Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital) – PGD application for Hirschsprung Disease OMIM #142623

Thursday, 31 March 2016

HFEA, Finsbury Tower, 103-105 Bunhill Row, London, EC1Y 8HF

Committee members	David Archard (Chair) Anne Lamp Margaret Gilmore Anthony Rutherford	
Members of the Executive	Trent Fisher Ian Brown	Secretary Head of Corporate Governance
External adviser	Dr Alison Male	
Legal Adviser	Ros Foster	Browne Jacobson LLP
Observers	none	

Declarations of interest:

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

The committee had before it:

- 8th edition of the HFEA Code of Practice
- standard licensing and approvals pack for committee members

The following papers were considered by the committee:

- executive summary
- three supporting articles
- PGD application form
- redacted peer review
- Genetic Alliance opinion

1. Consideration of application

1.1. The committee welcomed the advice of its specialist advisor, Dr Alison Male, who confirmed that the condition is as described in the papers.

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

1.3. The committee noted that the application had previously been submitted to the Statutory Approvals Committee in March. The committee noted that the decision was to adjourn its decision on receipt of clarification and further information

1.4. The committee was now satisfied that they had sufficient clarification and further information from the centre and was confident that it would be able to make an informed decision

1.5. 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.

1.6. The committee noted that the condition being applied for is not on the approved PGD condition list.

1.7. 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'.

1.8. The committee noted that the condition is inherited in an autosomal dominant pattern and there is a 1 in 2 chance of an embryo inheriting the gene change where one parent is affected.

1.9. The committee noted that the condition affects the motility of the intestinal wall, primarily the colon, leading to bowel obstruction. Symptoms frequently present soon after birth.

1.10. The committee noted that symptoms include constipation, vomiting, abdominal pain and distention.

1.11. The committee noted that the severity of the condition depended on the length of the affected bowel wall. About 80 percent of individuals with this condition will present with a short segment disease and the other 20 percent will present with a severe form of the condition with a long, non-functioning stretch or involvement of the entire colon. In the most severe cases, the small bowel may also be affected.

1.12. The committee noted that the condition demonstrates a 65 percent penetrance in males and 45 percent penetrance in females. The committee noted further that the condition is extremely variable even within the same family.

1.13. The committee noted that the treatment of this condition is surgical and involves resection of the affected bowel wall. Depending on the extent and severity of the condition the surgery may need to be carried out in several stages and may involve a colostomy which may be lifelong in the most severe cases. In cases where total bowel resection is necessary a bowel transplant may be required. This is a high-risk surgical procedure. Affected individuals may have extended periods of hospitalisation and there is a significant risk of morbidity associated with this condition and complications following surgery.

2. Decision

2.1. The committee considered that the condition is serious due to the variability of the condition and the significant effect that the symptoms of the condition may have on the individual.

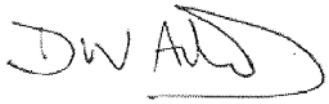
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious 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.

2.3. The committee agreed to authorise the testing of embryos for Hirschsprung Disease caused by a mutation in the RET gene.

3. Chair's signature

3.1. I confirm this is a true and accurate record of the meeting.

Signature



Name

David Archard

Date

14/04/2016