HFEA Licence Committee Meeting

7 March 2013

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

Minutes – Item 4

Centre 0102 (Guy's Hospital) – PGD for Gorlin Syndrome OMIM #109400

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford
Rebekah Dundas (lay)	
Debbie Barber (professional)	
Jane Dibblin (lay)	Legal Adviser:
Andy Greenfield (lay)	Juliet Oliver, Field Fisher
Bishop Lee Rayfield	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

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 supporting documents with this application did not contain a Genetic Alliance Opinion. The Committee were satisfied that they had, in any event, sufficient information before them on which to make a decision.

3. 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'.

4. The Committee noted that Gorlin Syndrome, OMIN #109400 is inherited in an autosomal dominant manner. There is therefore a 1 in 2 chance of the embryo being affected by this condition where a parent is affected.

5. The Committee noted that, in its most severe form, the condition would apparent from birth, with the most noticeable feature being a large head with frontal bossing that may require delivery by Caesarean section. Congenital anomalies may include abnormal ribs or vertebrae, cleft lip and/or palate, polydactyly, strabismus, cataract or other eye problems and cardiac fibromas, that may cause arrhythmia or obstruct cardiac flow.

6. The Committee noted that there may be some delay of motor development in the early years, and that about 5% will develop a malignant brain tumour called a medulloblastoma. Multiple jaw cysts are common and mostly develop in teenage years. There are several different skin manifestations including facial milia, skins pits on the palms and soles, sebaceous and dermoid cysts. The most significant skin lesions are basal cell carcinomas, which are usually multiple and develop continuously from late teenage years to adulthood. Ovarian fibromas can also occur, which may cause problems during pregnancy.

7. The Committee noted that although the condition shows intra and interfamilial variation in expression, experience clinically and from molecular testing is compatible with complete penetrance. 60% of individuals have macrocephaly, 90% have skeletal abnormalities, 90% develop multiple keratocysts, 90% develop basal cell carcinomas, 5% develop medulloblastoma in childhood, 20% females develop ovarian fibromas, 2% develop cardiac fibromas.

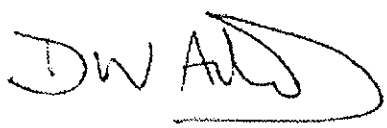
8. The Committee note that affected individuals are radio-sensitive which limits the treatment options available for malignancies.

9. The Committee considered that the condition is serious because individuals with Gorlin syndrome require surveillance from birth with input from multiple specialists including paediatricians, dentists, orthodontists, ophthalmologists and dermatologists. Investigations include regular physical examinations, -scans with the least radiation to identify congenital anomalies and monitor for new lesions. Symptoms of the condition are likely to develop continuously throughout life and require ongoing management. Physical impairment may result from treatment of multiple BCCs depending on the site and severity of the lesions.

10. The Committee noted that the application is supported by the Peer Reviewer.

11. 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.

12. The Committee agreed to authorise the testing of embryos for Gorlin Syndrome, OMIM #109400. The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed:  Date: 26/03/2013

David Archard (Chair)