Statutory Approvals Committee – minutes

Centre 0119 (Birmingham Women's Hospital) Pre-implantation Genetic Diagnosis (PGD) application for Neurodevelopmental Disorder with Progressive Microcephaly Spasticity and Brain Anomalies (NDMSBA), OMIM #617527

Thursday, 13 December 2018

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe Anthony Rutherford Emma Cave	
Members of the Executive	Dee Knoyle Catherine Burwood Paula Robinson	Committee Secretary Senior Governance Manager (Observer) Head of Planning and Governance (Observer)
Legal Adviser	Eve Piffaretti	Blake Morgan LLP
External adviser	Dr Jenny Carmichael	
Observers		

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:

- 8th 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 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 application for PGD for Neurodevelopmental Disorder with Progressive Microcephaly Spasticity and Brain Anomalies (NDMSBA), OMIM #617527 is consistent with the peer review.

1.3. The committee noted that the condition being applied for is not on the list of conditions approved for PGD.

1.4. The committee noted that the Genetic Alliance opinion provided a patient perspective and supported the 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 the condition is inherited in an autosomal recessive pattern which means there is a 25% chance of an embryo being affected with the condition in each pregnancy, if both parents have a relevant mutation. NDMSBA is fully penetrant.

1.8. The committee noted that this is a neurodevelopmental condition resulting from mutations in the PLAA gene which leads to death in infancy or childhood in the majority of cases. Those that survive have little spontaneous movement, no speech and profound intellectual disability.

1.9. The committee noted that babies are born with small heads and have progressive microcephaly, degeneration of the brain, vision problems, developmental delay and intellectual disability, absent or delayed speech. Individuals also have reduced muscle tone caused by deficits in the brain or spinal cord, and elevated muscle tone often associated with spasticity, contractures restricting movement in the hands and feet, involuntary muscle contractions that cause slow repetitive movements or abnormal postures, rigid spasms in limbs and poor coordination, and curvature of the spine. The condition also causes impaired control of the muscles used in swallowing, eating, drinking and talking, respiratory insufficiency, chest abnormalities and feeding difficulties. Some individuals have seizures.

1.10. The committee noted that these symptoms and features have some variability in onset, with infant death sometimes occurring in the most severe.

1.11. The committee noted that there is no curative treatment for this condition. Treatment is supportive and there are no treatments for NDMSBA which modify disease progression. Some affected children die in early childhood, others may have severe life limitations.

1.12. The committee noted the inspectorate's request to consider whether Neurodevelopmental Disorder with Progressive Microcephaly Spasticity and Brain Anomalies (NDMSBA), OMIM #617527 should be approved for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

2. Decision

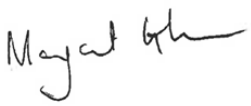
2.1. The committee considered that, in the worst case scenario the condition is serious. The condition is present at birth and in the majority of cases leads to death in infancy or early childhood. The condition affects the brain primarily and causes seizures, intellectual impairment and physical disability. Affected individuals have reduced muscle tone and problems with movement due to impaired control of the muscles, including the muscles used in swallowing, eating, drinking and talking. The condition is progressive and causes degeneration of the brain, breathing difficulties and severe problems with mobility. There is no curative treatment and the quality of life of affected individuals may be severely 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, given the condition's worst symptoms, that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that Neurodevelopmental Disorder with Progressive Microcephaly Spasticity and Brain Anomalies (NDMSBA), OMIM #617527 meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act and agreed to authorise testing.

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

17 January 2019