Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital)

Preimplantation Genetic Diagnosis (PGD) application for D-bifunctional protein deficiency, OMIM #261515

Date:	25 February 2021
Venue:	Microsoft Teams Meeting
Committee Members:	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde
Specialist Adviser:	Alan Fryer
Legal Adviser:	Sarah Ellson - FieldFisher LLP
Members of the Executive:	Moya Berry - Committee officer Catherine Burwood - Licensing Manager
Observers:	Sarah Steadman - Inspector (Induction) Karen Campbell - Inspector (Induction)
Apologies:	No apologies were received for the meeting
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:

- HFEA Code of Practice 9th edition
- Standard Licencing and Approvals Pack

The following papers were considered by the committee:

- Executive Summary
- PGD Application form
- Redacted Peer review
- 2011-09-29 Licence Committee Minutes, PGD for Peroxisome Biogenesis Disorders PBD (Zellweger Syndrome Spectrum ZSS)

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr Alan Fryer, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for D-bifunctional protein deficiency, OMIM #261515 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 a Genetic Alliance (UK) statement had not been provided for this application.

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 D-bifunctional protein deficiency, OMIM #261515, is inherited in an autosomal recessive manner, 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 that the penetrance of the condition is 100%.

1.9. D-bifunctional protein deficiency, OMIM #261515, is characterised by neonatal onset of hypotonia and severe and recurrent seizures, poor vision, and hearing impairment. Hardly any of these children show any developmental progress. This is a progressive disorder and most of those affected die in infancy or very early childhood.

1.10. There is no cure for this condition and treatment is focused on improving nutrition and growth, controlling symptoms, and limiting the progression of liver disease.

1.11. The committee noted the executive's request to consider D-bifunctional protein deficiency, OMIM #261515, 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, D-bifunctional protein deficiency, OMIM #261515, is a rare, degenerative condition leading to severe developmental delay, with additional features such as progressive vision loss, deafness, and intractable seizures. There is no cure for condition and most children die in infancy or by early childhood. The committee considered the devastating impact on the quality of life of those affected.

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 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.

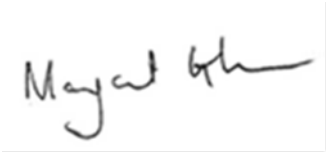
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- D-bifunctional protein deficiency, OMIM #261515

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

17 March 2021