HFEA Statutory Approvals Committee

19 December 2013

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

Minutes – Item 3

Centre 0035 (Oxford Fertilty Unit) – PGD application for Craniofrontal Dysplasia OMIM #304110

Members of the Committee: David Archard (lay) Chair Sue Price (professional) Hossam Abdalla (professional) Jane Dibblin (lay) Advisor: Dr Ed Blair	Committee Secretary: Lauren Crawford Legal Adviser: Graham Miles, Morgan Cole Observing: Sam Hartley, Head of Governance and Licensing, HFEA

Declarations of Interest: The Members declared no conflicts in relation to this item. The Advisor, Ed Blair, works with the Oxford PGD unit, but this was not seen as a conflict as he did not partake in the decision making process and also declared that he had no commercial, scientific or professional interest in this item.

The following papers were considered by the Committee

- Executive summary
- Application form
- Redacted peer review
- Genetic Alliance opinion
- Correspondence from centre confirming testing methodology
- Executive Summary of additional information
- Correspondence from centre providing additional information including peer reviewed publications

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 Craniofrontal Dysplasia (OMIM #304110) is an X-linked disorder which affects females. The particular risk of this disorder is dependent on which parent carries the mutated gene. If the male parent is a carrier then all of his female children will be affected and none of the male children will be affected. If the female parent is affected there is a 1 in 2 chance of her female children being affected by the condition.

4. The Committee noted that Craniofrontal Dysplasia is characterised in females by widely-spaced eyes (hypertelorism), a broad flattened nose with a vertical groove on the top and a prematurely fused skull with a flattened and prominent forehead (coronal craniosynostosis with brachycephaly and frontal bossing). They may be born deaf. They may have a cleft palate. They may have various dental abnormalities. Their shoulders may be sloping with abnormally formed collar bones. They may develop curvature of the spine and may have a number of abnormalities relating to their fingers and toes.

5. The Committee noted that the condition can be extremely disfiguring causing severe psychological distress. A severely affected child is likely to spend a significant amount of time in hospital following repeated surgery for fused skull sutures, and widely spaced eyes. There may also be surgery related to a cleft lip and palate. There may also be mild intellectual impairment.

6. The Committee noted that treatment for this condition include may be treated by craniosynostosis correction or surgical correction of the hypertelorism and other deformities. Craniosynostosis can result in increased intracranial pressure and if it is not corrected it can have severe aesthetic and developmental complications.

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

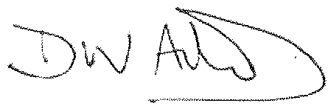
8. The Committee welcomed the advice of its Advisor, Ed Blair, who confirmed that the condition was as described in the papers. He explained about the possible mental impairment and that it can occur if the disorder is left untreated and that affected individuals suffer damage to the optic nerves due to compression of the skull. He also advised the committee that affected males as described in these papers may not have the exact same condition.

9. The Committee considered that the condition is serious because repeated surgery is necessary for many children with this condition from an early age and may also need to undergo further surgeries relating to skeletal problems as they grow.

10. 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) and (2) of Schedule 2 to the Act.

11. The Committee agreed to authorise the testing of embryos for Craniofrontal Dysplasia (OMIM #304110).

Signed: Date:



David Archard (Chair)