Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Familial Infantile Myoclonic Epilepsy (FIME), OMIM #605021

Thursday, 12 December 2019

HFEA, Spey Meeting Room, Level 2, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Anne Lampe Emma Cave Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Professor Peter Turnpenny	
Legal Adviser	Sarah Ellson	FieldFisher - LLP
Observer	Souhaila Cherkaoui	Inspections and Logistics Officer (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:

- 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
- Supporting Statement from patient couple with an affected child
- Statutory Approvals Committee Minutes - PGD application for Early Infantile Epileptic Encephalopathy 28 (EEIE-28), OMIM 616211 (includes approval for EIEE-16)

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Professor Peter Turnpenny, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Familial infantile myoclonic epilepsy (FIME), OMIM #605021, is consistent with the peer review.

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

1.4. The committee noted that the Genetic Alliance UK were requested to provide a perspective on the impact of the condition on patients, their families and carers, but unfortunately did not respond within the allocated deadline for their statement to be considered.

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 that Familial infantile myoclonic epilepsy (FIME), OMIM #605021, is inherited in an autosomal recessive pattern, 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 the penetrance of the condition is 100%.

1.9. Familial infantile myoclonic epilepsy (FIME), OMIM # 605021, is characterised by early onset myoclonic seizures, febrile convulsions and tonic/clonic seizures, of variable duration and frequency. More than half of those affected will have drug resistant epilepsy making the condition very difficult to treat. Some children may also demonstrate significant intellectual disability and developmental delay. The condition can have a significant impact on quality of life and may cause death in the first years of life.

1.10. There is no cure for this condition.

1.11. The committee noted the executive's request to consider Familial infantile myoclonic epilepsy (FIME), OMIM #605021, for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that, in the worst-case scenario Familial infantile myoclonic epilepsy (FIME), OMIM #605021, is a rare and very serious condition. The committee noted that over half of those affected are likely to have drug resistant epilepsy which makes this condition very difficult to control. Seizures may lead to death in infancy. The committee noted the serious implications and impact on the quality of life for those affected with the condition.

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 abnormalities in question and that there is a significant risk that a person with such an abnormality will, given the condition's worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

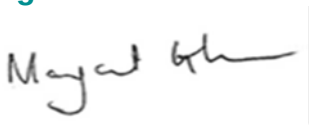
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Familial infantile myoclonic epilepsy (FIME), OMIM #605021

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
6 January 2020