Human Fertilisation and Embryology Authority

Minutes of the Statutory Approvals Committee

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

Minutes – Item 3

Centre 0102 (Guys Hospital) – PGD application for spinal muscular atrophy type 2 OMIM #253550 and type 3 OMIM #253400

Members of the Committee:	David Archard (Chair, lay) Jane Dibblin (lay) Sue Price (professional) Tony Rutherford (professional)
Legal Adviser:	Philip Grey, Mills and Reeve
Specialist Attending:	Dr Anne Lampe
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)
- 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

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 conditions being applied for are not on the approved PGD conditions 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 spinal muscular atrophy type 2 (OMIM #253550) and type 3 (OMIM #253400) are inherited in an autosomal recessive pattern and there is a 1 in 4 chance of an embryo being affected with one of the conditions where both parents are carriers.

5. The committee noted that individuals affected by these conditions develop progressive muscle weakness and reduced muscle tone which in its severe form can cause the inability to walk and the individual being confined to a wheel chair. More than thirty percent of individuals with spinal muscular atrophy type 2 will die before the age of twenty-five.

6. The committee noted that other symptoms that may develop are reduced respiratory capacity, pneumonia, chest infections, curvature of the spine, joint contractures and finger tremors.

7. The committee noted that the conditions demonstrate complete penetrance. If a couple have had an affected child, the symptoms of subsequent affected children they have together would be similar. For a couple who have not had an affected child together it can be very difficult to predict the severity of an

affected child that they might have. Symptoms may present from infancy to teenage years.

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

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

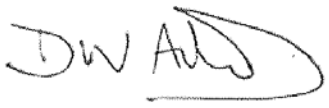
10. The committee welcomed the advice of its advisor, Dr Anne Lampe, who confirmed that the conditions were as described in the papers but highlighted that the executive summary in points 1.1, 2.1, 6.2 and 6.3 referred to incorrect OMIM numbers. In agreement with the peer reviewer she explained that rather than distinguishing four distinct categories of spinal muscular atrophy it is now recognised that there is a phenotypic continuum and early on during an infant or child's disease process it can be difficult to decide whether a child is best described as type 2 or type 3.

11. The committee considered that the conditions are serious due to the complete penetrance, lack of curative treatment, range of symptoms and the diminished life expectancy.

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 conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 to the Act.

13. The committee agreed to authorise the testing of embryos for spinal muscular atrophy types 2 (OMIM #253550) and type 3 (OMIM #253400).

Signed: Date: 12 March 2015



David Archard(Chair)