HFEA Licence Committee Meeting

1st December 2011

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

Minutes – Item 1

Centre 0035 (Oxford Fertility Unit) – PGD for Townes-Brock Syndrome (OMIM #107480)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Sally Cheshire (lay) (videoconference) Rebekah Dundas (lay) (videoconference) Sue Price (professional) Mair Crouch (lay) Jane Dibblin (lay)	Lauren Crawford Legal Adviser: Tom Rider, Field Fisher Waterhouse

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
- Research review: Kohlhase J (2007) in Gene Reviews [Internet] (editors: Pagon RA, Bird TD, Dolan CR, Stephens K), Seattle (WA): University of Washington, Seattle; 1993-2007
- 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

Background

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 Townes-Brock Syndrome (OMIM #107480) is a disorder that is inherited in an autosomal dominant manner. Only one mutated copy of the affected gene is required to cause the disorder. An embryo produced using gametes from an affected parent and an unaffected parent will have a 50% chance of inheriting the condition.

4. The Committee noted that although the degree of penetrance is 100%, expression can be highly variable, but at its most serious, patients born with Townes-Brock Syndrome are diagnosed with a triad of imperforate anus, dysplastic ears and typical thumb malformation. In addition to the triad symptoms affected individuals may also exhibit congenital heart disease, renal impairment, hearing impairment, foot malformation, genitourinary malformations and in some cases mental retardation.

5. The Committee noted that there is no curative treatment for Townes-Brock Syndrome. The malformations that result from this gene mutation will be present at birth. Some problems may however develop or be detected later

– there is a significant risk that renal function may deteriorate in those with involvement until the affected individual enters chronic renal failure; learning and behavioural problems can occur and be detected later in childhood.

6. The Committee considered that the condition is serious because there is a significant risk of congenital abnormalities, including congenital heart defects, imperforate anus, dysplastic ears and typical thumb malformation, requiring invasive surgery at an early stage of life. If detected at birth, an imperforate anus would require immediate surgical intervention. Renal function needs to be regularly monitored with a significant risk that those affected will suffer chronic renal failure and need a renal transplant.

7. The Committee noted that the application is supported by the Peer Reviewer as well as by 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. Accordingly, it was appropriate to grant the application under paragraph 1ZA(1)(b) of Schedule 2 to the Act.

9. The Committee agreed to authorise the testing of embryos for Townes-Brock Syndrome (OMIM #107480). The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 12/12/2011

David Archard (Chair)