Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Thrombocytopenia Absent Radius Syndrome, OMIM #274000

Thursday, 28 May 2020

HFEA, 10 Spring Gardens, London, SW1A 2BU via Teleconference

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 Alison Male	
Legal Adviser	Jane Williams	Mills & Reeve LLP
Observer	Bernadette O'Leary	Clinical Inspector, Induction

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 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 Thrombocytopenia Absent Radius (TAR) Syndrome, OMIM #274000, 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 Thrombocytopenia Absent Radius Syndrome (TAR), OMIM #274000, 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. The condition is characterised by the absence of the radius bone in the forearms and low platelet numbers (thrombocytopenia). In some cases, further skeletal abnormalities may be seen and haemorrhage secondary to thrombocytopenia can lead to permanent cognitive and/or motor impairment, damage to organs other than the brain, and death.

1.10. There is no cure for this condition and treatment focuses on managing the symptoms of individual patients.

1.11. The committee noted the executive's request to consider Thrombocytopenia Absent Radius Syndrome (TAR), OMIM #274000, 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 Thrombocytopenia Absent Radius (TAR) Syndrome, OMIM #274000, is a rare and very serious condition that can present in infancy. There is no cure for the condition which, in the worst-case scenario, can lead to life-threatening bleeding in the brain or in other organs. The committee also noted that malformations of the limbs could affect mobility and everyday living. The committee considered the serious implications for, and 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 or mental disability, a serious illness or any other serious medical condition.

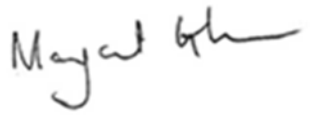
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Thrombocytopenia Absent Radius (TAR) Syndrome, OMIM #274000

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
24 June 2020