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 3

Centre 0102 (Guys Hospital) – PGD application for Leber congenital amaurosis types 3 – 17 OMIM #604232, #604393, #604537, #613826, #613829, #613835, #608553, #611755, #613837, #610612, #612712, #613341, #613843, #614186, #615360

Members of the Committee	David Archard (Chair, lay) Rebekah Dundas (Deputy Chair, lay) Sue Price (professional)
Legal Advisor	Dawn Brathwaite, Mills & Reeve
Specialist Advisor	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
- email clarification from the centre about the types to be included in this application

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 noted that the PGD application is for Leber congenital amaurosis types 3 to 17 inclusive. The centre's application form states all types of this condition are inherited in an autosomal recessive pattern.

2. The committee were made aware by the peer reviewer that types 7 and 11 of this condition are inherited in an autosomal dominant pattern. This was confirmed by the committee's advisor, Prof Peter Turnpenny, who added that Leber congenital amaurosis type 7 could be inherited in both an autosomal dominant and autosomal recessive pattern.

3. The committee received further advice from Prof Turnpenny that the dominant form of type 7 and type 11 can differ in significance and seriousness.

4. The committee, on considering the comments by the Peer Reviewer and advice of its advisor, decided that it would continue to consider the PGD application for types 3 to 6, 8 to 10, 12 to 17 and the autosomal recessive form of type 7. The Committee would not consider the autosomal dominant form of type 7 or type 11 as the members did not consider they had the evidence relating to the dominant types.

5. 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.

6. The committee noted that the conditions being applied for are not on the approved PGD condition list.

7. 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'.

8. The committee noted that Leber congenital amaurosis types 3 to 6, 8 to 10, 12 to 17, and the recessive form of type 7 are inherited in an autosomal recessive pattern and there is a 1 in 4 chance of an embryo being affected with the conditions where both parents are carriers.

9. The committee noted that the conditions are associated with abnormal retina and premature loss of light sensitive cells at the back of the eye before birth causing infants to be born with blind or with limited vision. Those born with limited vision may show progression to complete visual loss over time.

10. The committee noted that infants inheriting these conditions have a tendency to push, poke and rub their eyes that can cause further damage including developing sunken eyes.

11. The committee noted that the conditions demonstrate complete penetrance and signs are usually present from birth.

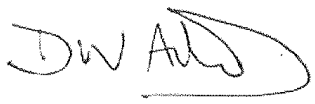
12. The committee noted that there is no curative treatment for the conditions and the only treatment options available are those to manage the symptoms that arise from the condition.

13. The committee considered that the condition is serious due to the extreme visual impairment with an early onset in infancy.

14. 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.

15. The committee agreed to authorise the testing of embryos for the autosomal recessive forms of Leber congenital amaurosis types 3 to 6, 8 to10, 12 to 17 inclusive (OMIMs #604232, #604393, #604537, #613826, 613835, #608553, #611755, #610612, #612712, #613341, #613843, #614186, #615360) and the form recessive form of type 7 (OMIM #613829). For the avoidance of doubt, the Committee did not agree to authorise the dominant form of type 7 and type11, but invited the centre to resubmit an application for these types with evidence based on the correct inheritance patterns of these types.

Signed: Date: 12 June 2015



David Archard (Chair)