# Statutory Approvals Committee - minutes

## Centre 0102 (Guys Hospital) – PGD application for FAS-associated death domain protein deficiency OMIM #613759

Friday, 5 August 2016

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

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| Committee members | Rebekah Dundas (Chair)<br>Margaret Gilmore<br>Anthony Rutherford<br>Anne Lampe | |
| Members of the Executive | Ian Brown<br>Trent Fisher | Head of Corporate Governance<br>Secretary |
| External adviser | Peter Turnpenny | |
| Legal Adviser | Sarah Ellson | Fieldfisher 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
- two additional supporting papers
- redacted peer review
- Genetic Alliance opinion

## 1. Consideration of application

**1.1.** The committee welcomed the advice of its specialist advisor, Professor Peter Turnpenny, 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 the 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 autosomal recessive pattern and there is a 1 in 4 chance of an embryo being affected with the condition where both parents are carriers.

**1.7.** The committee noted that the condition disrupts the development of the spleen so children suffer from an impaired immune system resulting in recurrent episodes of infection, fever and liver dysfunction. There is a high mortality rate associated with the condition.

**1.8.** The committee noted that episodes of encephalopathy may result in memory loss, changes in personality, seizures and occasionally coma. These recurrent episodes last for several days during which the child requires intensive care.

**1.9.** The committee noted that the condition demonstrates complete penetrance. Age of onset is usually in early childhood, where affected children often present with pneumococcal meningitis or encephalopathy

**1.10.** The committee noted that there is no curative treatment for the condition. Many affected children have to be on permanent pneumococcal prophylaxes. Bone marrow transplants are a potential treatment option but these are not always successful and the procedure comes with its own significant risks.

## 2. Decision

**2.1.** The committee considered that the condition is serious due to the severe symptoms associated with the condition which are often fatal.

**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 FAS-associated death domain protein deficiency OMIM #613759.

## 3. Chair's signature

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

### Signature

### Name

Rebekah Dundas

### Date

19 August 2016