# Statutory Approvals Committee - minutes

## Centre 0102 (Guy's Hospital) – PGD application for Mitochondrial Complex 1 Deficiency OMIM #611126

Thursday, 24 September 2015

HFEA, Finsbury Tower, 103-105 Bunhill Row, London, EC1Y 8HF

| | | |
|---|---|---|
| Committee members | David Archard (Chair, lay)<br>Rebekah Dundas (Deputy Chair, lay)<br>Sue Price (professional)<br>Margaret Gilmore (lay) | |
| Members of the Executive | Sam Hartley<br>Trent Fisher | Head of Governance and Licensing<br>Secretary |
| External adviser | Dr Anne Lampe | |
| Legal Adviser | Ros Foster | Browne Jacobson |
| Observers | Erin Barton | Projects, Inspections and Logistics Officer |

## 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:

- 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

## 1. Consideration of application

**1.1.** 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.

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

**1.3.** The committee noted that the proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of schedule 2 to the Human Fertilisation and Embryology Act 1990 (as amended) ("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'.

**1.4.** 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 where both parents are carriers.

**1.5.** The committee noted that the condition is a neuro-degenerative multisystem disorder that has a range of symptoms which can affect the brain (leukencephalopathy), liver, heart(cardiomyopathy) and skeletal muscle (exercise intolerance). It is a life threatening condition and can cause death within a few weeks of birth. Children who survive are affected with long-term secondary conditions that require lifelong monitoring.

**1.6.** The committee noted that the condition applied for is likely to demonstrate a high penetrance by late adult life. Affected children are usually diagnosed in infancy.

**1.7.** The committee noted that there is no curative treatment for the underlying condition and treatment is smyptomatic.

**1.8.** The committee noted that the application is consistent with the Peer Review.

**1.9.** The committee welcomed the advice of its specialist advisor, Dr Anne Lampe, who confirmed that the condition is as described in the papers and that the condition type applied for is caused by a mutation in the nuclear ACAD9 gene. Dr Lampe added further that early death is common and can occur as early as the neonatal period.

## 2. Decision

**2.1.** The committee considered that the condition is serious due to the range of symptoms associated, the risk of death in infancy and lack of any curative treatment.

**2.2.** The committee had regard to its explanatory note and on the basis of the information presented, given the condition's worst symptoms, 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.

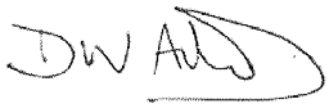
**2.3.** The committee decided that to avoid confusion the condition for which testing had been authorised will appear as 'Mitochondrial Complex 1 Deficiency caused by mutations in ACAD9' on the HFEA approved PGD conditions list.

**2.4.** The committee agreed to authorise the testing of embryos for Mitochondrial Complex 1 Deficiency caused by a mutations in ACAD9, OMIM #611126.

## 3. Chair's signature

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

**Signature**



**Name**

David Archard

**Date**

8 October 2015