HFEA Statutory Approvals Committee

27 March 2014

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

Minutes – Item 1

Centre 0102 (Guy's Hospital) – PGD application for Optic Atrophy 1 OMIM #165500

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford
Sue Price (professional)	
Hossam Abdalla (professional)	Legal Adviser:
Jane Dibblin (lay)	Sarah Ellson, Field Fisher Waterhouse
	Also in attendance:
	Sam Hartley, Head of Governance and Licensing, HFEA
	Dawn Braithwaite, Mills and Reeve

Declarations of Interest: The Members declared no conflicts in relation to this item.

The following papers were considered by the Committee:

- Executive summary
- 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) ("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 Optic Atrophy 1 (OMIM #165500) is inherited in an autosomal dominant pattern and there is a 1 in 2 chance of an embryo being affected with the condition where one parent is affected. This means that there is a particular risk to the embryo given the mode of inheritance.

4. The Committee noted that Optic Atrophy 1 is caused by alterations in a gene known as OPA1. The OPA1 gene has an important function in the mitochondria (cellular energy producing factories). The cells in the retina of the eye are rich in mitochondria. When the OPA1 gene is not functioning properly it can lead to an increase rate of cell death in the retina, which can lead to degeneration of the optic nerve. This means that the signal from the eye to the brain cannot be transmitted properly.

5. The Committee noted that the severity of the vision loss varies among affected people, including among members of the same family. Affected individuals can range from having nearly normal vision to complete blindness. The vision loss usually progresses slowly, and problems can include with colour vision (making it difficult or impossible to distinguish between shades of blue and green), and progressive tunnel vision. Up to 10% of people with an optic atrophy 1 gene alteration have additional problems, most commonly

sensorineural hearing loss and weakness and lack of voluntary coordination of muscles.

6. The Committee noted that affected individuals usually start to experience visual decline between four and six years of age.

7. The Committee noted that there is no curative treatment for this condition. Low-vision aids can be used to support affected individuals.

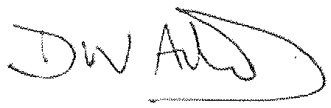
8. The Committee noted that the application is consistent with the peer review and the Genetic Alliance opinion.

9. The Committee considered that the condition is serious because it is a severe progressive disorder which begins in childhood and is unable to be cured.

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 Optic Atrophy 1 (OMIM #165500).

Signed: Date: 08/04/2014



David Archard (Chair)