HFEA Licence Committee Meeting

27 September 2012

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

Minutes – Item 5

Centre 0102 (Guy's Hospital) – PGD for Spinocerebellar ataxia type 1 (SCA1) (OMIM#164400)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Sue Price (professional) Jane Dibblin (lay) Anna Carragher (lay) Mair Crouch (lay) – Rebekah Dundas (lay)– Videoconference	Lauren Crawford 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

- Cover sheet
- Executive Summary
- PGD Application form
- Supporting information on Ataxia
- 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 Spinocerebellar ataxia type 1 (SCA1) (OMIM#164400) is a disorder that is inherited in an autosomal dominant manner. There is a 1 in 2 chance of an embryo inheriting this condition from an affected parent.

4. The Committee noted that Spinocerebellar ataxia type 1 (SCA1) is a genetic condition which is caused by a gene change in the ATXN1 gene. Within the ATXN1 gene, a number of triplet repeat sequences are normally present. When the number of triplet repeat sequences is above 39, a person is likely to develop symptoms of SCA1. SCA1 is a progressive neurodegenerative disorder.

5. The Committee noted that the onset of symptoms is most commonly between 30 and 40 years and the time frame from onset to death is approximately 10 to 30 years.

6. The Committee noted that early symptoms include gait disturbance leading to difficulty with balance, slurred speech, swallowing difficulties, eye movement problems and brisk reflexes. With the progression of the symptoms patients will experience difficulty with eye movements and poor muscle tone leading to weakening of muscles. A number of patients will have clinical depression. In the advanced stages affected individuals develop rigidity, chorea (involuntary movements) and cognitive

impairments (in verbal skills, memory, executive function deterioration). With advancing disease swallowing becomes more difficult leading to choking and aspiration. Respiratory failure is the main cause of death. Although rare, a juvenile form of SCA1 has been seen and there is a relationship between the repeat size and the age of onset and severity of symptoms. However it is not possible to predict on an individual basis what the age of onset will be. Similar to other neurological “repeat” disorders there is evidence of anticipation, which means that the onset of SCA1 can be earlier in subsequent generations.

7. The Committee noted that the condition is serious because no curative treatment is currently available and treatment is aimed only at relieving symptoms. The disease progresses at a variable rate but usually leads to death within an average of 20 years of onset. The rate of progression may be faster in younger cases. The phenomenon of anticipation means that the age of onset tends to decrease as the gene is passed on from generation to generation so the child of an affected parent may be affected at an earlier age.

8. The Committee had regard to its explanatory note, in particular paragraph 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’*, and noted that on the basis of the information presented, given the conditions worst symptoms, it was also 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 noted that the application is supported by the Peer Reviewer and the Genetic Alliance UK.

10. The Committee agreed to authorise the testing of embryos for Spinocerebellar ataxia type 1 (SCA1) (OMIM#164400). The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 09/10/2012

David Archard (Chair)