Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital) – PGD application for Facioscapulohumeral Muscular Dystrophy Type 2 OMIM #158901

Friday, 24 June 2016

HFEA, Level 2, 10 Spring Gardens, London, SW1A 2BU

Committee members	David Archard (Chair) Rebekah Dundas (Deputy Chair) Margaret Gilmore Anthony Rutherford Anne Lampe Ruth Wilde	
Members of the Executive	Ian Brown Trent Fisher	Head of Corporate Governance Secretary
External adviser	Jenny Carmichael	
Legal Adviser	Dawn Brathwaite	Mills & Reeve LLP
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
- two supporting research publications submitted with the application
- redacted peer review
- Genetic Alliance opinion
- Licence Committee minutes from 14 April 2008 which approved Facioscapulohumeral Dystrophy OMIM #158900.

1. Consideration of application

1.1. The committee noted that the papers contained Statutory Approvals Committee minutes concerning a previous condition which had been authorised for PGD. The committee's legal adviser confirmed that the committee is not bound by previous decisions made and that it needed to be satisfied that the particular risk and significant risks tests were met for each condition on the paperwork and advice provided.

1.2. The committee welcomed the advice of its specialist advisor, Dr Jenny Carmichael, who confirmed that the condition is as described in the papers with the exception of the explanation of the inheritance pattern within the application form. The inheritance pattern was correctly described in the executive summary which was confirmed by the specialist adviser and the peer review for the committee.

1.3. Dr Carmichael added further that due to the muscle weakness individuals with this condition may be unable to close their eyelids which can cause corneal ulcers and erosion. As the muscle weakness affects the facial muscles it can be difficult for individuals to express emotion as their face takes on a mask like appearance.

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

1.5. The committee noted the Genetic Alliance opinion which helped the committee gain an understanding of the condition from the patient perspective.

1.6. 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.

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

1.8. 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'.

1.9. The committee noted that the condition requires a mutation in the gene SMCHD1 and either one or two copies of a permissive haplotype at locus D4Z4 (located at chromosome 4q35). This effectively means that the recurrence risk is between 25-50% because an affected person has one mutated copy of SMCHD1 and either one or two copies of the 4q35 permissive haplotype, as may their partner.

1.10. The committee noted that the condition affects a wide range of muscles which includes muscles of the face, shoulder blades, upper arms and lower legs. The condition is progressive

1.11. The committee noted as a result of the muscle weakness a quarter of affected individuals will require use of a wheelchair within 6 years of diagnosis. Other associated problems include loss of ability to lift the arms above the shoulders and therefore loss of ability for self-care, as well as loss of ability to lift objects, curvature of the spine, pain in the muscles or joints, abnormal development of the blood vessels of the retina, breathing problems and moderate to severe hearing loss.

1.12. The committee noted that the condition is likely to demonstrate a very high penetrance by late adult life. The onset of symptoms is typically in the teenage years however there are reported cases on symptoms developing in infancy.

1.13. The committee noted that there is no curative treatment for the condition. The committee noted that physiotherapy and occupational therapy may help to retain function for longer.

2. Decision

2.1. The committee considered that the condition is serious due to severity of symptoms and intensive treatment needed.

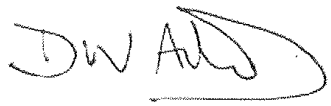
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious 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 Facioscapulohumeral Muscular Dystrophy Type 2 OMIM #158901.

3. Chair's signature

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

Signature



Name

David Archard

Date

11 July 2016