HFEA Licence Committee Meeting

28 March 2013

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

Minutes – Item 2

Centre 0102 (Guy's Hospital) – PGD for Fried Syndrome (OMIM ~300630)

Members of the Committee: Sue Price (professional) Chair Debbie Barber (professional) Jane Dibblin (lay) Andy Greenfield (lay)	Committee Secretary: Lauren Crawford Legal Adviser: Stephen Hocking, Beachcroft LLP

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

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 supporting documents with this application did not contain a Genetic Alliance Opinion. The Committee were satisfied that they had, in any event, sufficient information before them on which to make a decision.

3. The Committee noted that the Centre's proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(c) of schedule 2 of the Act, ie. '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, serious illness or serious medical condition' and also paragraph 1ZA(1)(b) of Schedule 2 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'.

4. The Committee noted Fried Syndrome, OMIM #300630 is a rare condition caused by mutations in the AP1S2 gene. Fried Syndrome is inherited in an X-linked recessive manner. This means for male embryos there is a 1 in 2 chance of the embryo being affected where the mother is a carrier of the condition..

5. The Committee noted that Fried syndrome is a rare X-linked condition leading to intellectual disability, which is usually severe. Other features that have been described in affected individuals have included: low muscle tone in childhood with delayed walking and motor development, spastic diplegia (a form of cerebral palsy), poor language skills, difficult and aggressive behaviour, hydrocephalus, calcification of the basal ganglia (a brain finding visible on MRI scan), and mild facial dysmorphism (unusual facial appearance).

6. The Committee considered that the condition is serious because the quality of life for affected males is reduced by both mental and physical impairment. At worst, affected individuals will require full time care for their health and social wellbeing - some individuals use a wheelchair and will be fully dependant requiring help with toilet and dressing. The only treatments available are

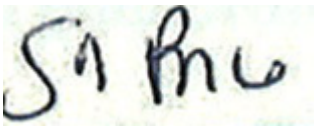
supportive- speech, physical, educational and occupational therapy may be appropriate.

7. The Committee noted that the purpose for the proposed testing is to establish whether the embryo has an X-linked condition. 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 male 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 for either and both of the purposes specified under paragraph 1ZA(1)(b) and (c) of Schedule 2 to the Act.

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

9. The Committee agreed to authorise the testing of embryos for Fried Syndrome (OMIM #300630). The Committee confirmed that this authorisation will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 15/04/2013



Sue Price (Chair)