Statutory Approvals Committee – minutes

Item 2

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Spondylometaepiphyseal Dysplasia Short Limb Hand Type (SMED-SL), OMIM #271665

Thursday, 30 May 2019

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

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe Tony Rutherford Rachel Cutting	
Members of the Executive	Moya Berry Catherine Burwood Debbie Okutubo Nora Cooke- O'Dowd	Committee Secretary Licensing Manager (Observer) Governance Manager (Induction) Head of Research & Intelligence (Induction)
Specialist Adviser	Dr Alison Male	
Legal Adviser	Ros Foster	Browne Jacobson LLP

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
- Supporting document – Bargal et al paper

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 Spondylometaepiphyseal Dysplasia Short Limb Hand type (SMED-SL), OMIM #271665 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 Spondylometaepiphyseal Dysplasia Short Limb Hand type (SMED-SL), 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. Spondylometaepiphyseal Dysplasia Short Limb Hand type (SMED-SL), is a progressive condition and is characterised by skeletal abnormalities including shortening of upper and lower limbs, spinal abnormalities and a narrowed or ‘caved-in’ appearance to the chest. Patients are frequently wheelchair dependent. Skeletal abnormalities may lead to fatal complications (spinal compression and recurrent respiratory disease). Affected individuals can also show delayed motor and intellectual development.

1.10. There are no treatments available for Spondylometaepiphyseal Dysplasia Short Limb Hand type (SMED-SL), however medication and physiotherapy can be given to treat recurrent respiratory infections. Some patients require neurosurgery, but this may not prevent death.

1.11. The committee noted the inspectorate’s request to consider Spondylometaepiphyseal Dysplasia Short Limb Hand type (SMED-SL), OMIM #271665 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 Spondylometaepiphyseal Dysplasia Short Limb Hand type (SMED-SL), OMIM #271665 is a severe, progressive condition which is disabling and potentially life-limiting. The symptoms of the condition, which are present from birth, can lead to lethal complications including spinal compression and recurrent respiratory disease. There is no cure for the condition and there is a significant risk of death in childhood.

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 condition's worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

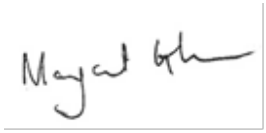
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Spondylometaepiphyseal Dysplasia Short Limb Hand type (SMED-SL), OMIM #271665

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

25 June 2019