HFEA Licence Committee Meeting

25 March 2010

21 Bloomsbury Street London WC1B 3HF

Minutes – Item 9

Centre 0044 (Centre for Reproductive and Genetic Health) – Variation application to perform PGD for Hypophosphatemic Rickets: X-linked dominant (Xlh), OMIM# 307800

Members of the Committee:	Committee Secretary:
David Archard (lay) – Chair Rebekah Dundas (lay) Sue Price (Professional)	Terence Dourado Legal Advisers: 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
- Signed application form (including any appendices)
- 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

1. The Committee had regard to its Decision Tree. The Committee was satisfied that the Centre already has considerable experience of carrying out PGD and had conducted 46 PGD cycles between 1 January 2008 and 31 December 2009.

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 that Hypophosphatemic Rickets: X-linked dominant (Xlh) is inherited in an X-linked pattern but girls are also affected. In this type of inheritance a male or female inheriting the X with the genetic change would be affected i.e. there is a 50% chance of the embryo inheriting the affected gene from an affected mother. The Committee noted that the condition usually affects males more than females.

4. The Committee noted that there is a significant risk that those born with the affected gene will develop an abnormality because it is 100% penetrant.

5. The Committee noted that the condition is serious because it is characterised by growth retardation, rachitic and osteomalacic bone disease, hypophosphatemia, and renal defects in phosphate reabsorption and vitamin D metabolism. The condition usually presents with limb deformity and slow growth which may be evident within one year of age or by early childhood. The main symptoms are short stature, bowing of legs, muscle weakness and bone pain. However, craniosynostosis, dental abnormalities and loss of hearing may also occur. Males are more severely affected than females, but affected females will have some features of the condition. Treatment for the condition includes oral phosphate therapy and Vitamin D analogues. However, it is not always effective and treatment will be required indefinitely. Furthermore, treatment includes daily intramuscular injection of growth hormone and surgical limb lengthening. However, these methods are not always successful.

6. On the basis of the information presented, 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.

7. The Committee agreed that the licence should be varied to authorise the testing of embryos for Hypophosphatemic Rickets: X-linked dominant (Xlh), OMIM# 307800 and that no conditions should be put on the licence in relation to the variation. The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed..... Date.31/03/2010..

David Archard (Chair)