Statutory Approvals Committee – minutes

Item 1

Centre 0102 (Guy's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Microcephaly, Short Stature and Polymicrogyria with or without seizures (MSSP), OMIM #614833

Thursday, 29 November 2018

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe Anthony Rutherford Ruth Wilde Emma Cave	
Members of the Executive	Dee Knoyle Catherine Burwood Paula Robinson Victoria Askew	Committee Secretary Senior Governance Manager (Observer) Head of Planning and Governance (Observer) Inspections & Logistics Officer (Observer)
Specialist Adviser	Dr Alan Fryer	
Legal Adviser	Ros Foster	Browne Jacobson LLP
Observers	Rachel Cutting (New Member Induction - Observer)	

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 Alan Fryer 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 Microcephaly, Short Stature and Polymicrogyria with or without seizures (MSSP), OMIM #614833 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. Penetrance is difficult to determine as MSSP is very rare but current evidence seems to indicate that the condition is fully penetrant.

1.8. The committee noted that the condition causes dwarfism, microcephaly and a variety of brain malformations including polymicrogyria as well as severe intellectual disability, seizures and delayed speech and motor development. MSSP is an incurable condition so treatment focuses on the alleviation of symptoms. Treatment options include physiotherapy, speech therapy and educational aids. Anti-epileptic medications are administered in the case of seizures. Affected individuals will require lifelong treatment and support and there are significant impacts on daily quality of life.

1.9. The committee noted the inspectorate's request to consider whether Microcephaly, Short Stature and Polymicrogyria with or without seizures (MSSP), OMIM #614833 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

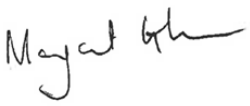
2.1. The committee considered that, in the worst case scenario MSSP, OMIM #614833 is a serious condition characterised by dwarfism and severe intellectual disability. Babies are born very small, with small heads and brain malformations, and in some cases other abnormalities have been reported including duodenal atresia and single kidneys. In the worst case scenario, babies can fail to thrive and may die within a few months of life. Affected individuals who survive have severe intellectual disability, seizures and delayed speech and motor development. Treatment is therapeutic, focussed on the alleviation of symptoms. Anti-epileptic medications used for treating seizures are not always successful and the seizures can be intractable. There is no curative treatment. Affected individuals require lifelong treatment and support and their quality of life is severely impacted.

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 Microcephaly, Short Stature and Polymicrogyria with or without seizures (MSSP), OMIM #614833 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

19 December 2018