Statutory Approvals Committee – minutes

Centre 0044 (The Centre for Reproductive and Genetic Health) Pre-implantation Genetic Diagnosis (PGD) application for Bailey-Bloch Congenital Myopathy (BBCM), OMIM #255995

Thursday, 13 December 2018

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe Anthony Rutherford Emma Cave	
Members of the Executive	Dee Knoyle Catherine Burwood Paula Robinson	Committee Secretary Senior Governance Manager (Observer) Head of Planning and Governance (Observer)
Legal Adviser	Eve Piffaretti	Blake Morgan LLP
External adviser	Dr Jenny Carmichael	
Observers		

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 UK statement
- Supporting paper, Stamm et al. 2008
- Supporting paper, Stewart et al 1998
- Supporting paper, Telegrafi et al 2017
- Supporting paper, Grzybowski et al 2017

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 application for PGD for Bailey-Bloch Congenital Myopathy (BBCM), OMIM #255995 is consistent with the peer review.

1.3. The committee noted that the OMIM website lists the condition as Myopathy, Congenital, Bailey-Bloch and Native American Myopathy is listed as an alternative name. However, the condition is no longer considered limited to the Lumbee Native American population.

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

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

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 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'.

1.8. The committee noted that the condition is inherited in an autosomal recessive pattern which means there is a 25% chance of an embryo being affected with the condition in each pregnancy, if both parents have a relevant mutation. The condition is completely penetrant.

1.9. The committee noted that BBCM is a rare condition, caused by mutations in the STAC3 gene. The condition is characterised by congenital muscle weakness, arthrogryposis, cleft palate, ptosis, myopathic facies, short stature, kyphoscoliosis, clubfoot malformation, susceptibility to malignant hyperthermia provoked by anaesthesia, oral hypotonia and poor feeding, which has led to a tracheostomy and feeding problems. A small case series study in 2008 (Stamm et al) suggested premature death, before the age of 18 years in 36% of those affected, and for those living with the condition it may have a major effect on the quality of life due to complex musculoskeletal issues, progressive muscle weakness and feeding problems.

1.10. The committee noted that treatment to manage some of the symptoms of the condition is available but there is no curative treatment. Management of symptoms may include mobility aids, a tracheostomy, feeding tubes, and cleft palate repair.

1.11. The committee noted the inspectorate's request to consider whether Bailey-Bloch Congenital Myopathy (BBCM), OMIM #255995 should be approved for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

1.12. The inspectorate also suggested that the condition is referred to as Myopathy, Congenital, Bailey-Bloch, OMIM #255995, as per the OMIM website, or Bailey-Bloch Congenital Myopathy (BBCM), OMIM #255995.

2. Decision

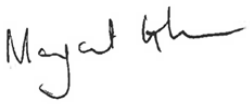
2.1. The committee considered that, in the worst case scenario Bailey-Bloch Congenital Myopathy (BBCM), OMIM #255995 is a serious progressive disabling disease. The condition is present from birth and causes muscle weakness, feeding difficulties, breathing problems and developmental delay affecting motor skills. Affected individuals have joint contractures presenting as clubfoot, abnormal positioning of the hands and abnormal curve of the spine. Hearing may be affected too. Some individuals may be tube fed through to teenage years. The condition requires invasive treatment including surgery to correct the congenital anomalies or to insert a feeding tube and affected individuals may have a severe reaction to particular medications used during general anaesthesia which could be life-threatening. There is no cure for this condition and death could occur during infancy.

2.2. 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 abnormality in question and that there is a significant risk, given the condition's worst symptoms, 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 Bailey-Bloch Congenital Myopathy (BBCM), OMIM #255995 meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act and agreed to authorise testing.

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

17 January 2019