HFEA Licence Committee Meeting

26 April 2012

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

Minutes – Item 2

Centre 0044 (Centre for Reproductive and Genetic Health) – PGD for Noonan Syndrome (OMIM #163950)

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, 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 Noonan Syndrome (OMIM #163950) is a disorder that is inherited in an autosomal dominant manner. If one parent carries an affected copy of a relevant gene there is a 1 in 2 chance (50% risk) of the embryo exhibiting the syndrome.

4. The Committee noted that penetrance is incomplete and expression is variable and unpredictable. Some individuals may not exhibit the syndrome and others may be identified at birth depending on the severity of the symptoms.

5. The Committee noted that effects of Noonan Syndrome include delayed puberty, down slanting or wide-set eyes, variable hearing loss, low-set or abnormally, shaped ears, mild mental retardation (in about 25% of cases), drooping eyelids, short stature (can be treated with growth hormone), small penis, undescended testicles, unusual chest shape, webbed and short appearing neck.

6. The Committee noted that the Noonan Syndrome affects: the heart, gastrointestinal system, genito-urinary system and lymphatic system. Developmental problems include: musculoskeletal, hematological and neurological. While growth hormone may aid growth to normal height, many aspects will not be possible to treat.

7. The Committee noted Noonan Syndrome is not a curable condition, treatment may involve growth hormone to aid growth to normal height, however many aspects will not be treatable.

8. The Committee considered that the condition amounts to a serious physical or mental disability because 20-30% of infants with Noonan Syndrome will develop hypertrophic cardiomyopathy which may lead to early death (6-12% within 15 years of diagnosis). 10-15% of affected individuals require special education.

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

10. The Committee had regard to its explanatory note, in particular paragraphs 5.3 *Where a condition has a variability of 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.

11. 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).

12. The Committee agreed to authorise the testing of embryos for Noonan Syndrome (OMIM #163950). 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)