# Statutory Approvals Committee – minutes

## Centre 0102 (Guys Hospital)

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

## Isolated Microphthalmia 2 (MCOP2), OMIM #610093

Thursday, 23 February 2017

HFEA, 10 Spring Gardens, London, SW1A 2BU

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| Committee members | Margaret Gilmore (Chair)<br>Anne Lampe<br>Ruth Wilde<br>Bobbie Farsides | |
| Members of the Executive | Dee Knoyle<br>Siobhain Kelly<br>Erin Barton | Secretary<br>Interim Head of Corporate Governance<br>Governance Manager |
| External adviser | Dr Mary Porteous | |
| Legal Adviser | Shelley Edwards | 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
- Two Academic papers
- Genetic Alliance Opinion

## 1. Consideration of application

**1.1.** The committee welcomed the advice of its specialist adviser, Dr 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 Isolated Microphthalmia 2 (MCOP2), OMIM #610093 is consistent with the Peer Review.

**1.3.** The committee noted that the Genetic Alliance opinion provided a patient's 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 pattern and there is a 1 in 4 chance of an embryo being affected with the condition if each parent is a carrier of a relevant mutation.

**1.8.** The committee noted that the condition demonstrates 100% penetrance and the onset of symptoms is from birth.

**1.9.** The committee noted that MCOP2 is a condition resulting in eye malformation of varying severity.

**1.10.** In some patients there will be complete absence of the structure and components of one or both eyes (anopthalmia). In others, the eyes will be under developed (micropthalmia) and may be affected by additional features including coloboma (failure of full development of parts of the eye) and cataract (opacity of lens). These characteristics result in complete or significant visual impairment from birth with affected individuals being registered blind.

**1.11.** Affected children are taught to manage their complete or significant visual impairment since there is no treatment available to improve visual ability. They require developmental and educational support to maximise their psychomotor function, life skills and mobility. Prosthetic eyes are surgically implanted, at regular intervals, to enable the growth of the orbits to match the requirements of the growing child; these prostheses have no impact on visual function.

**1.12.** The committee noted that there is no curative treatment for the condition.

**1.13.** The committee noted the inspectorate’s recommendation to consider the approval of Isolated Microphthalmia 2 (MCOP2) to be included on the PGD List. The committee agreed to consider the application on this basis.

## 2. Decision

**2.1.** The committee considered that the condition is serious due to the risk of complete blindness, absence of eyes at birth and the need for surgery at regular intervals to implant prosthetic eyes which have no impact on visual function but enable the growth of the orbits to match the requirements of the growing child. The committee also considered the psychological impact on the family and child affected by the condition.

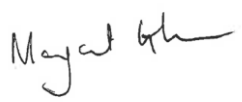
**2.2.** The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability,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

**2.3.** The committee agreed to authorise the testing of embryos for Isolated Microphthalmia 2 (MCOP2), OMIM #610093.

## 3. Chair's signature

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

### Signature



### Name

Margaret Gilmore

### Date

10 March 2017