# Statutory Approvals Committee - minutes

## Centre 0102 (Guys Hospital) – PGD application for Danon Disease, OMIM #300257

Friday, 24 June 2016

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

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| Committee members | David Archard (Chair)<br>Rebekah Dundas (Deputy Chair)<br>Margaret Gilmore<br>Anthony Rutherford<br>Anne Lampe<br>Ruth Wilde | |
| Members of the Executive | Ian Brown<br>Trent Fisher | Head of Corporate Governance<br>Secretary |
| External adviser | Jenny Carmichael | |
| Legal Adviser | Dawn Brathwaite | Mills & Reeve 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
- redacted peer review
- Genetic Alliance opinion
- additional paper provided by the peer reviewer entitled “Danon Disease: Clinical Features, Evaluation, and Management.”

## 1. Consideration of application

**1.1.** The committee welcomed the advice of its specialist advisor, Dr Jenny Carmichael, who confirmed that the condition is as described in the papers. Dr Carmichael gave the committee a detailed overview of the condition.

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

**1.3.** The committee noted the Genetic Alliance opinion which helped the committee gain an understanding of the condition from the patient perspective.

**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 approved PGD condition list.

**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 X-linked dominant manner which means there is 50% chance of having an affected child in each pregnancy, if either parent is a carrier of a relevant mutation.

**1.8.** The committee noted further that if the female partner has a causative mutation, there is a 50% chance male offspring inheriting the condition and a 50% chance female offspring inheriting the condition. If the male partner is affected, all male offspring will not inherit the condition and all female offspring will inherit the condition

**1.9.** The committee noted that the condition primarily causes cardiomyopathy and cardiac conduction abnormalities as well as being associated with learning difficulties in some males. Individuals who inherit the condition have a reduced life expectancy.

**1.10.** The committee noted that symptoms include heart palpitations, breathlessness on exertion, fatigue and fainting episodes progressing to heart failure which can be fatal.

**1.11.** The committee noted that other symptoms associated with the condition are generalised muscle weakness, intellectual impairment, eye problems and poor mental concentration.

**1.12.** The committee noted that the penetrance for the condition is likely to be complete for males and high for females. The condition is extremely variable even within the same family

**1.13.** The committee noted that that papers state the onset of symptoms for males can be during infancy and onset of symptoms is generally later for females.

**1.14.** The committee noted that there is no curative treatment for the entire condition. Treatment for the cardiac aspect of the condition is a heart transplant which carries the risks associated with surgery.

## 2. Decision

**2.1.** The committee considered that the condition is serious due to the range of symptoms associated with the condition which can be fatal, the individual's reduced life expectancy and the lack of any curative treatment.

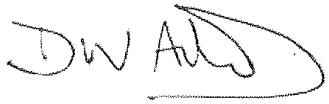
**2.2.** The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, 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 Danon Disease, OMIM #300257.

## 3. Chair's signature

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

**Signature**



**Name**

David Archard

**Date**

11 July 2016