price (professional) Debbie Barber (professional) Jane Dibblin (lay) Anna Carragher (lay) Mair Crouch (lay) – VC Rebekah Dundas (lay) – VC	Lauren Crawford Legal Adviser: Graham Miles, Morgan Cole

Declarations of Interest: members of the Committee declared that they had no conflicts of interest in relation to this item.

The following papers were considered by the Committee

- Cover sheet
- Executive Summary
- PGD Application form
- Redacted Peer Review
- 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)
- 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 Centre's proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of schedule 2 of the Act, ie. '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 Bardet-Biedl syndrome (BBS) (OMIM #209900) is a disorder that is inherited in an autosomal recessive manner. Therefore there is a 1 in 4 chance that an embryo will have the condition if both parents are carriers.

4. The Committee noted that Bardet-Biedl syndrome (BBS) is a rare genetic disorder that affects the brain and can cause multiple physical problems including a deterioration of the intellect and neurological functions. The age of onset depends on the severity of the phenotype but the majority of cases should be diagnosed in childhood. A high frequency of obesity, usually beginning at 1 - 2 years of age with night blindness usually appearing by age 9, and legal blindness often reached by age 15. Penetrance is high (over 90%) but the phenotype can be variable.

5. The Committee noted the disorder is characterized by these symptoms: rod-cone dystrophy (a degeneration of light sensitive cells in the periphery of the retina). This eye disorder causes night blindness, tunnel vision, decreased visual acuity and photophobia (extreme sensitivity of the eyes to light). Other visual symptoms may include nystagmus (involuntary eye movement), near sightedness, strabismus (cross-eye), glaucoma, and cataracts. Some of the other common characteristics of BBS are extra fingers and/or toes, ranging from a single skin tag to a fully formed digit, short, stubby fingers and/or toes, more frequently in the feet than in the hands and webbing of toes, kidney disease, obesity and mental retardation/learning difficulties.

6. The Committee noted that there is no known cure, but some treatments of specific organs and systems may be beneficial. Treatment is limited to complications such as diabetes and does not address the underlying condition. There is no known treatment for the progressive vision problems that often occur in people with BBS. However, there is much that can be done to prepare for a life with low vision. An ophthalmologist should be consulted to accurately diagnose the specific problems, and other vision professionals, including low vision specialists, can assist in making life easier for affected children and adults. The quality of life for affected individuals and their families is significantly reduced.

7. The Committee considered the condition is serious because the condition is 90% penetrant, it is diagnosed in early childhood, severely affects the eyesight, includes kidney disease and that there is little or no effective treatment. The quality of life for affected individuals and their families is significantly reduced. The condition is variable, but also unpredictable.

8. The Committee noted that the application is supported by the Peer Reviewer and the Genetic Alliance UK.

9. The Committee had regard to its explanatory note, in particular paragraph 5.5 '*Where a condition has variable symptoms, the Licence Committee will base its determination of how serious the disability, illness or condition is, on the worst possible symptoms'*, and noted that on the basis of the information presented, given the conditions worst symptoms, it was also 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. Accordingly, it was appropriate to grant the application under paragraph 1ZA(1)(b) of Schedule 2 to the Act.

10. The Committee agreed to authorise the testing of embryos for Bardet-Biedl syndrome (BBS) (OMIM #209900)**.** The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 15/11/2012

David Archard (Chair)