HFEA Statutory Approvals Committee

19 December 2013

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

Minutes – Item 2

Centre 0119 (Birmingham Women's Hospital) – PGD application for Desbuquois Dysplasia (DBDQ) OMIM #251450

Members of the Committee: David Archard (lay) Chair Sue Price (professional) Hossam Abdalla (professional) Jane Dibblin (lay) Advisor: Dr Ed Blair	Committee Secretary: Lauren Crawford Legal Adviser: Graham Miles, Morgan Cole Observing: Sam Hartley, Head of Governance and Licensing, HFEA

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
- 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 Desbuquois Dysplasia (DBQD) (OMIM #251450) 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 DBQD is a severe disease caused by a mutation in a gene known as CANT1. Children who inherit two copies of the mutated gene have growth retardation before and after their birth, leading to severe dwarfism. They also suffer from severe joint problems including hand and foot anomalies, and progressive scoliosis. Pregnancies may not reach birth, but those children who do survive the first few years of life have extremely restricted movement. One characteristic feature of many cases is major breathing difficulties due to the small size of the body which can often end up being a lethal problem.

5. The Committee noted that treatments centre around the temporary alleviation of complications, such as surgery, but the symptoms continue to progress and this condition cannot be cured.

6. The Committee noted that the application is supported by the Genetic Alliance UK and the Peer Reviewer.

7. The Committee welcomed the advice of its Advisor, Ed Blair, who confirmed that the condition was as described in the papers. He also emphasised the

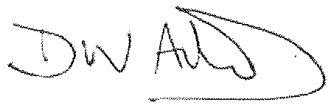
fact that in the worst case scenario it is a very severe disorder; this variability in disease expression is unpredictable.

8. The Committee considered that the condition is serious because quality of life is severely impaired due to breathing and mobility difficulties and in some cases death in utero can occur.

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 Desbuquois Dysplasia (DBQD) (OMIM #251450).

Signed: Date: 08/01/2014



David Archard (Chair)