HFEA Statutory Approvals Committee

30 October 2014

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

Minutes – Item 6

Centre 0102 (Guy's Hospital) – PGD application for Nance-Horan Syndrome OMIM #302350

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford, Head of Governance and Licensing
Sue Price (professional)	
Debbie Barber (professional)	
Rebekah Dundas (lay)	Legal Adviser:
Jane Dibblin (lay)	Dawn Brathwaite, Mills & Reeve
Advisor: Dr Peter Turnpenny	

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

The following papers were considered by the Committee:

- Executive summary
- PGD application form
- Redacted Peer Review form
- 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 Nance-Horan Syndrome OMIM #302350 is inherited in an X-linked manner but the applicant has provided information that female embryos with a single copy of the gene can also be affected by this condition and on this basis there is a 1 in 2 chance of having an affected child in each pregnancy. Males will usually be more severely affected than females.

4. The Committee noted that there are three main features of Nance-Horan syndrome. Males are born with congenital cataracts and the front part of the eyes is smaller than it should be. Affected children have impaired vision and nystagmus leads to repetitive, involuntary to-and-fro oscillations of the eyes. About 50% of older affected males develop glaucoma, which can damage the optic nerve connecting the eye to the brain and the nerve fibres from the retina. Female carriers may have cataracts but usually have normal vision. Nance-Horan syndrome causes the teeth, in particular the front teeth, to have an abnormal shape in both baby and permanent teeth. There may be gaps between teeth and female carriers may exhibit problems with the teeth. Approximately 30% of male patients have mild to moderate learning difficulties. Other features include the shortening of the fifth bone in the hand and facial dysmorphism, which can include a long, sometimes narrow and rectangular face, a marked and long chin, a large nose with a high, narrow nasal bridge and large, often protruding ears. The applicants have provided information that although far less common, severe learning difficulties have been described in some females. This is not reported by the peer reviewer.

5. The Committee noted that affected males have congenital cataracts from birth and about 50% develop glaucoma (eye conditions that cause optic nerve damage

and can affect sight), as young adults. The dental abnormalities become apparent when the teeth develop and the learning difficulties become apparent in childhood.

6. The Committee welcomed the advice of its advisor, Dr Peter Turnpenny. Dr Turnpenny confirmed that the condition is as described in the papers. He advised that the level of the intellectual impairment is variable from mild to moderate to in some cases severe. He also advised that females can also develop serious intellectual impairment.

7. The Committee noted that the application is consistent with the Peer Review and is also supported by the Genetic Alliance.

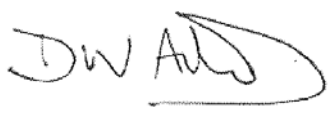
8. The Committee considered that the condition is serious because it involves multiple surgeries on the eyes from a young age with the possibility of complete visual loss.

9. 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.

10. The Committee noted that there are some other symptoms of the condition which they did not consider as meeting the test of seriousness. These features include the shortening of the fifth bone in the hand and facial dysmorphism, which can include a long, sometimes narrow and rectangular face, a marked and long chin, a large nose with a high, narrow nasal bridge and large, often protruding ears. There may be gaps between teeth and female carriers may exhibit problems with the teeth.

11. The Committee agreed to authorise the testing of embryos for Nance-Horan Syndrome OMIM #302350.

Signed: Date: 13/11/2014



David Archard (Chair)