Statutory Approvals Committee – minutes

Item 2

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Surfactant Dysfunction Metabolism 3 (SMDP3),OMIM #610921

Thursday, 28 March 2019

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

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Emma Cave Anne Lampe Tony Rutherford	
Members of the Executive	Moya Berry Catherine Burwood	Committee Secretary Licensing Manager (Observer)
Specialist Adviser	Dr Alison Male (PGD)	
Legal Adviser	Tom Rider	Fieldfisher LLP
Observers	Hannah Carpenter	Policy Officer

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

- SAC minutes 20 November 2014, PGD for Surfactant Metabolism Dysfunction, Pulmonary type 2 (SMDP2)
- Licence Committee minutes 23 May 2013, PGD for Surfactant Metabolism Dysfunction, Pulmonary, type 1 (SMDP1)

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 Surfactant Dysfunction Metabolism 3 (SMDP3), OMIM # 610921, 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. The committee noted that Surfactant Metabolism Dysfunction, Pulmonary, Type 1 (SMDP1), OMIM #265120 and Surfactant Metabolism Dysfunction, Pulmonary, Type 2 (SMDP2), OMIM #610913 are already approved for PGD and to ensure consistency with the other condition types, the condition being applied for will be termed Surfactant Metabolism Dysfunction, Pulmonary, Type 3 (SMDP3), OMIM #610921.

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 Surfactant Metabolism Dysfunction, Pulmonary Type 3 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 penetrance of the condition is 100%.

1.9. Surfactant Metabolism Dysfunction, Pulmonary Type 3 is caused by a mutation in the ABCA3 gene. It is characterised by the absence, or abnormal forms of pulmonary surfactant. Pulmonary surfactant increases the ability of the lungs to expand when breathing in and prevents the lungs from collapsing when breathing out. Patients with Surfactant Metabolism Dysfunction, Pulmonary Type 3 either completely lack or have abnormal forms of these pulmonary surfactant. The condition is associated with babies having difficulty breathing independently after birth and lung disease in children and adults.

1.10. The condition normally becomes apparent in babies shortly after birth and 80% of patients will die either within the first 6 months of their life or before the age of 5 years.

1.11. There is no cure for the condition. In new-born babies, artificial respiration can be used to help establish breathing. Patients who survive may be required to have ongoing respiratory support using tracheostomy and mechanically assisted breathing at home. Patients are also required to take medications such as steroids and immunosuppressants which have significant side effects including an increased risk of infections. The effectiveness of these therapeutic interventions is poor, and a very small number of patients will require a lung transplant.

1.12. The committee noted the inspectorate's request to consider Surfactant Metabolism Dysfunction, Pulmonary Type 3, OMIM #610921 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 Surfactant Metabolism Dysfunction, Pulmonary Type 3, OMIM #610921 is a severe lethal condition. There is no cure and most patients will die either within the first 6 months of life or before the age of 5 years.

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.

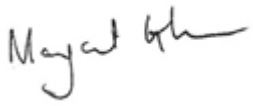
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Surfactant Metabolism Dysfunction, Pulmonary Type 3, OMIM #610921

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

24 April 2019