# Statutory Approvals Committee – minutes

## Centre 0119 (Birmingham Women's Hospital)

## Pre-implantation Genetic Diagnosis (PGD) application for

## Multiple Pterygium Syndrome, Escobar Variant, EVMPS, OMIM #265000

Thursday, 25 May 2017

Church House Westminster, Dean's Yard, Westminster SW1P 3NZ

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| Committee members | Margaret Gilmore (Chair)<br>Anne Lampe<br>Ruth Wilde<br>Anthony Rutherford<br>Bobbie Farsides | |
| Members of the Executive | Bernice Ash<br>Paula Robinson | Secretary<br>Head of Planning & Governance |
| External adviser | Dr Jenny Carmichael | |
| Legal Adviser | Sarah Ellson | Fieldfisher |
| Observers | | |

## Declarations of interest

- Members of the panel 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 opinion
- Licence Committee Minutes

## 1. Consideration of application

**1.1.** The committee welcomed the advice of its specialist adviser, Dr Jenny Carmichael, 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 Pterygium Syndrome, Escobar variant, EVMPS, OMIM #265000 is consistent with the Peer Review.

**1.3.** The committee noted that the Genetic Alliance opinion provided a patient perspective and supported the application.

**1.4.** 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.

**1.5.** The committee noted that the condition being applied for is not on the list of approved PGD conditions.

**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 manner which means there is 25% chance of having an affected child in each pregnancy, if each parent has a relevant mutation.

**1.8.** Multiple Pterygium Syndrome, Escobar variant, EVMPS is a condition that is evident before birth, with webbing of the skin at the joints and a lack of muscle movement. The combined effect, termed arthrogryposis, is to restrict joint movement which in turn often results in an inability to fully extend arms and legs. Sometimes this condition results in intrauterine death.

**1.9.** The committee noted that, webbing typically affects the skin of the neck, fingers, forearms, inner thighs, and backs of the knee. In addition to arthrogryposis, sometimes a side-to-side curvature of the spine occurs. At birth, affected babies often have respiratory distress due to underdeveloped lungs, and fatal congenital heart defects have been reported. Penetrance is thought to be 100%

**1.10.** There is currently no curative treatment for this condition. This is frequently a lethal condition. Surviving children are born with a combination of features which lead to a number of serious, though usually non-progressive, handicaps and may require prolonged periods of treatment to improve mobility e.g. surgery to improve joint function and release webs, and physiotherapy.

**1.11.** The committee noted the inspectorate’s recommendation to consider the approval of Multiple Pterygium Syndrome, Escobar variant, EVMPS, OMIM #265000 to be included on the PGD List. The committee agreed to consider the application on this basis.

## 2. Decision

**2.1.** The committee considered Multiple Pterygium Syndrome, Escobar variant, EVMPS, OMIM #265000 is serious given the early onset and the effects on the quality of life of this condition including severe physical difficulties, prolonged periods of treatments including surgeries and physiotherapy, and the lack of a cure. The committee considered the impact on the individuals' quality of life and the family caring for affected individuals.

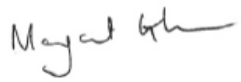
**2.2.** The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition of Multiple Pterygium Syndrome, Escobar variant, EVMPS, OMIM #265000 does meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

**2.3.** The committee agreed to authorise the testing of embryos for Multiple Pterygium Syndrome, Escobar variant, EVMPS, OMIM #265000.

## 3. Chair's signature

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

### Signature



### Name

Margaret Gilmore

### Date

8 June 2017