Statutory Approvals Committee - minutes

Centre 0035 (Oxford Fertility Unit) – PGD application for Spondyloepimetaphyseal dysplasia, Strudwick type, OMIM #184250

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) Anthony Rutherford (professional) 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)(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'.

1.4. 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.

1.5. The committee noted that the condition may result in individuals having an abnormally curved and shortened spine. This can cause progressive disability and compromise breathing. There is often instability of the spinal bones in the neck which creates a risk of spinal cord damage and paralysis, and for which surgery may be needed. Other joints are affected by the skeletal dysplasia leading to early arthritis. Vision is affected at a young age, with severe short sightedness and sometimes retinal detachment. Hearing may also be affected. Although intellect is normal, there is considerable and progressive physical disability.

1.6. The committee noted that the severity of symptoms for this type can be quite variable. The condition can result in severe short stature from birth, with a very short trunk and shortened limbs. Symptoms develop very early in life.

1.7. The Committee noted that this is a highly penetrant condition.

1.8. There are no treatments available for this condition and the only treatment options available are those to manage the symptoms that arise from the condition.

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

1.10. The committee welcomed the advice of its specialist advisor, Professor Peter Turnpenny, 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 the lack of curative treatment, and the risk of abnormally curved and shortened spine that can cause progressive disability and compromise breathing. The committee further considered it a serious condition due to the risk of spinal cord damage, and the effect on vision at a young age. The committee agreed that the described effect on the stature of patients would not in itself lead the committee to consider this a serious condition, but that the other symptoms and factors described above would.

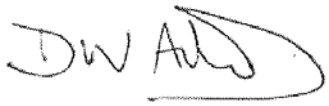
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious 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 Spondyloepimetaphyseal dysplasia, Strudwick type, OMIM #184250.

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