Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital) – PGD application for Cockayne syndrome type A and B OMIM #216400 #133540

Thursday, 25 February 2016

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

Committee members	David Archard (Chair) Anne Lampe Margaret Gilmore	
Members of the Executive	Trent Fisher	Secretary
External adviser	Professor John Walter	
Legal Adviser	Dawn Brathwaite	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
- redacted peer review
- Genetic Alliance opinion
- two redacted public comments
- email correspondence with the centre in response to the public comments

1. Consideration of application

1.1. The committee welcomed the advice of its specialist advisor, Professor John Walter, who confirmed that the condition is as described in the papers, subject to the comment at 1.2 below..

1.2. The committee noted that that, within the application, the centre had stated that Cockayne syndrome types A and B are also known as types 1 and 2. This was corrected by the peer reviewer. Two comments from members of the public also commented on the classification. With the advice of our specialist advisor the committee established that Cockayne Syndrome types A and B are clinically subdivided into type 1(classic or moderate form), type 2 (a more severe form) and type 3 (a milder form).

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 in each pregnancy of an embryo being affected with the condition where both parents are carriers of a relevant gene abnormality.

1.7. The committee noted that the condition is a very rare multisystem neurodegenerative disorder. In type 1, growth and developmental delay become noticeable during the first 2 years of life along with a characteristic facial appearance with sunken eyes and often marked photosensitivity. There is then a progressive course resulting in very short stature, delayed or absent sexual maturation, neurological problems, poor vision, hearing loss, intellectual disability and impaired renal and endocrine function. Death typically occurs in the first or second decade.

1.8. The committee noted that in type 2 growth failure is present at birth with little or no postnatal neurological development. Congenital cataracts may be present. Affected children have early postnatal contractures of the spine and joints. Death usually occurs by age seven years.

1.9. The committee noted that the condition demonstrates complete penetrance and symptoms can be present from birth.

1.10. The committee noted that there is no curative treatment for the condition, only treatment for the symptoms that present.

2. Decision

2.1. The committee considered that the condition is serious as the condition is a multisystem and debilitating condition that can be fatal.

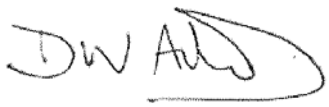
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious 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 Cockayne syndrome types A and B OMIM #216400, #133540.

3. Chair's signature

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

Signature



Name

David Archard

Date

10/03/2016