Statutory Approvals Committee - minutes

Centre 0102 (Guy's Hospital) – PGD application for Congenital Contractual Arachnodactyly (Beals Syndrome) OMIM #121050

Thursday, 24 September 2015
HFEA, Finsbury Tower, 103-105 Bunhill Row, London, EC1Y 8HF

Committee members	David Archard (Chair, lay) Rebekah Dundas (Deputy Chair, lay) Sue Price (professional) Margaret Gilmore (lay)	
Members of the Executive	Sam Hartley Trent Fisher	Head of Governance and Licensing Secretary
External adviser	Dr Anne Lampe	
Legal Adviser	Ros Foster	Browne Jacobson
Observers	Erin Barton	Projects, Inspections and Logistics Officer

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 to the Human Fertilisation and Embryology Act 1990 (as amended) ("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 risk of an embryo being affected with the condition where one parent is affected.

1.5. The committee noted that the condition is a multisystem disorder with individuals presenting with joint contractures, restricting movement of the hips, knees, ankle and elbows. Symptoms can also include long limbs and long slender fingers and toes. Some individuals will have a rounded upper back which will contribute to walking and movement difficulties. Heart problems may occasionally occur, in particular aortic root dilatation where individuals can tear their dilated aortic root causing a life threating situation that requires emergency surgery.

1.6. The committee noted that in a rare, severe form of this condition infants have significant joint contractures, gastrointestinal and heart congenital malformations with a high risk of death in infancy.

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

1.8. The committee noted that there is no curative treatment for this condition and the only treatment available is to treat the symptoms that arise. This can include invasive traumatic surgery and individuals will require regular hospital visits.

1.9. The committee noted that the Peer Review does support the application however the Peer Review does not make reference to eye problems such as short-sightedness and risk of retinal detachment.

1.10. The committee welcomed the advice of its specialist advisor, Dr Anne Lampe, who confirmed that the condition is as described in the Peer Review and that due to the contractures in the joints it can be a very disabling condition as individuals can have very restricted movement. Dr Lampe also pointed out that the description of the condition contained at paragraph 4.2 of the Executive Summary and the lay summary of the application was inaccurate with respect to the references to 'eye problems' in that these are not recognised to be part of the condition

2. Decision

2.1. The committee considered that the condition is serious due to the disabling restriction of movement which impacts on quality of life and lack of any curative treatment available.

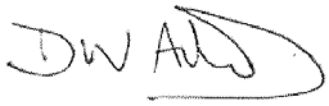
2.2. The committee had regard to its explanatory note and on the basis of the information presented, given the condition's worst symptoms, t was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness 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 Congenital Contractual Arachnodactyly (Beals Syndrome) OMIM #121050

3. Chair's signature

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

Signature



Name

David Archard

Date

8 October 2015