Statutory Approvals Committee - minutes

Centre 0044 (Centre for Reproductive and Genetic Health) – PGD application for Dyskeratosis Congenita types A2 and B4, due to TERT mutation OMIM #613989

Thursday, 28 April 2016

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

Committee members	David Archard (Chair) Rebekah Dundas (Deputy Chair) Margaret Gilmore Anthony Rutherford Ruth Wilde Anne Lampe	
Members of the Executive	Ian Brown Trent Fisher	Head of Corporate Governance Secretary
External adviser	Mary Porteous	
Legal Adviser	Philip Grey	Mills & Reeve
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
- OMIM website entry for #613989
- Genetic Alliance opinion

1. Consideration of application

1.1. The committee welcomed the advice of its specialist advisor, Professor Mary Porteous, who confirmed that the condition is as described in the papers adding that the age of onset can vary depending on the symptoms present.

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

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 dominant pattern and there is a 1 in 2 chance of an embryo being affected with the condition where one parent is affected.

1.7. The committee noted that the condition is a rare genetic form of bone marrow failure, the inability of the marrow to produce sufficient blood cells. The condition is diagnosed when patients present with the triad of abnormal skin, malformation of the nails, and white, thickened patches on the mucous membranes of the mouth (oral leukoplakia).

1.8. The committee noted that symptoms include pulmonary and liver fibrosis. Bone marrow failure may occur. In severe forms there may be learning difficulties, tooth loss, lack of blood supply to the hips, thickening of palmar skin and ridged finger nails, progressive bone marrow failure, narrowing of the oesophagus, severe diarrhoea and under-developed male genitalia. The skin changes may be present before the development of bone marrow failure.

1.9. The committee noted that other symptoms include short stature, eye and tooth abnormalities, thin and early greying of the hair, lung disease, liver disease, gut abnormalities, bone thinning, infertility, learning difficulties, and delays in reaching developmental milestones. An increased incidence of leukaemia and cancer has also been documented.

1.10. The committee noted, due to the condition being extremely rare, penetrance is hard to determine. The committee noted further that the condition is extremely variable from individuals presenting as asymptomatic to individuals being extremely handicapped by the condition.

1.11. The committee noted that there is no curative treatment for the condition and that the only treatment available is to manage the symptoms that present.

2. Decision

2.1. The committee considered that the condition is serious. While symptoms vary, in its more severe manifestations the condition would have a major impact on an individual's quality of life.

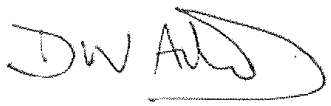
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious 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 Dyskeratosis Congenita types A2 and B4, due to TERT mutation OMIM #613989

3. Chair's signature

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

Signature



Name

David Archard

Date

13 May 2016