# HFEA Licence Committee Meeting
## 30 April 2010

21 Bloomsbury Street London WC1B 3HF

### Minutes – Item 3

**Centre 0044 Centre for Reproductive and Genetic Health – Variation application to perform PGD for Central Core Disease of Muscle - OMIM# 117000**

| Members of the Committee: | Committee Secretary: |
|---|---|
| Anna Carragher (lay) – Chair<br>Debbie Barber (Professional)<br>Mair Crouch (lay) | Terence Dourado<br><br>Legal Advisers:<br>Mary Timms |

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
- Signed application form (including appendix)
- Redacted peer review
- Licence Committee minutes 25 February 2010
- New information from the centre concerning the 'seriousness' of this disease
- Clarification from the centre concerning the purpose for the testing of embryos

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

1. The Committee had regard to its Decision Tree. The Committee was satisfied that the Centre already had considerable experience of carrying out PGD and had conducted 46 PGD cycles between 1 January 2008 and 31 December 2009. Additionally, the Centre was CPA accredited in accordance with standards for the medical laboratory incorporating ISO 15189:2007 in December 2009.

2. The Committee noted that the item was considered and adjourned by the Licence Committee on 25 February 2010, as it was unable to make a decision due to "the insufficient evidence put before it; particularly in respect of the seriousness of the condition". The Committee also noted that the peer reviewer had been unable to give "…a definitive view as to the significance of the risk that a person with the abnormality would develop a serious physical or mental disability, serious illness or other serious medical condition."

3. The Committee which met on 25 February 2010, also wished to give the PR an opportunity to clarify whether they had intended to apply under 1ZA(1)(a) of schedule 2 of the Act, as it was not clarified on their initial application. This information had since been provided.

4. The Centre had since clarified that the 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 Central Core Disease of Muscle is inherited in an autosomal dominant pattern. If one parent is affected there is a 50% chance of the embryo having the condition.

6. The Committee noted that there is a significant risk that those born with the affected gene will develop an abnormality because it is nearly 100% penetrant. There is no curative treatment only supportive.

7. The Committee noted the additional information which had been received from a Consultant Paediatric Neurologist regarding the significance, risk and seriousness of the condition. The Committee noted that the condition is serious because in its worst form it can manifest as severe congenital myopathy, and may be associated with unusual growth of the hip, delayed motor milestones and scoliosis (curvature of the spine). The condition could also present with hypotonia (diminished muscle tone), weakness and ventilator dependence. The Committee also noted that infants are generally affected to the same extent as the affected parent.

8. On the basis of the information presented about the most serious form of the condition, the Committee 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 any other serious medical condition. Accordingly, it was appropriate to grant the application under 1ZA(1)(b) of Schedule 2 to the Act.

9. The Committee noted paragraph 10.5 of the Code of Practice (8th edition)/ HFEA guidance for Centres: 'The use of PGD should be considered only where there is a significant risk of a serious genetic condition being present in the embryo. When deciding if it is appropriate to provide PGD in particular cases, the seriousness of the condition in that case should be discussed between the people seeking treatment and the clinical team. The perception of the level of risk for those seeking treatment will also be an important factor for the centre to consider.' Since infants with this condition are generally affected to the same extent as the affected parent, this is something which the centres will need to take into account when deciding to provide PGD in particular cases.

10. The Committee agreed that the licence should be varied to authorise the testing of embryos for Central Core Disease of Muscle, OMIM# 117000. Although the Committee authorised the condition, it noted that the Authority would be reviewing PGDs periodically. It requests that when the Authority reviews this condition, it should take into account any clinical advances in the understanding of the Genotype/phenotype relationship. The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed..... Date...10.5.2010

Anna Carragher (Chair)