Statutory Approvals Committee – minutes

Centre 0044 (The Centre for Reproductive and Genetic Health)

Pre-implantation Genetic Diagnosis (PGD) application for Donnai-Barrow Syndrome, OMIM #222448

Thursday, 30 April 2020

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 Statement
- Peer Review Supporting Document (Anglani et al, 2018)

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 Donnai-Barrow Syndrome, OMIM #222448, is consistent with the peer review.

1.3. The committee noted that the condition being applied 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 Donnai-Barrow Syndrome, OMIM #222448, 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 the penetrance of the condition is close to 100%.

1.9. Donnai-Barrow Syndrome, OMIM #222448, is characterised by diaphragmatic hernia (a birth defect where there is a hole in the diaphragm), exomphalos (weakness of the abdominal wall where the umbilical cord joins, allowing the abdominal contents to protrude outside of the abdominal cavity), absent corpus callosum (the nerve fibre bundle that connects the left side of the brain to the right side), high myopia (severe short-sightedness) and sensorineural deafness. The condition presents prenatally or shortly after birth.

1.10. There is no cure for this condition and treatment focuses on managing the symptoms of individual patients. This may include hearing aids and/or cochlear implants, corrective lenses for vision loss and surgery for certain physical abnormalities.

1.11. The committee noted the executive's request to consider Donnai-Barrow Syndrome, OMIM #222448, 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 Donnai-Barrow Syndrome, OMIM #222448, is a rare and serious multi-system disorder which can present prenatally or at birth. There is no cure for the condition which in some cases can lead to fatal renal disease. The committee also noted that surgery to correct severe birth anomalies is considered to be very high-risk. The committee considered the serious implications and impact on the quality of life for those affected with 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 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.

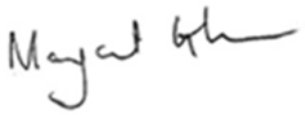
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Donnai-Barrow Syndrome, OMIM #222448

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