

HFEA Statutory Approvals Committee

19 December 2013

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

Minutes – Item 5

Centre 0102 (Guy’s Hospital) – PGD application for Diarrhoea 5 with Tufting Enteropathy, Congenital (OMIM #613217)

Members of the Committee: David Archard (lay) Chair Sue Price (professional) Hossam Abdalla (professional) Jane Dibblin (lay)	Committee Secretary: Lauren Crawford
Advisor: Dr Ed Blair	Legal Adviser: Graham Miles, Morgan Cole
	Observing: Sam Hartley, Head of Governance and Licensing, HFEA

Declarations of Interest: The Members declared no conflicts in relation to this item

The following papers were considered by the Committee

- Executive summary
- 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) (“the Act”)
- 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
- HFEA Pre-Implantation Diagnostic Testing (“PGD”) Explanatory Note For Licence Committee

Discussion

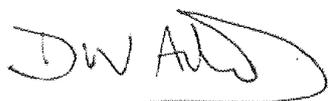
1. The Committee had regard to its Decision Tree. The Committee noted that the Centre is licensed to carry out PGD. The Committee was also satisfied that the Centre has experience of carrying out PGD and that generic patient information about its PGD programme and associated consent forms had previously been received by the HFEA.
2. The Committee noted that the 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’.
3. The Committee noted that Diarrhoea 5 with tufting enteropathy, congenital (OMIM #613217) 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.
4. The Committee noted that the condition causes severe chronic diarrhoea characterized by large amounts of watery stools which contain a high level of chloride salts. This results in dehydration, low levels of potassium and a higher level than normal of alkalis in the tissues. It is caused by diseased villi in the gut with characteristic ‘tufts’ on the surface of the gut lining.
5. The Committee noted that Diarrhoea 5 is fully penetrant and that affected children need total care day and night. They have a severely limited oral diet, need total parenteral nutrition, daily medication and care of their central line. They will suffer impaired growth and failure to thrive caused by an electrolyte imbalance.
6. The Committee noted that the only treatments available are total parenteral nutrition (TPN) and / or a bowel transplant.
7. The Committee noted that in cases where TPN is tolerated by the baby they can survive for some time although nutrition is rarely sufficient to meet full growth and developmental expectations. They may never eat food. They will

spend the majority of their time having nutrients pumped slowly into their blood stream. This method of feeding brings many risks. Infection of the intravenous access point is the primary cause of death in patients using parenteral feeding. Blood clots developing at the point of access may also result in death when they travel to the lungs. Liver failure is a major cause of death in these babies too.

8. The Committee noted that the application is supported by the Genetic Alliance UK and the Peer Reviewer.
9. The Committee welcomed the advice of its Advisor, Ed Blair, who confirmed that the condition was as described in the papers.
10. The Committee considered that the condition is serious because although growth development may be normal with TPN feeding this is not a straightforward option and leads to serious risk of infection and in some cases death.
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 Diarrhoea 5 with tufting enteropathy, congenital (OMIM #613217).

Signed:

Date: 08/01/2014

A handwritten signature in black ink, appearing to read 'DWA' followed by a stylized flourish.

David Archard (Chair)