HFEA Statutory Approvals Committee

27 March 2014

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

Minutes – Item 2

Centre 0102 (Guy's Hospital) – PGD application for Homocystinuria OMIM #236200

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford
Sue Price (professional)	
Rebekah Dundas (lay)	Legal Adviser:
Hossam Abdalla (professional)	Sarah Ellson, Field Fisher Waterhouse
Jane Dibblin (lay)	
	Also in attendance:
	Sam Hartley, Head of Governance and Licensing, HFEA
	Dawn Braithwaite, Mills and Reeve

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

The following papers were considered by the Committee:

- Executive summary
- 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 Homocystinuria (OMIM #236200) 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. The Committee was satisfied that there was therefore a particular risk for an embryo given the mode of inheritance.

4. The Committee noted that Homocystinuria is a multi-system disorder and affects the connective tissues. Individuals affected by vascular involvement have an increased tendency to clotting. Clotting is the major cause of morbidity and even death in affected individuals. Vascular damage may have occurred prior to diagnosis and is irreversible even with treatment. Ectopia lentis (displaced lens) is common with severe short sightedness in some people. Surgery can correct physical abnormalities such as scoliosis and ectopia lentis.

5. The Committee noted that affected individuals have long thin bones and are prone to osteoporosis, as well as suffering other skeletal abnormalities. Individuals affected by vascular involvement have an increased tendency to clotting. Most of those affected will have a learning disability and 20% of those affected develop seizures.

6. The Committee noted that the age of onset for the disorder is variable from infancy to adulthood.

7. The Committee noted that irreversible vascular damage may have occurred prior to diagnosis. These individuals require long term surveillance and care. Patients need to have surgery to correct physical abnormalities such as scoliosis and ectopia lentis. Medical management is the mainstay and involves diet restriction and life long medications such as pyridoxine (vitamin B6) to stabilise and maintain the homocysteine levels. A severely protein restricted diet is required in B6 responsive individuals. In those who are B6 non responsive, a severely methionine restricted (non meat, dairy soya) diet with frequent monitoring is required although this is difficult to tolerate if not introduced from infancy. They also need Betaine which can have side effects as well as poor compliance. It is difficult to achieve compliance with diet in all individuals particularly those diagnosed late.

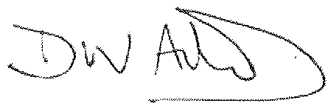
8. The Committee noted that the application is consistent with the peer review and the Genetic Alliance opinion.

9. The Committee considered that the condition is serious because there are significant lifelong health problems, which can include psychiatric problems and early death.

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 Homocystinuria (OMIM #236200).

Signed: Date: 08/04/2014



David Archard (Chair)