Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital)

Preimplantation Genetic Diagnosis (PGD) application for Transcobalamin II Deficiency, OMIM #275350

Date:	27 May 2021
Venue:	Microsoft Teams Meeting
Committee Members:	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde
Specialist Adviser:	Alison Male
Legal Adviser:	Graham Miles – Blake Morgan LLP
Members of the Executive:	Moya Berry - Committee officer Catherine Burwood - Licensing Manager
Observers:	Dina Halai- Senior Scientific Policy Manager HFEA India Hickey – Research Officer (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 Licensing and Approvals Pack

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. Alison Male, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Transcobalamin II Deficiency, OMIM #275350, 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 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 Transcobalamin II Deficiency, OMIM #275350, 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. Transcobalamin II Deficiency, OMIM #275350, is characterised by vomiting, diarrhoea, failure to thrive, and susceptibility to infection. If not treated, the condition is rapidly progressive and life threatening, resulting in permanent brain damage and severe haematological complications.

1.10. Treatment for the condition relies on life-long weekly vitamin B12 injections and regular blood monitoring. Non-compliance with treatment can lead to significant neurological complications which can be ameliorated but not always corrected by the resumption of optimal treatment.

1.11. The committee noted the executive's request to consider Transcobalamin II Deficiency, OMIM #275350, 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, Transcobalamin II Deficiency, OMIM #275350, is a potentially severe, life-threatening, and rapidly progressive disease that presents in the early weeks of life. If the condition is left untreated, or there is non-compliance with treatment, individuals may suffer from long-term neurological consequences, permanent brain damage and severe haematological complications. Those living with the condition will require lifelong painful weekly injections and regular blood monitoring. The committee noted the potential physical and psychological impact on the quality of life of those 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 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.

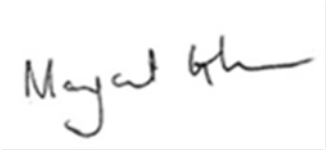
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Transcobalamin II Deficiency, OMIM #275350

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

21 June 2021