Statutory Approvals Committee – minutes

Centre 0035 (Oxford Fertility)

Pre-implantation Genetic Diagnosis (PGD) application for Benign Chronic Pemphigus (BCPM) also known as Hailey-Hailey Disease (HHD), OMIM #169600

Thursday, 30 April 2020

HFEA, HFEA Teleconference Meeting

Committee members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr Jenny Carmichael	
Legal Adviser	Graham Miles	Blake Morgan - LLP
Observer	Ermal Kirby	Authority Member

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
- Genetic Alliance UK Statement

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 Benign Chronic Pemphigus (BCPM), OMIM #169600, 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 Benign Chronic Pemphigus (BCPM), OMIM #169600 is also known as Hailey-Hailey Disease. To ensure consistency with the OMIM website, the condition for the purposes of this application, will be known as Benign Chronic Pemphigus (BCPM), OMIM #169600.

1.5. The committee noted that the Genetic Alliance UK statement provided a perspective on the impact of the condition on patients, their families and carers.

1.6. 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 Benign Chronic Pemphigus (BCPM), OMIM #169600, is inherited in an autosomal dominant manner which means there is 50% chance of having an affected child in each pregnancy if either parent has a relevant mutation.

1.9. The committee noted the penetrance of the condition is 100%.

1.10. Benign Chronic Pemphigus (BCPM), OMIM #169600, is characterised by blisters and erosions of the skin most often affecting the neck, armpits, groin, perianal region, genitals, and other skin folds. These can present in adolescence or early adulthood. The lesions may come and go repeatedly, are prone to secondary infections and can have a significant impact on a patient's quality of life.

1.11. There is no cure for this condition and treatment focuses on managing the symptoms or preventing an exacerbation of the condition.

1.12. The committee noted the executive's request to consider Benign Chronic Pemphigus (BCPM), OMIM #169600, 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 Benign Chronic Pemphigus (BCPM), OMIM #169600, is a rare skin disorder that usually presents in adolescence or early adulthood with relentless and painful skin lesions/ blisters. These can often become susceptible to secondary infections which can be resistant to treatment and in some cases, treatment can result in scarring. The committee considered the possible serious adverse physical and psychological impact on the quality of life for patients living with this 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.

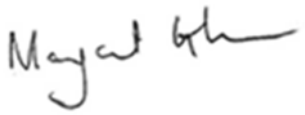
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Benign Chronic Pemphigus (BCPM) OMIM #169600

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
20 May 2020