# Statutory Approvals Committee – minutes

## Item 4

## Centre 0101 (CARE Fertility Nottingham)

## Pre-implantation Genetic Diagnosis (PGD) application for Laryngo-onycho-cutaneous (LOC) syndrome, OMIM #245660

Thursday, 28 February 2019

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

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| Committee members | Margaret Gilmore (Chair)<br>Bobbie Farsides (Deputy Chair)<br>Emma Cave<br>Rachel Cutting<br>Ruth Wilde | |
| Members of the Executive | Moya Berry<br>Dee Knoyle<br>Catherine Burwood | Committee Secretary<br>Committee Secretary (Observer)<br>Senior Governance Manager (Observer) |
| Specialist Adviser | Dr Alan Fryer | |
| Legal Adviser | Sarah Ellson | Fieldfisher LLP |
| Observers | | |

## 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

## 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 application for PGD application for Laryngo-onycho-cutaneous (LOC) syndrome, OMIM #245660 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 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 Laryngo-onycho-cutaneous Syndrome 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 either parent has a relevant mutation.

**1.8.** The committee noted that Laryngo-onycho-cutaneous Syndrome is extremely rare. No information on penetrance was provided by either the peer reviewer or applicant. As an autosomal recessive condition however, it is likely to be 100% penetrant.

**1.9.** The committee noted the condition is characterised by excessive granulation of the mucous membranes in the respiratory and ocular systems as well as the teeth, skin and nails. Involvement of the respiratory and ocular systems can lead to obstructed airways and respiratory failure and visual impairment resulting in blindness. Patients also suffer from delayed healing of wounds with subsequent risk of sepsis. Those affected have symptoms likely to significantly reduce their quality of life and it has caused death in childhood.

**1.10.** There is no cure for Laryngo-onycho-cutaneous syndrome, so treatments are focused on managing symptoms.

## 2. Decision

**2.1.** The committee considered that, in the worst-case scenario Laryngo-onycho-cutaneous syndrome, OMIM #245660 is a serious condition which affects multiple systems within the body. Most of those affected will not survive past early childhood and those who do, may become blind and may be prone to respiratory failure with serious effects on the quality of life.

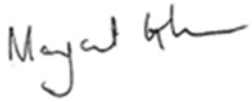
**2.2.** The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, given the conditions' worst symptoms, that a person with the abnormality will have or develop a serious physical disability, a serious illness or any other serious medical condition.

**2.3.** The committee was therefore satisfied that Laryngo-onycho-cutaneous (LOC) syndrome, OMIM #245660 meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act and agreed to authorise testing.

## 3. Chairs signature

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

### Signature



### Name

Margaret Gilmore

### Date

14 March 2019