HFEA Licence Committee Meeting

30 June 2011

21 Bloomsbury Street London WC1B 3HF

Minutes – Item 5

Centre 0102 (Guy's Hospital) – PGD for Spinal and Bulbar Muscular Atrophy X-linked (Kennedy disease) OMIM# 313200

Members of the Committee:	Committee Secretary:
David Archard (lay) – Chair Debbie Barber (professional) Anna Carragher (lay) Mair Crouch (lay)	Terence Dourado Legal Adviser: 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:

- Executive Summary
- Application form
- Genetic Alliance opinion
- Redacted peer review

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

1. The Committee noted that a licence committee, which convened on 5th May 2011, had begun to consider the application variation in respect of PGD Spinal and Bulbar Muscular Atrophy X-linked (Kennedy disease) but that it was aware that in effect only males suffer from the condition and females carry the condition; it was concerned that the application did not make explicit whether the Centre will only seek to exclude affected male embryos from implantation, so it deferred consideration of the application for receipt of a clear explicit statement that the Centre seeks a licence variation in respect of PGD Spinal and Bulbar Muscular Atrophy X-linked (Kennedy disease), for male embryos only.

2. The Committee noted that it had since received a letter from the PR of the Centre, on 25th May 2011, confirming that 'All embryos except the affected male embryos would be considered suitable for embryo transfer as female carriers would not be expected to manifest serious symptoms of this condition'.

3. The Committee had regard to its Decision Tree. The Committee was satisfied that the Centre has considerable 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.

4. 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'.

5. The Committee noted that Spinal and Bulbar Muscular Atrophy X-linked (Kennedy disease) OMIM# 313200 is an X-linked recessive form of spinal muscular atrophy. Males only have one copy of the X chromosome and the inheritance of a copy of the recessive gene in that X chromosome will result in the manifestation of the disease. Females have two X chromosomes and inheritance of one copy of the affected gene manifests in a carrier status. Thus each son of a female carrier has a 50% chance of being affected. Each daughter has a 50% chance of being a carrier.

6. The Committee noted that there is a significant risk that a male with the abnormality will develop a serious medical condition because it is fully penetrant in males with 38 or more CAG triplet base repeats in the Androgen Receptor gene and in males who live long enough for the disease to manifest symptoms.

7. The Committee noted that the condition is clinically similar to, but genetically distinct from, classic forms of autosomal spinal muscular atrophy. The Committee considered that the condition is serious because severely affected individuals (many of whom are non-ambulatory) are at risk for asphyxiation or aspiration pneumonia because of weakness of the bulbar musculature. The Committee considered that age of onset is usually in the third to fifth decade of life, but earlier onset has been reported. The disorder is characterised by slowly progressive limb and bulbar muscle weakness with fasciculations (muscle twitches), muscle atrophy, and gynecomastia (breast enlargement in men). Furthermore, variability in disease severity and progression occurs both within and between families. However, there is no cure and progression cannot be halted.

8. On the basis of the information presented, the Committee was satisfied that there is a significant risk that a male 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 that the licence should be varied to authorise the testing of embryos for Spinal and Bulbar Muscular Atrophy X-linked (Kennedy disease) OMIM# 313200 and that no conditions should be put on the licence in relation to the variation. The Committee confirmed that this condition in affected male embryos will be added to the published list of conditions for which PGD may be carried out. The authorisation of testing for this condition is limited to affected male embryos as female carriers would not be expected to manifest serious symptoms of this condition.

Signed..... Date..13/7/2011

David Archard (Chair)