HFEA Licence Committee Meeting

29 March 2012

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

Minutes – Item 2

Centre 0102 (Guys Hospital) – PGD for Calpainopathy previously known as Limb girdle muscular dystrophy type 2A (OMIM #253600)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Anna Carragher (lay) Rebekah Dundas (lay) (videoconference) Mair Crouch (lay) (videoconference) Sue Price (professional) Debbie Barber (professional)	Lauren Crawford Legal Adviser: Juliet Oliver, 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

- 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 Calpainopathy (OMIM #253600) is a disorder that is inherited in an autosomal recessive manner. There is a 1 in 4 chance of an embryo being affected by this condition where both parents are carriers..

4. The Committee noted that the condition is fully penetrant by adulthood. The age of onset can vary from early infancy to adulthood, with most of those affected having symptoms between 8 and 15 years. The initial symptoms are weakness and wasting (loss of muscle bulk) in the hip, thigh and shoulder muscles. This weakness is usually even on both sides of the body and the condition usually affects the legs before the shoulders and arms. The weakness can result in frequent falls, difficulty in running, climbing stairs and rising from the floor. As the condition progresses, affected individuals can have problems with walking.

5. The Committee noted that Calpainopathy is one of the most common recessively inherited forms of limb girdle muscular dystrophy. Many of those with this disorder will become wheelchair dependent. As the disorder progresses the respiratory muscles can weaken and lead to breathing difficulties in the later stages of the disorder. The impact of this can

include: poor sleep, nightmares, tiredness or headaches after waking up in the morning, lack of appetite and falling asleep during the day.

6. The Committee noted that there is no curative treatment for Calpainopathy and treatment is supportive only. Treatment requires surgery to correct foot problems and curvature of the spine. Respiratory aids may be required to help those who develop respiratory failure as the condition progresses.

7. The Committee considered that the condition is serious because the muscular dystrophy associated with the condition is progressive and untreatable, and causes severe disability. The Committee noted that Calpainopathy does not decrease life expectancy but has a high morbidity, with early onset and patients may require lifelong therapy and care and/or prolonged respiratory support. From onset, patients are significantly weakened, limiting both their choice of occupation and their recreational options. For those with early onset Calpainopathy, the condition will impact upon both their education and their early social life. For those with late onset Calpainopathy the condition's onset may cast a shadow over their future, and may cause them to second guess every instance of weakness or tiredness as a potential first symptom.

8. The Committee noted that the application is supported by the Peer Reviewer as well as by Genetic Alliance UK. The Genetic Alliance statement is supported by Muscular Dystrophy Campaign.

9. 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.

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 Calpainopathy (OMIM #253600). The Committee confirmed that this

condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 12/04/2012

David Archard (Chair)