Statutory Approvals Committee – minutes

Centre 0119 (Birmingham Women's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Merosin Deficient Congenital Muscular Dystrophy Type 1A, OMIM #607855

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 Merosin Deficient Congenital Muscular Dystrophy type 1A, OMIM #607855 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 an autosomal recessive manner which means there is 25% chance of having an affected child in each pregnancy, if each parent has a relevant mutation.

1.8. Merosin Deficient Congenital Muscular Dystrophy type 1A (MDC1A), causes weakness and wasting (also known as atrophy) of the skeletal muscles used for movement. The condition can appear as a severe, early-onset type which is apparent at birth, or a milder, late-onset form which manifests later in childhood.

1.9. The committee noted that early-onset affected infants have severe muscle weakness and joint contractures which can cause feeding problems, problems with speech and respiratory problems which can lead to life-threatening lung infections. Affected children cannot walk unassisted and often develop curvatures of the spine. Affected individuals usually do not survive past adolescence. Penetrance is thought to be 100%.

1.10. Children with late-onset disease may show delayed walking or delay of other motor-skills but are generally able to walk without assistance. In time, those with late-onset disease may show other features such as joint contractures, breathing problems, and scoliosis. However, they will usually remain mobile and their life expectancy is not compromised.

1.11. The committee noted there is currently no treatment available for this condition.

1.12. The Committee noted the PRs comments that the condition is also known as LAMA2-related Muscular Dystrophy and the inspectorate's suggestion that the condition be added to the HFEA website as Merosin Deficient Congenital Muscular Dystrophy type 1A (MDC1A) (also known as LAMA2-related Muscular Dystrophy).

1.13. The committee noted the inspectorate's recommendation to consider the approval of Merosin Deficient Congenital Muscular Dystrophy type 1A (MDC1A), OMIM #607855 to be included on the PGD List, named as Merosin Deficient Congenital Muscular Dystrophy type 1A (MDC1A) (also known as LAMA2-related Muscular Dystrophy), OMIM #607855 which is in line with the information published by OMIM. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered Merosin Deficient Congenital Muscular Dystrophy type 1A (MDC1A) (also known as LAMA2-related Muscular Dystrophy), OMIM #607855 is serious given the early onset and effects of this condition, including severe muscle weakness and wasting (atrophy), life-threatening lung infections and the lack of a cure. The Committee noted, in its worst case scenario, the condition brings early death and most people who have early onset will never walk. The committee considered the impact on the individuals' quality of life and the family caring for affected individuals.

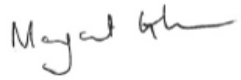
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition of Merosin Deficient Congenital Muscular Dystrophy type 1A (MDC1A) (also known as LAMA2-related Muscular Dystrophy), OMIM #607855 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 Merosin Deficient Congenital Muscular Dystrophy type 1A (MDC1A) (also known as LAMA2-related Muscular Dystrophy), OMIM #607855 and agreed that the condition should be included on the PGD List on the HFEA website as Merosin Deficient Congenital Muscular Dystrophy type 1A (MDC1A) (also known as LAMA2-related Muscular Dystrophy), OMIM #607855.

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