Human Fertilisation and Embryology Authority

Minutes of the Statutory Approvals Committee

Meeting held at Finsbury Tower, 103-105 Bunhill Row, London, EC1Y 8HF on
28 May 2015

Minutes – item 1

Centre 0035 (Oxford Fertility Unit) – PGD application for Joubert syndrome 5, OMIM #610188

Members of the Committee	David Archard (Chair, lay) Rebekah Dundas (Deputy Chair, lay) Sue Price (professional)
Legal Adviser	Dawn Brathwaite, Mills & Reeve
Specialist Attending	Prof Peter Turnpenny
Members of the Executive	Sam Hartley, Head of Governance and Licensing Trent Fisher, Secretary

Declarations of interest: members of the committee declared that they had no conflicts of interest in relation to this item.

The following papers were considered by the committee:

- executive summary
- PGD application form
- redacted peer review
- Genetic Alliance opinion
- one redacted public comment
- correspondence clarifying the OMIM applied for by the centre

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)

- HFEA decision trees
- 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

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 condition being applied for is not on the approved PGD condition list.

3. 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'.

4. The committee noted that Joubert syndrome 5 OMIM #610188 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.

5. The committee noted that the condition is a rare genetic disorder affecting the cerebellum, the area of the brain that controls coordination. Individuals can suffer from seizures, intellectual disabilities and progressive vision loss.

6. The committee noted that other symptoms that may develop are abnormal breathing patterns, lack of muscle control, sleep apnoea, decreased muscle tone, abnormal eye and tongue movements, cleft lip or palate, tongue abnormalities and extra fingers and toes.

7. The committee noted that the condition demonstrates complete penetrance and symptoms usually develop from birth.

8. The committee noted that there is no curative treatment for the condition and the only treatment options available are those to manage the symptoms that arise from the condition.

9. The committee noted that the application is consistent with the Peer Review.

10. The committee welcomed the advice of its advisor, Prof Peter Turnpenny, who confirmed that the condition is as described in the papers.

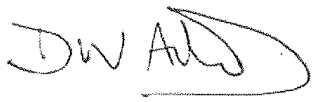
11. The committee welcomed the comment submitted by a member of the public.

12. The committee considered that the condition is serious due the associated symptoms, their early onset, and lack of any curable treatment.

13. 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.

14. The committee agreed to authorise the testing of embryos for Joubert syndrome 5 OMIM #610188.

Signed: Date: 12 June 2015



David Archard (Chair)