Statutory Approvals Committee - minutes

Centre 0119 (Birmingham Women's Hospital) – PGD application for mitochondrial complex 1 deficiency OMIM #252010

Thursday, 17 December 2015

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

Committee members	David Archard (Chair) Rebekah Dundas (Deputy Chair) Sue Price Margaret Gilmore Anthony Rutherford Bishop Lee Rayfield	
Members of the Executive	Trent Fisher	Secretary
External adviser	Dr Edward Blair	
Legal Adviser	Graham Miles	Blake Morgan
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
- PGD application form
- redacted peer review
- redacted peer review with amendments
- Genetic Alliance opinion

1. Consideration of application

1.1. The committee noted that the centre has applied for mitochondrial complex 1 deficiency OMIM #252010 which can be caused by mutations in multiple different genes.

1.2. The committee's clinical adviser specified that the information located within the application related primarily to mitochondrial complex 1 deficiency caused by mutation in the NDUFS6 gene OMIM *603848 which was also affirmed by the peer reviewer.

1.3. The committee, in consultation with their legal adviser, decided that as the information within the application related to mitochondrial complex 1 deficiency caused by mutation in the NDUFS6 gene OMIM *603848, they would only be able to consider the application for this form of mitochondrial complex 1 deficiency.

1.4. 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.

1.5. The committee noted that mitochondrial complex 1 deficiency caused by mutation in the NDUFS6 gene OMIM #603848 is not on the approved PGD condition list.

1.6. 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'.

1.7. The committee noted that the condition 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.

1.8. The committee noted that the condition is a disorder affecting the ability of the body to produce energy via the respiratory chain in the mitochondria. This creates lactic acidosis, and often causes a progressive multisystem disorder with a wide range of clinical phenotypes. Children can have macrocephaly and with neurological presentations including progressive leukodystrophy, nonspecific encephalopathy or Leigh syndrome. It can affect muscle causing myopathy or hypertrophic cardiomyopathy. Some individuals will have optic atrophy or liver disease. Parkinson disease can be a more adult presentation.

1.9. The committee noted further that the condition can be lethal neonatally, present in childhood or with adult onset neurodegeneration.

1.10. The committee noted that the condition demonstrates a near complete penetrance. The condition also is extremely variable and it is near impossible to predict how severely a child will be affected but all individuals who carry the mutation are usually affected at some stage of their lives.

1.11. The committee noted that there is no curative treatment for the condition, but treatment that is purely symptomatic.

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

1.13. The committee welcomed the advice of its specialist advisor, Dr Edward Blair, who confirmed that the condition is as described in the papers.

2. Decision

2.1. The committee considered that the condition is serious due to progressive multi system disorders the condition causes and lack of any curable treatment.

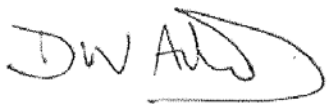
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious 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 mitochondrial complex 1 deficiency caused by mutation in the NDUFS6 gene OMIM #603848.

3. Chair's signature

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

Signature



Name

David Archard

Date

6 January 2016