HFEA Statutory Approvals Committee

30 October 2014

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

Minutes – Item 1

Centre 0102 (Guy's Hospital) – PGD application for Trichothiodystrophy, OMIM #601675

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford, Committees Officer
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 Trichothiodystrophy, OMIM #601675 is inherited in an autosomal recessive pattern and there is a 1 in 4 chance of an embryo being affected with the condition where both parents are carriers.

4. The Committee noted that Trichothiodystrophy is a rare ectodermal disorder affecting epidermic tissues, including nails, hair, glands of the skin and the nervous system. The clinical symptoms are variable and include intellectual impairment, developmental delay, poor coordination, nail abnormalities, congenital cataracts (cloudy lens), microcephaly (small head), eye and dental abnormalities, severe infections, ichthyotic skin (dry and scaly skin), sparse sulphur deficient hair, small stature, bone anomalies and hypogonadism (poor hormone production from ovaries and testes). Recurrent infections with a high mortality rate (a 20 fold increase below the age of 10), and maternal pregnancy complications are well recognised with this condition. Early onset and progressive hip necrosis (tissue death) is an additional complication. Around 50% of affected individuals exhibit photosensitivity (light sensitivity), due to the underlying defective DNA repair. Most children with this condition will not survive to see adulthood.

5. The Committee noted that the abnormalities are usually identified at birth, however a number will be detected in the foetus before birth.

6. The Committee also noted that there is no cure is available. Supportive care is available, including fluid management in Collodian infants, emollients for icthyosis (with some benefit but not cure), treatment of infections with antibiotics and supportive care, surgery for cataracts. Individuals with intellectual disability will need educational support and later social and/or ongoing extensive parental support, especially for those unable to live independently.

7. The Committee welcomed the advice of its advisor, Dr Peter Turnpenny. Dr Turnpenny confirmed that this condition is as explained within the papers. He confirmed that early onset is usual with this condition but there are also some rare cases of affected individuals surviving to adult life. The Committee accepted the advice.

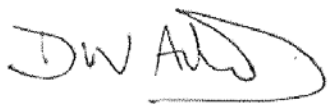
8. The Committee noted that the application is consistent with the Peer Review and is also supported by the Genetic Alliance.

9. The Committee considered that the condition is serious because it is a severe neurological condition, affected children will need a multi-disciplinary care approach with multiple specialists involved, regular hospital admissions and full time care from their parents. There is a significant increase in mortality before the age of 10.

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 Trichothiodystrophy, OMIM #601675.

Signed: Date: 13/11/2014



David Archard (Chair)