# Statutory Approvals Committee - minutes

## Centre 0102 (Guy's Hospital) – PGD application for X-linked Adrenal Hypoplasia Congenita (XL-AHC) OMIM #300200

Thursday, 24 November 2016

HFEA, Level 2, 10 Spring Gardens, London, SW1A 2BU

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| Committee members | Margaret Gilmore (Chair)<br>Rebekah Dundas (Deputy Chair)<br>Anne Lampe<br>Ruth Wilde<br>Anthony Rutherford | |
| Members of the Executive | Siobhain Kelly<br><br>Trent Fisher | Head of Corporate Governance (interim)<br>Secretary |
| External adviser | Professor Mary Porteous | |
| Legal Adviser | Graham Miles – Blake Morgan LLP | |
| Observers | None | |

## 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
- An additional academic paper submitted with the application
- redacted peer review

- Genetic Alliance opinion

## 1. Consideration of application

**1.1.** The committee welcomed the advice of its specialist advisor, Professor Mary Porteous, who confirmed that the condition is as described in the papers.

**1.2.** The committee noted that the application is consistent with the Peer Review and Genetic Alliance opinion.

**1.3.** 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.

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

**1.5.** 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'.

**1.6.** The committee noted that the condition is inherited in an X-linked pattern and for a female carrier of an altered gene there is a 25% chance of having an affected child (male) in each pregnancy: one in two boys born to a carrier woman will be affected and one in two girls will inherit the affected gene and be a carrier.

**1.7.** The committee noted that affected males usually present in infancy with vomiting, feeding difficulty, dehydration and shock due to a loss of important salts.

**1.8.** The committee noted that in affected males, the condition causes adrenal insufficiency which can be triggered by episodes of stress and if treatment is not received during an acute episode, the condition can be fatal. Also, puberty can be delayed and can be associated with infertility.

**1.9.** The committee noted that the condition is fully penetrant and usually presents in the first weeks of life but can present throughout childhood and up to early adulthood.

**1.10.** The committee noted that lifelong specialist treatment from endocrinologists and other medical support would be needed by affected males.

## 2. Decision

**2.1.** The committee considered that the condition is serious due to the ongoing risk of adrenal crises which untreated can lead to death and the requirement for life long compliance with steroid treatment. In addition, the committee noted that the majority of affected males experience delayed puberty and variable infertility.

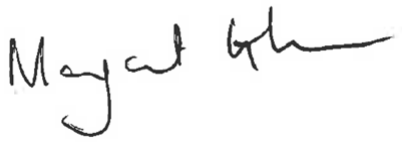
**2.2.** The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, 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 X-linked Adrenal Hypoplasia Congenita (XL-AHC) #300200.

## 3. Chair's signature

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

**Signature**



**Name**

Margaret Gilmore

**Date**

09 December 2016