Statutory Approvals Committee – minutes

Centre 0119 (Birmingham Women's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Myotonia Congenita, Autosomal Dominant, OMIM #160800 and Myotonia Congenita, Recessive, OMIM #255700

Thursday, 30 April 2020

HFEA Teleconference Meeting

Committee members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr Jenny Carmichael	
Legal Adviser	Graham Miles	Blake Morgan - LLP
Observer	Ermal Kirby	Authority Member

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:

- 9th 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 UK Statement

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr Jenny Carmichael, who confirmed that the conditions were as described in the papers.

1.2. The committee noted that the description in the PGD application for Myotonia Congenita, Autosomal Dominant, OMIM #160800, and Myotonia Congenita, Autosomal Recessive, OMIM #255700, are consistent with the peer review.

1.3. The committee noted that the conditions being applied for are not on the list of approved PGD conditions.

1.4. The committee noted that the Genetic Alliance UK statement provided a perspective on the impact of the condition on patients, their families and carers.

1.5. 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.

1.6. 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'.

1.7. The committee noted Myotonia Congenita, Autosomal Dominant, OMIM #160800, is inherited in an autosomal dominant manner which means there is a 50% chance of having an affected child in each pregnancy if either parent has a relevant mutation. Myotonia Congenita, Autosomal Recessive, OMIM #255700, is inherited in an autosomal recessive manner, which means there is a 25% chance of an embryo being affected by the condition in each pregnancy if each parent has a relevant mutation.

1.8. The committee noted Myotonia Congenita, Autosomal Recessive, OMIM #255700, is likely to be completely penetrant. The penetrance of Myotonia Congenita, Autosomal Dominant, OMIM #160800, is unknown.

1.9. Both types of Myotonia Congenita affect skeletal muscles with onset in infancy or early childhood and manifest as periods of muscle stiffness and myotonia (the condition where relaxation of a muscle is impaired). Symptoms can be aggravated by exposure to cold temperatures. In females, symptoms are exacerbated during menstruation and pregnancy. In some of those affected, symptoms can be severe and result in muscle weakness, pain and reduced quality of life. Transient muscle weakness will occur in 75% of patients which can lead to falls and other accidents. Difficulties with hand grip can cause difficulties with daily life. Eye movement abnormalities can affect vision and tongue involvement can affect speech and swallowing.

1.10. There is no cure for these conditions. Medical treatment is available to help manage the symptoms but these are not without side-effects.

1.11. The committee noted the executive's request to consider for Myotonia Congenita, Autosomal Dominant, OMIM #160800, and Myotonia Congenita, Autosomal Recessive, OMIM #255700, for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

1.12. The committee noted the recommendation of the executive to consider a further similar condition type for inclusion on the list of conditions approved for PGD. The condition Paramyotonia Congenita of von Eulenburg (PMC), OMIM #168300, is characterised by spells of muscle stiffness and myotonia, aggravated by exposure to cold temperatures and after physical activity. The peer reviewer considers the condition to be of a similar phenotype, with equivalent, or more severe symptoms to Myotonia Congenita, Autosomal Dominant, OMIM #160800, and Myotonia Congenita, Autosomal Recessive, OMIM #255700. The condition is inherited in an autosomal dominant manner.

2. Decision

2.1. The committee considered that, in the worst-case scenario Myotonia Congenita, Autosomal Dominant, OMIM #160800, and Myotonia Congenita, Autosomal Recessive, OMIM #255700, are rare, debilitating and severely painful conditions that usually present in infancy. The conditions interfere with movement and may be severe enough to affect walking, running and other daily activities of everyday life. The committee noted the possible psychological and physical impact on the quality of life for those affected with the conditions.

2.2. The committee considered in the worst-case scenario the condition Paramyotonia Congenita of von Eulenburg OMIM #168300, was of a similar phenotype and presentation to Myotonia Congenita, Autosomal Dominant, OMIM #160800, and Myotonia Congenita, Autosomal Recessive, OMIM #2557003, but is often more severe with more frequent pain and weakness.

2.3. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormalities in question and that there is a significant risk that a person with such an abnormality will, given the conditions' worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

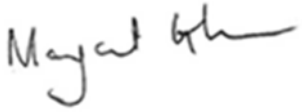
2.4. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Myotonia Congenita, Autosomal Dominant, OMIM #160800
- Myotonia Congenita, Autosomal Recessive, OMIM #255700
- Paramyotonia Congenita of von Eulenburg OMIM #168300

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
20 May 2020