Statutory Approvals Committee – minutes

Item 4

Centre 0044 (The Centre for Reproductive and Genetic Health)

Pre-implantation Genetic Diagnosis (PGD) application for Multiple Sulfatase Deficiency (MSD), OMIM #272200

Thursday, 25 April 2019

HFEA Spey Meeting Room, Level 2, 10 Spring Gardens, London, SW1A 2BU

Committee Members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Emma Cave Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Secretary Licensing Manager (Observer)
Specialist Adviser	Professor Mary Porteous	
Legal Adviser	Graham Miles	Blake Morgan LLP
Observers	Amanda Evans Jennifer Rogerson	Research Manager (Induction) Research Manager (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 UK statement
- 31 March 2011 Licence Committee minutes - PGD for Mucopolysaccharidosis type VI
- 25 June 2008 Licence Committee minutes – PGD for Metachromatic Leukodystrophy

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Professor Mary Porteous, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Multiple Sulfatase Deficiency (MSD), OMIM #272200, was 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 Multiple Sulfatase Deficiency, 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 each parent has a relevant mutation.

1.8. The committee noted the penetrance of the condition is 100%.

1.9. Multiple Sulfatase Deficiency is a rare genetic condition caused by a mutation in the SUMF1 gene. All affected individuals are severely affected.

1.10. Multiple Sulfatase Deficiency is characterised by leukodystrophy (leading to movement problems, seizures, developmental delay, cognitive impairment and slow growth), dry scaly skin, excess hair growth and skeletal abnormalities such as abnormal spinal curvature and joint stiffness. Other features can include hearing loss, heart malformations and an enlarged liver and spleen.

1.11. Affected individuals only survive a few years after the symptoms of the condition appear. Although life expectancy varies depending on the severity of the condition and how quickly neurological problems worsen, in all cases the condition is progressive, symptoms will worsen with time and life expectancy will be shortened. Most of those affected would be expected to die in childhood.

1.12. Multiple Sulfatase Deficiency is not curable. Physiotherapy and other supportive services are available to help manage the effects of the condition.

1.13. The committee noted the inspectorate’s request to consider Multiple Sulfatase Deficiency, OMIM #272200 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 Multiple Sulfatase Deficiency (MSD), OMIM #272200 is a severe life-limiting neurodegenerative condition, which leads to death in childhood. In all cases the condition is severe. The committee considered the severe effects on the quality of life of those affected by this condition for which there is currently no cure.

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 the abnormality will, given the conditions' worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

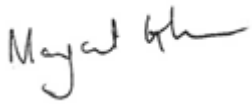
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Multiple Sulfatase Deficiency (MSD), OMIM #272200

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

15 May 2019