Statutory Approvals Committee – minutes

Centre 0314 (Leeds Fertility)

Pre-implantation Genetic Diagnosis (PGD) application for Multiple Joint Dislocations, Short Stature and Craniofacial Dysmorphism with or without Congenital Heart Defects (JDSCD)

OMIM #245600

Thursday, 28 November 2019

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

Committee members	Margaret Gilmore (Chair) Anne Lampe Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr Alan Fryer	
Legal Adviser	Eve Piffaretti	Blake Morgan LLP

Declarations of interest

- Tony Rutherford declared an interest with this item and withdrew from discussion during this part of the meeting.
- There were no conflicts of interest declared by any other members of the committee.

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
- 2019-09-26 Statutory Approval Committee Minutes - Spondyloepiphyseal Dysplasia with Congenital Joint Dislocations (SEDCJD), OMIM #143095 and Larson Syndrome, OMIM #150250

- 2019-05-30 Statutory Approvals Committee Minutes - Spondylometaepiphyseal Dysplasia Short Limb Hand type, OMIM #271665
- 2015-08-27 Statutory Approvals Committee Minutes - Spondyloepimetaphyseal dysplasia, Strudwick, OMIM #184250
- 2011-05-05 Licence Minutes - Spondyloepiphyseal Dysplasia Congenita, OMIM #183900

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 PGD application for Multiple Joint Dislocations, Short Stature, and Craniofacial Dysmorphism with or without Congenital Heart Defects (JDSCD), OMIM #245600 is 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 Multiple Joint Dislocations, Short Stature, and Craniofacial Dysmorphism with or without Congenital Heart Defects (JDSCD), OMIM #245600 is inherited in an autosomal recessive pattern, which means there is a 25% chance of an embryo being affected by the condition in each pregnancy if each parent has a relevant mutation.

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

1.9. Multiple Joint Dislocations, Short Stature, and Craniofacial Dysmorphism with or without Congenital Heart Defects (JDSCD), OMIM #245600 is characterised by skeletal dysplasia (abnormalities of cartilage and bone growth) which can cause short, bowed long bones resulting in disproportionate short stature. Skeletal abnormalities also include spinal defects such as scoliosis and kyphosis, club foot, joint dislocations and contractures. Respiratory and cardiac abnormalities can also occur, which may be fatal. Multiple surgeries may be required to correct skeletal and other abnormalities. Those affected by the condition may experience a significant level of disability which affects their health, independence, life choices, education, and work-related opportunities. Quality of life may also be affected by severe pain from joint dislocations and abnormal joint function.

1.10. There is no cure for this condition, so treatment focuses on managing the symptoms.

1.11. The committee noted the executive's request to consider Multiple Joint Dislocations, Short Stature, and Craniofacial Dysmorphism with or without Congenital Heart Defects (JDSCD), OMIM #245600 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 Joint Dislocations, Short Stature, and Craniofacial Dysmorphism with or without Congenital Heart Defects (JDSCD), OMIM #245600 is a rare and severely painful condition which presents at birth. There is no cure for this condition and death may occur in infancy as a result of cardiac anomalies and respiratory problems. The committee considered the possible adverse physical and psychological impact on the quality of life for patients living with the condition.

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 abnormalities in question and that there is a significant risk, that a person with such an abnormality will, given the conditions' worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

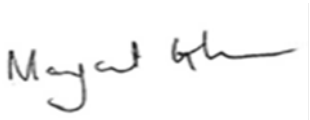
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Multiple Joint Dislocations, Short Stature, and Craniofacial Dysmorphism with or without Congenital Heart Defects (JDSCD), OMIM #245600

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
24 December 2019