HFEA Licence Committee Meeting

14 June 2012

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

Minutes – Item 2

Centre 0044 (The Centre for Reproductive and Genetic Health) – PGD for Epilepsy, female restricted, with mental retardation (EFMR) (OMIM #300088)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Sue Price (professional) Mair Crouch (lay) Jane Dibblin (lay) Debbie Barber (professional)	Lauren Crawford Legal Adviser: Juliet Oliver, Field Fisher Waterhouse

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

1. HFEA Protocol for the Conduct of Licence Committee Meetings and Hearings
2. 8th edition of the HFEA Code of Practice
3. Human Fertilisation and Embryology Act 1990 (as amended)
4. Decision trees for granting and renewing licences and considering requests to vary a licence (including the PGD decision tree); and
5. Guidance for members of Authority and Committees on the handling of conflicts of interest approved by the Authority on 21 January 2009.
6. Guidance on periods for which new or renewed licences should be granted
7. Standing Orders and Instrument of Delegation
8. Indicative Sanctions Guidance
9. HFEA Directions 0000 – 0012

10. Guide to Licensing
11. Compliance and Enforcement Policy
12. Policy on Publication of Authority and Committee Papers
13. 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 considerable 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 received legal advice that they were entitled to authorise testing of an embryo for one or more of the purposes under paragraph 1ZA(1) of schedule 2 to the Act. Where the centre wishes to test an embryo to establish its sex, the Committee must consider whether the criteria in paragraph (1)(c) are met; they must then go on, if the centre wishes also to test whether the embryo has an abnormality or any other gene, chromosome or mitochondrion abnormality, to consider whether the criteria in paragraph (1)(b) and (2) are met.

3. The Committee noted that the Centre's proposed purpose of testing the embryos was both: (1) 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'; and (2) as set out in paragraph 1ZA(1)(c) of schedule 2, 'to establish the sex of the embryo in case where there is a particular risk that any resulting child will have or develop a gender-related serious physical or mental disability'.

4. The Committee noted that Epilepsy, female-restricted, with mental retardation (EFMR) (OMIM #300088) is a disorder that is inherited in an X-linked dominant manner. Therefore 50% of the female embryos from a mildly affected mother are likely to inherit the mutation and display the phenotype. All the male embryos from such a female would be phenotypically normal; however half of them could carry the mutation and transmit to the females of the next generation.

5. The Committee considered that there is a significant risk that a female person with the abnormality will develop a serious medical condition. It is

90% penetrant in affected females. The Committee considered that the condition is serious in females for the following reasons:

6. The onset of EFMR is usually in the first year of life. Affected girls suffer from frequent seizures often occurring in groups, declining in frequency from the third year, and usually ending by the end of the sixth year of life. Seizures occur unpredictably and can require immediate admission to hospital. The condition is rare and poorly understood and there is no known effective treatment regime.

7. The Committee noted that these seizures can lead to the developmental delay and learning difficulties associated with EFMR. The level of developmental delay and learning difficulties is variable, from mild to severe. Severely affected girls will require 24 hour care for life and may never obtain any form of independence.

8. Further, the Committee considered that the condition is serious because EFMR symptoms include mild to severe epilepsy episodes that could be very frequent and last for a few days. Although the seizures decrease in frequency in adult life, the seizures in infancy are generally not responsive to treatment, consequently causing varying degrees of mental retardation and cognitive impairment. Many of the clinical cases have also reported obsessive and aggressive behaviours in patients, as well as displaying a range of various autistic features. EMFR can have a varying age of onset from approximately 7 months to 5 years of age.

9. The Committee noted that the condition was both variable, and unpredictable. It 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 female 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 noted that the condition affected only the female sex, or affected the female sex significantly more than the male sex and was also therefore satisfied that there is a particular risk that any resulting child will have or develop a gender-related serious physical or mental disability. It considered that it was appropriate to grant the application under paragraph 1ZA(1)(c) of Schedule 2 to the Act.

11. The Committee noted that the application is supported by Genetic Alliance UK.

12. The Committee agreed to authorise the testing of embryos for Epilepsy, female-restricted, with mental retardation (EMFR) (OMIM #607208) to define affected female embryos only. The Committee confirmed that this authorisation will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 28/06/2012

David Archard (Chair)