Human Fertilisation and Embryology Authority

Minutes of the Statutory Approvals Committee

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

Minutes – item 5

Centre 0102 (Guys Hospital) – PGD application for pyruvate dehydrogenase E1-alpha deficiency X-linked OMIM #312170

Members of the Committee	David Archard (Chair, lay) Sue Price (professional) Tony Rutherford (professional) Margaret Gilmore (lay)
Legal Adviser	Sarah Ellson, Fieldfisher
Specialist Attending	Professor Mary Porteous
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

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'. The committee noted that this was not an application for embryo testing to establish the sex of the embryo, under paragraph 1ZA(1)(c) of Schedule 2 to the Act.

4. The committee noted that pyruvate dehydrogenase E1-alpha deficiency X-linked OMIM #312170 is inherited in an X-linked pattern and there is a 1 in 2 chance of a male embryo being affected with the condition and a 1 in 2 chance of a female embryo being a carrier or affected where the mother is a carrier or affected by the condition. A female may be an asymptomatic carrier or can be as severely affected as a male. The committee noted that this was not an application for sex testing.

5. The committee noted that individuals with this condition form lactic acidosis. This can cause severe breathing problems, global developmental delay, and seizures. Abnormalities in the structure of the brain may be present with the wasting away of the cerebral cortex and incomplete development or total absence of the corpus callosum. Most males born with this condition do not survive beyond childhood.

6. The committee noted that other symptoms that may develop are an abnormal heartbeat, weak muscle tone, poor coordination, difficulties in walking, nausea and vomiting.

7. The committee noted that the condition demonstrates complete penetrance in males. The condition is very unpredictable and variable in females. Some symptoms are present from birth and others develop later in life.

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.

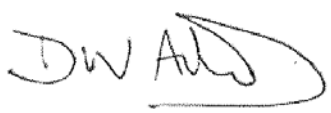
10. The committee welcomed the advice of its advisor, Professor Mary Porteous, who confirmed that the condition is as described in the papers and added some females can be as severely affected as males in inheriting the condition and displaying symptoms. Regard was had to the further information provided by the centre that described the unpredictability of how females may may be affected.

11. The committee considered that the condition is serious due to complete penetrance in males, the unpredictability in females, severity of the symptoms and the high likelihood of death in childhood.

12. 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. The committee noted that it was not approving embryo testing under paragraph 1ZA(1)(c) of Schedule 2 to the Act (i.e. in order to establish the sex of the embryo).

13. The committee agreed to authorise the testing of embryos for pyruvate dehydrogenase E1-alpha deficiency X-linked OMIM #312170.

Signed: Date: 12 May 2015



David Archard(Chair)