Statutory Approvals Committee – minutes

Centre 0102 (Guy's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Mitochondrial Myopathy and Ataxia (MMYAT), OMIM #617675

Thursday, 25 October 2018

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe Anthony Rutherford Ruth Wilde	
Members of the Executive	Bernice Ash Dee Knoyle Paula Robinson Catherine Burwood	Committee Secretary Committee Secretary (Observer) Head of Planning and Governance (Observer) Senior Governance Manager (Observer)
Specialist Adviser	Professor Mary Porteous	
Legal Adviser	Gerard Hanratty	Browne Jacobson LLP
Observers	Emma Cave	(New Member Induction)

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
- Nasca et al 2017 (Human mutation) paper
- Gal et al 2017 paper

1. Consideration of application

1.1. The committee welcomed the advice of its Specialist Adviser, Professor Mary Porteous 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 Mitochondrial Myopathy and Ataxia (MMYAT), OMIM #617675 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 in families studied by Gal et al. (2017), 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.

1.8. The committee noted that, a family was reported by Nasca et al. (2017), where the condition is thought to be inherited in an autosomal dominant pattern which would mean there would be a 50% chance of an embryo being affected with the condition in each pregnancy, if either parent has a relevant mutation. The Specialist Adviser concurred with the Peer Reviewer that currently no convincing data exists for a dominantly inherited form of this condition.

1.9. The committee noted that the penetrance of the autosomal recessive condition is 100%. The condition causes, from early in life, muscle weakness and wasting, poor growth, delayed motor development, impaired ability to perform rapid / alternating movements, lack of coordination and additional neurologic features. Some patients show skeletal and endocrine anomalies. The condition can lead to hearing and sight loss later in life. MMYAT may impact significantly on the quality of life of those affected. There are no curative treatments.

1.10. The committee noted the inspectorate's request to consider whether Mitochondrial Myopathy and Ataxia (MMYAT), OMIM #617675, should be approved for inclusion on the PGD List. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that, in the worst case scenario, Mitochondrial Myopathy and Ataxia (MMYAT), OMIM #617675 (autosomal recessive), is serious as the condition has an early onset, is multi-systemic and severely debilitating, causing motor development and mobility issues. The condition may severely impact on the individual's quality of life and the family.

2.2. The committee decided not to authorise testing for the autosomal dominant inherited form of Mitochondrial Myopathy and Ataxia (MMYAT), OMIM #617675, noting the advice provided by the Specialist Advisor regarding the deficiency of available data, as there is currently not enough information on which to make the decision.

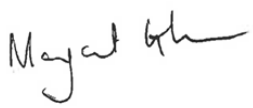
2.3. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, given the condition's worst symptoms, that a 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 the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act and agreed to authorise testing:

- Mitochondrial Myopathy and Ataxia (MMYAT) OMIM #617675 (**autosomal recessive**)

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

27 November 2018