# Statutory Approvals Committee - minutes

## Centre 0102 (Guy's Hospital) – PGD application for Bloom Syndrome OMIM #210900

Thursday, 24 September 2015
HFEA, Finsbury Tower, 103-105 Bunhill Row, London, EC1Y 8HF

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| 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), 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 applied for is inherited in an autosomal recessive pattern and there is a 1 in 4 risk of an embryo being affected with the condition where both parents are carriers.

**1.5.** The committee noted that Bloom Syndrome is a rare condition which is characterised by predisposition to the development of cancer, genomic instability and short stature.

**1.6.** The committee noted that individuals with this condition are at an increased risk of developing cancers including leukaemia, lymphoma, adenocarcinoma and squamous cell carcinoma. Other symptoms include chronic lung disease, reduced immunity to infection, learning difficulties, increased risk of diabetes, sun sensitive skin changes and distinct facial features. Men who inherit this condition will be infertile and affected women may experience early menopause.

**1.7.** The committee noted that the condition demonstrates complete penetrance and that symptoms may be present at birth.

**1.8.** The committee noted that there is no curative treatment for this condition. The only treatment available is to treat the symptoms that arise. Individuals will also have a hypersensitivity to treatments such as chemotherapy and radiotherapy.

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

**1.10.** The committee welcomed the advice of its specialist advisor, Dr Anne Lampe, who confirmed that the condition is as described in the papers.

## 2. Decision

**2.1.** The committee considered that the condition is serious due to the seriousness of symptoms, lack of curative treatment and early age of onset of symptoms.

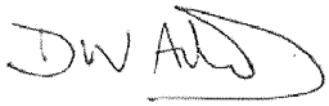
**2.2.** The committee had regard to its explanatory note and on the basis of the information presented, given the condition's worst symptoms, was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness 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 agreed to authorise the testing of embryos for Bloom Syndrome OMIM #210900

## 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