HFEA Statutory Approvals Committee

25 September 2014

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

Minutes – Item 8

Centre 0102 (Guy's Hospital) – PGD application for Osteogenesis Imperfecta Type VIII OMIM #610915

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Sam Hartley, Head of Governance and Licensing
Debbie Barber (professional)	
Rebekah Dundas (lay)	
Jane Dibblin (lay)	Legal Adviser:
Hossam Abdalla (professional)	Dawn Brathwaite, Mills & Reeve
Advisor: Dr Edward Blair	

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 Osteogenesis Imperfecta Type VIII (OI VIII) OMIM #610915 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 OI VIII is a serious condition which affects formation of the bones meaning that they are very fragile. There are overlaps between this type of OI and Types II and III, which have already been licensed for PGD by the HFEA. In some cases this condition is lethal in the first few weeks of life. Surviving children suffer numerous fractures which lead to increasing levels of disability as they grow. In the most severe cases the condition is lethal as it causes a chest abnormality which in turn leads to underdeveloped lungs. In this particular form of OI the bones are undermineralised, meaning that fractures occur without obvious cause and are present at birth, such as multiple fractures of the long bones and ribs. It also causes growth deficiency in the womb, meaning babies can be born with short limbs and a small chest leading to breathing difficulties, and the long bones are often malformed. Breathing and swallowing is often impaired, and cause of death is usually linked to respiratory or cardiac problems. Growth deficiency is severe, and over time fractures and associated problems lead to malformation and bowing of the long bones. Fractures and associated

problems with the spine can lead to curvature of the spine, very short stature and chest abnormailty. Pain can also be a feature of OI.

5. The Committee noted that penetrance is 100% as far as is known, and whilst the condition is variable it is always expected to be severe if not lethal. In some cases this condition is lethal in the first few weeks of life.

6. The Committee welcomed the advice of its advisor, Dr Edward Blair, who confirmed that the condition was as described in the papers.

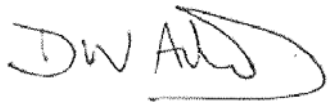
7. The Committee noted the responses from the Peer Reviewer and also noted that this application is supported by the Genetic Alliance.

8. The Committee considered that the condition is serious because it is always expected to be severe, if not lethal. The Committee also noted that the condition can be lethal in the first few weeks of life.

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 agreed to authorise the testing of embryos for Osteogenesis Imperfecta Type VIII OMIM #610915.

Signed: Date: 07/10/2014



David Archard (Chair)