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 March 2015

Minutes – item 3

Centre 0102 (Guys Hospital) – PGD application for rigid spine muscular dystrophy type 1 OMIM #602771

Members of the Committee:	David Archard (Chair, lay) Sue Price (professional) Debbie Barber (professional) Tony Rutherford (professional)
Legal Adviser:	Dawn Brathwaite, Mills & 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
- supporting information provided 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)
- 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 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 rigid spine muscular dystrophy type 1 OMIM #602771 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 those affected by the condition have hypotonia, generalised muscle weakness, develop rigidity of the spine and scoliosis. Individuals develop early and life threatening respiratory problems with 50% of those affected needing night-time ventilation by the age of 15.

6. The committee noted that the condition demonstrates complete penetrance and is variable in the severity of symptoms and complications. Symptoms in one third of those affected are present from birth.

7. The committee noted that there is no curative treatment for the underlying condition and that the only treatment options available are those to manage the symptoms that arise such as surgery and use of a ventilator.

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

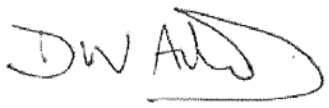
9. The committee welcomed the advice of its Advisor, Dr Anne Lampe, who confirmed that the condition is as described in the papers, stating that it is an extremely rare condition, and added that due to the requirement for long time ventilation there is an increased risk of death.

10. The committee considered that the condition is serious due the early onset of the condition and symptoms including respiratory issues that leads to the use of night-time ventilation in teenage years and the onset of scoliosis which may involve invasive surgery to manage.

11. 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.

12. The committee agreed to authorise the testing of embryos for rigid spine muscular dystrophy type 1 OMIM #602771.

Signed: Date: 13 April 2015



David Archard (Chair)