HFEA Licence Committee Meeting

26 April 2012

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

Minutes – Item 1

Centre 0044 (Centre for Reproductive and Genetic Health) – PGD for Ellis-Van Creveld Syndrome (EVC) (OMIM #225500)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Anna Carragher (lay) Rebekah Dundas (lay) (videoconference) Mair Crouch (lay) Sue Price (professional) Debbie Barber (professional) Jane Dibblin (lay)	Joanne McAlpine Legal Adviser: Stephen Hocking, Partner, Public Law Department For DAC Beachcroft LLP

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 much 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 Ellis-Van Creveld Syndrome (EVC) (OMIM #225500) is a disorder that is inherited in an autosomal recessive manner. Where both parents carry an affected copy of the relevant genes there is a 1 in 4 chance (25% risk) of the embryo exhibiting the syndrome.

4. The Committee noted that EVC has a varying degree of penetrance and a skeletal dysplasia characterized by short limbs, short ribs, postaxial polydactyly, and dysplastic nails and teeth. Congenital cardiac defects, most commonly a defect of primary atrial septation producing a common atrium, occur in 60% of affected individuals. The condition itself is not treatable, but many of the complications are. Many babies with this condition die in early infancy, usually due to a small chest or heart defect. Stillbirth is common. The outcome depends on which body system is involved and to what extent. Intelligence is normal.

5. The Committee noted that EVC symptoms caused by this condition may include cleft lip or palate, epispadias or undescended testicle, extra fingers, limited range of motion, nail problems, including missing or deformed nails, short arms and legs, especially forearm and lower leg,

short height (between 3 ½ and 5 feet tall) sparse, absent, or fine textured hair; tooth abnormalities.

6. The Committee noted that treatment is available for EVC through surgical intervention however the condition itself is not curable and treatment is supportive only through surgery and drugs. Due to the wide range of effects, the quality of life will be diminished in many ways.

7. The Committee considered that the condition amounts to a serious physical disability because it is associated with significant risk of defects or atrial separation and restricted rib growth resulting in potentially life threatening cardiorespiratory compromise in the neonatal period.

8. The Committee noted from the application that both the Peer Reviewer and Genetic Alliance UK consider the condition to be serious.

9. The Committee had regard to its explanatory note, in particular paragraphs 5.4 *Where a condition has a range of penetrance (eg. 40-60%), the Licence Committee will base its decision on the highest penetrance figure* and 5.5 '*Where a condition has variable symptoms, the Licence Committee will base its determination of how serious the disability, illness or condition is, on the worst possible symptoms'*. The Committee noted that on the basis of the information presented, given the highest degree of penetrance and 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 another serious medical condition. Accordingly, it was appropriate to grant the application under paragraph 1ZA(1)(b) of Schedule 2 to the Act.

10. The Committee noted that while they may grant approval, the centre providing PGD for the condition must also ensure that each individual case treated fulfills the severity criteria of Schedule 2 paragraph 1ZA of the HFE Act 1990 (as amended).

11. The Committee agreed to authorise the testing of embryos for Ellis-Van Creveld Syndrome (EVC) (OMIM #225500). The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 10/05/2012

David Archard (Chair)