HFEA Licence Committee Meeting

28 March 2013

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

Minutes – Item 4

Centre 0102 (Guy's Hospital) – PGD for Autosomal Dominant Retinitis Pigmentosa OMIM #180100

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
- Genetic Alliance and RP Fighting Blindness Opinion
- Additional email from peer reviewer regarding the approval of all types of RP

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 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 that Autosomal Dominant Retinitis Pigmentosa is one form of a heterogeneous group of inherited ocular diseases known as Retinitis Pigmentosa (RP – OMIM #268000). The condition can result from the autosomal recessive, autosomal dominant, or X-linked inheritance of a number of genetic mutations each of which is allocated an individual OMIM number.

4. The Committee noted that Autosomal Dominant Retinitis Pigmentosa (OMIM #180100) is inherited in an autosomal dominant manner. There is therefore a 1 in 2 chance of the embryo with an affected parent being affected by this condition.

5. The Committee noted that most individuals with this diagnosis will have some loss of vision by their 20s or 30s. However there is significant variability in the age of onset, with some signs being visible to an ophthalmologist or on electrical testing in the first decade of life.

6. The Committee noted that there is variability in the rate of progression of RP. The first symptoms usually relate to problems with vision in dim light, meaning that it can take longer to adjust to poor light, or it may not be possible to adjust. Over time, slow progression of peripheral visual loss leads to restrictedvision, which can deteriorate into tunnel vision. In some cases there can be further deterioration so that central vision, which is used for detailed vision, is either compromised or lost. Manypeople with RP also experience discomfort with bright lights (photophobia) which can worsen over the course

of the progression of the condition. Some individuals with RP develop cataracts and/or macular oedema (swelling of the retina caused by leaking blood vessels).

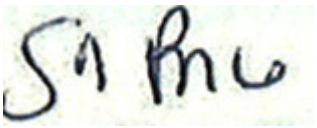
7. The Committee considered that the condition is serious because at the most extreme end of this disease spectrum a patient could be blind in the 2nd to 3rd decade of life with subsequent significant impact on quality of life. Although penetrance is incomplete and expression is variable it is difficult to predict which patient will have the most severe symptoms. Treatment for this condition is limited. RP is currently not curable. The effects of photophobia can be managed using UV-blocking sunglasses. Cataracts can be removed but this does not alter the course of the RP degeneration. Use of visual aids such as magnifiers can help maximize useful vision.

8. 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) of Schedule 2 to the Act.

9. The Committee noted that the application is supported by the Peer Reviewer and the Genetic Alliance (supported by RP Fighting Blindness).

10. The Committee agreed to authorise the testing of embryos for Autosomal Dominant Retinitis Pigmentosa (OMIM #180100). The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 15/04/2013



Sue Price (Chair)