Statutory Approvals Committee - minutes

Centre 0035 (Oxford Fertility Unit) – PGD application for Achondrogenesis types 1a and 1b, OMIM #200600 and #600972

Thursday, 27 August 2015

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

Committee members	David Archard (Chair, lay) Rebekah Dundas (Deputy Chair, lay) Margaret Gilmore (lay) Sue Price (professional)	
Members of the Executive	Jo McAlpine	Secretary
External advisor	Professor Peter Turnpenny	
Legal Advisor	Tom Rider	Fieldfisher
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 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.

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

1.3. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(a) of schedule 2 of the Act, i.e. 'establishing whether the embryo has a gene, chromosome or mitochondrion abnormality that may affect its capacity to result in a live birth'.

1.4. 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 of a mutation in either the TRIP11 or SLC26A2 genes respectively.

1.5. The committee noted that the condition is a rare and severe genetic disorder in which all affected babies die either during pregnancy, shortly after birth or during early infancy.

1.6. The committee noted that Achondrogenesis is a severe, perinatally lethal skeletal dysplasia. There are 3 sub-types. Type 1A (ACG1A) and 1B (ACG1B) are often clinically and radiologically indistinguishable; both are autosomal recessive. ACG1A is caused by mutations in TRIP11. ACG1B is caused by severe mutations in SLC26A2. ACG1A and B usually present prenatally. They cause extremely short limbs, short hands and fingers, short trunk, short neck, small chest with short, thin ribs and a flat face. There is severely reduced ossification of the vertebrae. Hydrops can occur. The lungs are small. Babies with type 1A sometimes have rib fractures. Hands and fingers are sometimes less short in type 1A compared to 1B. The majority of affected babies die in utero or shortly after birth.

1.7. The committee noted that the condition demonstrates complete penetrance when a fetus inherits the two mutations.

1.8. The committee noted that there is only palliative care available for those who inherit this condition.

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

1.10. The committee welcomed the advice of its advisor, Professor Peter Turnpenny, who confirmed that the condition is as described in the papers.

2. Decision

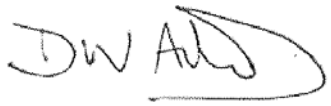
2.1. The committee was satisfied that the condition meets the criteria for testing under paragraph 1ZA(1)(a) of Schedule 2 to the Act.

2.2. The committee agreed to authorise the testing of embryos for Achondrogenesis types 1a and 1b, OMIM #200600 and #600972

3. Chair's signature

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

Signature



Name

David Archard

Date

10 September 2015