HFEA Licence Committee Meeting

30 August 2012

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

Minutes – Item 3

Centre 0044 (Centre for Reproductive and Genetic Health) – PGD for Multiple Lentigines Syndrome) (OMIM #151100)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Sue Price (professional) Debbie Barber (professional) Jane Dibblin (lay) Anna Carragher (lay) Mair Crouch (lay) – VC Rebekah Dundas (lay) – VC	Lauren Crawford Legal Adviser: Sarah Ellson, 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

- 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 Multiple Lentigines Syndrome (OMIM #151100) is a rare disorder that is inherited in an autosomal dominant manner. There is a 1 in 2 chance of an embryo being affected by this condition where 1 parent is affected.

4. The Committee noted Multiple Lentigines Syndrome (LEOPARD Syndrome) (OMIM #151100) is a multisystem disease caused by a mutation in the protein tyrosine phosphatase, non-receptor type 11 gene (PTPN11). It is a fully-penetrant condition, so the mutation will result in the manifestation of the disorder, although the syndrome has variable expressivity.

5. The Committee noted that symptoms of LEOPARD Syndromeinclude abnormal genitalia (undescended testicles cryptorchidism), absent or delayed puberty, cafe-au-lait spots (light brown birthmarks), hearing problems (partial deafness), hypogonadism, multiple spots on neck and trunk, pectus carinatum, or pectus excavatum, (abnormalities of the

sternum or breastbone) prominent ears, slow growth, wide-set eyes (hypertelorism).

6. The Committee noted that Onset of the disorder occurs in childhood. Lentigines (freckle-like spots) may be present at birth or develop during childhood, most frequently manifesting by 4-5 years of age. Although fully penetrant, the syndrome has variable expressivity, therefore one generation may have a mild expression of the syndrome while the next may be profoundly affected.

7. The Committee noted that the condition is serious because in the more extreme cases patients with LEOPARD syndrome will have significant problems related to a heart condition (especially cardiomyopathy), a level of hearing impairment that will affect many aspects of normal life and a level of intellectual disability that will affect educational and employment prospects. There is no cure for this condition. Symptoms are treated as appropriate, such as the use of hearing aids for hearing problems. Hormone treatment may be required at the expected time of puberty to cause the normal changes to occur. Laser or bleaching creams may help lighten some of the brown spots on the skin. Hearing and cardiac defects must be monitored annually. Prognosis is determined mainly by cardiac complications. LEOPARD Syndrome is a complex of features, mostly involving the skin, skeletal and cardiovascular systems, which may or may not be present in all patients.

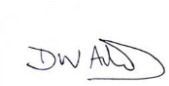
8. The Committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms and the multisystem nature of the disorder, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or a 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 noted that the Peer Reviewer had set out information as to why the genetic condition was serious and the significant problems experienced in the more extreme patient cases.

10. The Committee noted that the application is supported by the Genetic Alliance UK.

11. The Committee agreed to authorise the testing of embryos for Multiple Lentigines Syndrome (LEOPARD Syndrome) (OMIM #151100). The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 13/09/2012



David Archard (Chair)