Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Inclusion body myopathy and early-onset Paget disease and frontotemporal dementia 1 (IBMPFD1)

OMIM #167320

Thursday, 31 October 2019

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

Committee members	Margaret Gilmore (Chair) Rachel Cutting Emma Cave	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Professor Mary Porteous	
Legal Adviser	Ros Foster	Browne Jacobson LLP
Observer	Alistair Robertson	DAC Beachcroft LLP

Declarations of interest

- Members of the committee declared that they had no conflicts of interest in relation to this item.

Apologies:

- Apologies were received from Tony Rutherford and Bobbie Farsides.

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
- Genetic Alliance UK statement
- Comment has been received from the public regarding IBMPFD1

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 PGD application for Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1 (IBMPFD1), OMIM #167320, 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 statement provided a perspective on the impact of the condition on patients, their families and carers.

1.5. The committee noted and welcomed the personal representations received from the public regarding the impact of the condition.

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 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1 (IBMPFD1), OMIM #167320 is inherited in an autosomal dominant pattern, which means there is a 50% chance of an embryo being affected by the condition in each pregnancy if either parent has a relevant mutation.

1.9. The committee noted the penetrance of the condition is not known; however, it is high and thought to be around 90%.

1.10. The condition is characterised by myopathy leading to muscle weakness, cardiac failure, bone pain, bone fracture, lack of inhibition, loss of empathy, inability to recall words and loss of numerical skills. With BMPFD1, around 90% of people will experience progressive myopathy, 50% will develop Paget disease and 33% will develop frontotemporal dementia. While the severity can be variable, all symptoms of the condition have a significant impact on the quality of life. All of the features of the condition begin in adulthood and from the 30s onwards. Pain and weakness associated with the skeletomuscular symptoms can be significantly debilitating for patients, and can significantly impact the quality of life. They can only be partially treated with medications, and are not curative. The condition is life-limiting, with the average life expectancy being in the 50s or 60s.

1.11. The committee noted the executive's request to consider Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1 (IBMPFD1), OMIM #167320 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 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1 (IBMPFD1), OMIM #167320 is a rare, degenerative and life limiting condition with onset in adulthood. The condition can be associated with severe pain from overgrowth of weak bones and treatment options to manage pain are limited. There is no cure for the condition with death occurring within 10-15 years of onset due to a range of issues. The committee considered the possible impact on the quality of life for patients living with the condition and in particular noted the psychological impact on those who may live with the fear of developing a particularly damaging form of dementia. The committee took into account the information received from the public in relation to the application.

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 conditions' worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

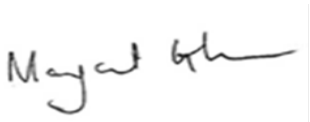
2.3. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1 (IBMPFD1), OMIM #167320

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
18 November 2019