HFEA Statutory Approvals Committee

23 May 2013

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

Minutes – Item 2

Centre 0102 (Guys Hospital) – PGD application Surfactant Metabolism Dysfunction, Pulmonary 1 (SMDP1) OMIM #265120

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford
Sue Price (professional)	
Rebekah Dundas (lay) (Videoconference)	Legal Adviser:
Jane Dibblin (lay)	Graham Miles, Morgan Cole

Declarations of Interest: members of the Committee declared that they had no conflicts of interest in relation to this item.

The following papers were considered by the Committee

- Cover sheet
- Executive Summary
- PGD Application form
- Redacted Peer Review
- Genetic Alliance Opinion

The Committee also had before it

- HFEA Protocol for the Conduct of Licence Committee Meetings and Hearings
- 8th edition of the HFEA Code of Practice
- Human Fertilisation and Embryology Act 1990 (as amended)
- Decision trees for granting and renewing licences and considering requests to vary a licence (including the PGD decision tree); and
- Guidance for members of Authority and Committees on the handling of conflicts of interest approved by the Authority on 21 January 2009.
- Guidance on periods for which new or renewed licences should be granted
- Standing Orders and Instrument of Delegation
- Indicative Sanctions Guidance
- HFEA Directions 0000 – 0012
- Guide to Licensing

- Compliance and Enforcement Policy
- Policy on Publication of Authority and Committee Papers
- HFEA Pre-Implantation Diagnostic Testing ("PGD") Explanatory Note For Licence Committee

Discussion

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.

2. The Committee noted that the Centre's proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of schedule 2 of the Act, ie. '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'.

3. The Committee noted that Surfactant Metabolism Dysfunction, Pulmonary 1 (SMDP1, OMIM #265120) is inherited in an autosomal recessive pattern. There is therefore a 1 in 4 chance of the embryo being affected by this condition if both parents are carriers of the mutation.

4. The Committee noted that SMDP1 is a respiratory condition affecting the lungs. Several genes encode surfactant proteins and this application concerns the gene encoding surfactant protein B (SFTPB). The purpose of pulmonary surfactant in the lungs is to regulate pulmonary activity and prevent the lung from collapsing when one exhales (deflation stability). Dysfunctions of surfactant metabolism lead to severe respiratory distress (breathing difficulties) in the neonatal period and can be fatal within a year.

5. The Committee noted that the symptoms of the condition are severe and respiratory failure usually occurs in the neonatal period. In addition lung cavities can fill with a granular or foamy material which can cause lung disease and result in death within three months of birth. Some patients with disorders due to mutations in other surfactant proteins may have a later age of onset.

6. The Committee considered that the condition is serious because respiratory distress usually presents in the neonatal period. Severe respiratory insufficiency in the neonatal period is associated with pulmonary alveolar proteinosis (where the alveoli fill with a granular or foamy material). The condition is fatal in early childhood. There has been one reported case of a

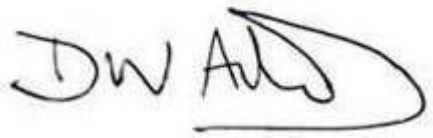
family with a milder phenotype, in which a child survived to age 6 and another child had a lung transplant at 4 months of age. However in most cases the affected children die within the first year of life. Children with SMDP1 have a very short window to find a lung donor for transplant; the average waiting time for a lung transplant is more than four times their median life expectancy.

7. The Committee noted that the application is supported by the Peer Reviewer and the Genetic Alliance UK.

8. 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) of Schedule 2 to the Act.

9. The Committee agreed to authorise the testing of embryos for Surfactant Metabolism Dysfunction, Pulmonary 1 (SMDP1, OMIM #265120). The Committee confirmed that this condition will be added to the published list of condition for which PGD may be carried out.

Signed: Date: 06/06/2013



David Archard (Chair)