Statutory Approvals Committee - minutes

Centre 0044 (The Centre for Reproductive and Genetic Health)

Preimplantation Genetic Diagnosis (PGD) application for Temtamy syndrome (TEMTYS), OMIM #218340

Date:	25 March 2021
Venue:	Microsoft Teams Meeting
Committee Members:	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde
Specialist Adviser:	Jenny Carmichael
Legal Adviser:	Tom Rider - FieldFisher
Members of the Executive:	Moya Berry - Committee officer Catherine Burwood - Licensing Manager
Observers:	Jason Kasraie - Authority Member (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 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 Temtamy syndrome, (TEMTYS), OMIM #218340, 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 Temtamy syndrome (TEMTYS), OMIM #218340, 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. Temtamy syndrome, (TEMTYS), OMIM #218340, is a rare neurological condition that causes brain abnormalities, mild to severe developmental delay, intellectual disability, seizures, and autism. It is also associated with eye abnormalities and heart problems such as aortic dilatation, aortic regurgitation, and myocardial impairment. Symptoms develop from birth or early childhood. The main brain abnormalities associated with this condition relate to the development of the corpus callosum (the connection between the two hemispheres of the brain). The eye abnormalities can include missing pieces of tissue in structures that form the eye (ocular coloboma), lens dislocation, optic atrophy and microphthalmia (abnormally small eyes), which cause visual impairment.

1.10. There is no cure for this condition. Affected children may require surgery for their heart and eye problems. Epilepsy medications may or may not be effective in treating seizures. Speech therapy, occupational therapy and physiotherapy may be needed to help those affected with their developmental delay.

1.11. The committee noted the executive's request to consider Temtamy syndrome (TEMTYS), OMIM #218340, 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, Temtamy syndrome (TEMTYS), OMIM #218340, is a severe, incurable, and progressive neurological disorder that presents from birth and can lead to profound learning disabilities and sight loss. The condition has no

effective treatment and may result in death in babies and children due to complications such as complex heart problems. Due to the variability of the condition, those affected do not know how severe their condition may become. The committee considered the serious implications for, and the physical and psychological impact on the quality of life of, those affected by 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 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.

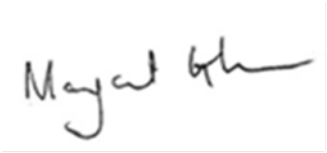
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Temtamy syndrome (TEMTYS), OMIM #218340

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

16 April 2021