HFEA Licence Committee Meeting

26 April 2012

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

Minutes – Item 3

Centre 0044 (Centre for Reproductive and Genetic Health) – PGD for Hypochondroplasia (OMIM #146000)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Anna Carragher (lay) Rebekah Dundas (lay) (videoconference) Mair Crouch (lay) Sue Price (professional) Debbie Barber (professional) Jane Dibblin (lay)	Joanne McAlpine Legal Adviser: Stephen Hocking, DACBeachcroft 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
- 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 much 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 Hypochondroplasia (OMIM #146000) is a disorder that is inherited in an autosomal dominant manner. Where one parent carries an affected copy of the relevant gene there is a 1 in 2 chance (50% risk) of the embryo exhibiting the syndrome.

4. The Committee noted that this condition is 100% penetrant, and a majority of the individuals diagnosed with this condition are diagnosed in early to mid-childhood. The condition is caused by a mutation in the FGFR3 gene.

5. The Committee noted that Hypochondroplasia is a form of short-limbed dwarfism and the severity of the condition can vary. It can result in only mild short stature at one end of the spectrum to more severe problems similar to achondroplasia at the other.

6. The Committee noted that occasional symptoms can include bowing of the lower legs associated pain and reduced mobility, requiring surgery, ventriculomegaly, occasionally requiring ventriculoperitoneal shunting, mild developmental delay, related to joint hypermobility, ENT problems causing speech delay, as well as generalized difficulty with learning.

7. The Committee noted that treatment options are available to manage specific complications through growth hormone therapy but these are generally not effective in improving final height and may exacerbate the physical and functional problems.

8. The Committee considered the seriousness of the condition and noted that they had conflicting information within the application on the description of the condition given between that of the peer reviewer and the Genetic Alliance.

9. The Committee agreed that it had insufficient information on which to make its determination on this condition and therefore agreed not to consider this application in its present form, pursuant to s.16(4) of the1990 Act .

10. The Committee would be happy to consider a new application for this condition from this centre or another centre that wishes to make an application in the future. The Committee would ask if a new application is made that the following information is included;

    - Clear statement on the impact on quality of life and the activities of day to day living of shortness of stature from relevant and informed interest groups
    - A statement on the probability of each of the possible effects of this syndrome manifesting themselves in an affected individual and how the syndrome meets the statutory test of a significant risk of a serious physical or mental disability, serious illness or other serious medical condition,

Signed: Date: 10/05/2012

David Archard (Chair)