HFEA Licence Committee Meeting

14 June 2012

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

Minutes – Item 1

Centre 0102 (Guy's Hospital) –PGD for X Linked Retinitis Pigmentosa (RP3) (OMIM 300029)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Mair Crouch (lay) Sue Price (professional) Debbie Barber (professional) Jane Dibblin (lay)	Joanne McAlpine Legal Adviser: Juliet Oliver, Field Fisher Waterhouse

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 PGD for X Linked Retinitis Pigmentosa (RP3) (OMIM #300029) is a disorder that is inherited in an X-linked manner. This means that carrier women have a 50% chance of having an affected son and a 50% chance of having a carrier daughter.

4. The Committee considered that there is a significant risk that a male person with the abnormality will develop a serious medical condition because it is fully penetrant in affected males. Although less likely, females who carry the condition may develop the full condition also.

5. The Committee noted that RP3 is caused by a gene change within the RPGR gene on the X chromosome. RP3 is a severe form of retinal degeneration which means a deterioration in part of the eye that receives light signals and as a result leads to visual loss and in many cases complete loss of sight.

6. The Committee noted that the effects of the genetic mutation causing X-linked Retinitis pigmentosa typically present in early adulthood but may present as early as 10 years of age. Complete visual loss may typically occur in affected individuals in their thirties and forties.

7. The Committee considered the symptoms of RP3 which include night blindness, the development of tunnel vision, and slowly progressive decreased central vision generally starting at approximately 20 years of age. Affected individuals may have, constricted visual fields and dyschromatopsia (impaired perception of colour). The classic fundus appearance includes dark pigmentary clumps in the midperiphery and perivenous areas ('bone spicules'), attenuated retinal vessels, cystoid macular edema, fine pigmented vitreous cells, and waxy optic disc pallor. Retintis Pigmentosa can be associated with posterior subcapsular cataracts, high myopia, astigmatism, keratoconus, and mild hearing loss.

8. The Committee noted that RP3 affects both males and females. It affects males severely. The range of severity in females is broad, from asymptomatic carriers to severe expression comparable to the male form of the condition. The severity of expression in females is not possible to predict.

9. The Committee noted that noted that treatments are limited for RP3 and mainly consist of visual aids and the support of visual disability specialists to help those affected to adapt to the visual impairment. The Committee noted that the condition itself is not curable. Due to the wide range of effects, the quality of life will be diminished in many ways.

10. The Committee noted that the application both the Peer Reviewer and Genetic Alliance UK consider the condition to be serious.

11. The Committee had regard to its explanatory note, in particular paragraphs 5.4 *Where a condition has a range of penetrance (eg. 40-60%), the Licence Committee will base its decision on the highest penetrance figure* and 5.5 '*Where a condition has variable symptoms, the Licence Committee will base its determination of how serious the disability, illness or condition is, on the worst possible symptoms'*. The Committee noted that on the basis of the information presented, given the highest degree of penetrance and 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 another serious medical condition. Accordingly, it was appropriate to grant the application under paragraph 1ZA(1)(b) of Schedule 2 to the Act.

12. The Committee noted that while they may grant approval, the centre providing PGD for the condition must also ensure that each individual case treated fulfills the severity criteria of Schedule 2 paragraph 1ZA of the HFE Act 1990 (as amended).

13. The Committee agreed to authorise the testing of embryos for X Linked Retinitis Pigmentosa (RP3) (OMIM #300029). The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 25/06/2012
David Archard (Chair)