Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital) – PGD application for SOPH Syndrome (Short stature, Optic nerve atrophy, Pelger-Huet anomaly) OMIM #614800

Thursday, 25 August 2016

HFEA, Level 2, 10 Spring Gardens, London, SW1A 2BU

Committee members	David Archard (Chair) Rebekah Dundas (Deputy Chair) Margaret Gilmore Anne Lampe Ruth Wilde	
Members of the Executive	Ian Brown Trent Fisher	Head of Corporate Governance Secretary
External adviser	Peter Turnpenny	
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
- PGD application form
- redacted peer review
- Genetic Alliance opinion

1. Consideration of application

1.1. The committee welcomed the advice of its specialist advisor, Professor Peter Turnpenny, 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 and the GA opinion paper.

1.3. 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.

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

1.5. 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'.

1.6. 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.

1.7. The committee noted that SOPH syndrome is an exceptionally rare condition caused by mutations in the NBAS gene. It causes severe postnatal growth retardation with abnormalities of the skeleton, unusual facial features with the appearance of premature aging, small hands and feet, an abnormality of the white blood cells and optic atrophy with loss of visual acuity and colour vision.

1.8. The committee further noted that patients may also have recurrent acute liver failure, which can be potentially fatal, and abnormalities of the cervical bones in the neck, leading to cervical instability, spinal cord compromise and paralysis.

1.9. The committee noted that liver crises are triggered by febrile infections; they become less frequent with age but are not restricted to childhood. Some patients who recover from the liver crises have been left with sequelae such as the consequences of brain haemorrhage with hemiparesis and epilepsy.

1.10. The committee noted that, individually, the symptoms of the condition are serious and as a whole it is an extremely debilitating condition.

1.11. The committee noted that the condition demonstrates complete penetrance however the clinical symptoms can be variable. Symptoms will be present at birth.

1.12. The committee noted that there is no curative treatment for the condition. The only available treatment is symptomatic. Some patients have required a liver transplant.

2. Decision

2.1. The committee considered that the condition is serious due to the wide range of severe symptoms and lack of any curable treatment.

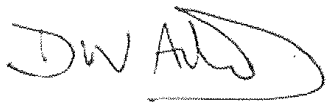
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious 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 SOPH Syndrome (Short stature, Optic nerve atrophy, Pelger-Huet anomaly) OMIM #614800.

3. Chair's signature

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

Signature



Name

David Archard

Date

8 September 2016