Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Paramyotonia Congenita (PMC), OMIM #168300

Thursday, 25 May 2017

Church House Westminster, Dean's Yard, Westminster SW1P 3NZ

Committee members	Margaret Gilmore (Chair) Anne Lampe Ruth Wilde Anthony Rutherford Bobbie Farsides	
Members of the Executive	Bernice Ash Paula Robinson	Secretary Head of Planning & Governance
External adviser	Dr Jenny Carmichael	
Legal Adviser	Sarah Ellson	Fieldfisher
Observers		

Declarations of interest

- Members of the panel 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 welcomed the advice of its specialist adviser, Dr Jenny Carmichael, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Paramyotonia Congenita (PMC), OMIM #168300 is consistent with the Peer Review.

1.3. The committee noted that the Genetic Alliance opinion provided a patient perspective and supported the application.

1.4. 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.

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

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 that the condition is inherited in autosomal dominant manner which means there is 50% chance of having an affected child in each pregnancy, if either parent has a relevant mutation.

1.8. Paramyotonia Congenita (PMC) is a very variable condition (both inter and intra-familial variation). Onset can be in the neonatal period, with hypotonia, laryngospasm (floppy larynx) and feeding difficulties, but it usually presents in early childhood, or occasionally much later. It is characterised by myotonia (sustained muscle contraction and inability to relax muscles) which can be painful, sometimes severely so, muscle weakness and stiffness. The face, neck, arms and hands are usually most affected, but other muscles, including respiratory muscles, can be involved.

1.9. Myotonia is frequently precipitated by cold. Other precipitating factors can include exercise, hunger, menstruation and pregnancy. Unlike congenital myotonia, there isn’t usually the ‘warm-up phenomenon' where muscle stiffness improves with exercise; frequently the opposite occurs. Muscle weakness can last hours to days and in older severely affected people can be permanent. There is often muscle hypertrophy (enlarged muscles).

1.10. The committee noted the condition is usually non-progressive. There is a risk of malignant hyperthermia with anaesthetics, which is life threatening, but if the risk is known in advance this can be prevented by using different anaesthetics agents and muscle relaxants. Penetrance is thought to be high but not complete.

1.11. Some patients get marked improvement from Mexiletine (an oral antiarrhymia drug), but not all respond. Side effects can occur, including abdominal pain, nausea, tremor and ataxis, but these usually resolve with dose reduction. Aniteplieptics including carbamazepine, sodium valproate and gabapentin are beneficial in some patients. In some patients, the symptoms are mild so treatment is not required. There is no cure, but many people live a near-normal lifestyle taking care with temperature, exercise, diet and sometimes medication.

1.12. The committee noted the inspectorate’s recommendation to consider the approval of Paramyotonia Congenita (PMC), OMIM #168300 to be included on the PGD List. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee noted that some patients get marked improvements from treatment and may lead a near normal lifestyle, but in most scenarios it considered Paramyotonia Congenita (PMC), OMIM #168300 is serious given the unpredictable onset and the effect on the quality of life including severe debilitating pain, physical symptoms, breathing difficulties, long periods of paralysis and lack of a cure. The committee considered the impact on the individuals' quality of life and the family caring for affected individuals.

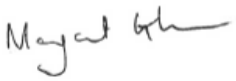
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical disability which may have a secondary effect on mental health. The committee was therefore satisfied that the condition of Paramyotonia Congenita (PMC), OMIM #168300 does meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

2.3. The committee agreed to authorise the testing of embryos for Paramyotonia Congenita (PMC), OMIM #168300.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

8 June 2017