HFEA Licence Committee Meeting

26 August 2010

21 Bloomsbury Street London WC1B 3HF

Minutes – Item 2

Centre 0044 (Centre for Reproductive and Genetic Health CRGH) –Application for Variation to include PGD for Non-Ketotic Hyperglycinaemia (NKH)/ Glycine Encephalopathy (GCE) OMIM# 605899)

Members of the Committee: Anna Carragher (lay) – (Chair) Jane Dibblin (lay) Sally Cheshire (lay) Sue Price (Professional) Debbie Barber (Professional) Mair Crouch (lay) Apologies: Rebekah Dundas (lay)	Committee Secretary: Joanne McAlpine Legal Advisers: Sarah Ellson – Field Fisher

Declarations of Interest: members of the Committee declared that they had no conflicts of interest in relation to this item; Debbie Barber noted that she works in a licensed centre.

The following papers were considered by the Committee:

- Executive summary
- Application form (including 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)
- HFEA (Licence Committees and Appeals) Regulations 1991 (SI 1991/1889)
- Decision trees for granting and renewing licences and considering requests to vary a licence (including the PGD decision tree)
- Guidance for members of Authority and Committees on the handling of conflicts of interest approved by the Authority on 21 January 2009.
- 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 has been licensed to perform PGD for a number of years and conducted a total of 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, i.e. ' 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 Non-Ketotic Hyperglycinaemia (NKH)/ Glycine Encephalopathy (GCE) is an autosomal recessively inherited condition, so each embryo has a 25% chance of inheriting mutations from both parents and being affected.

4. The Committee noted that the condition is thought to be 100% penetrant and mostly presents in the first few days of life, causing a significant risk that a person with the abnormality will develop a serious medical condition.

5. The Committee considered that the condition is serious. The condition, Non-Ketotic Hyperglycinaemia (NKH) / Glycine Encephalopathy (GCE), is not treatable. Affected neonates deteriorate and die within a few days of birth. Those who survive the neonatal period develop intractable seizures and profound mental retardation. In the infantile form GCE, patients present with seizures and have various degrees of mental retardation after a symptom-free interval and seemingly normal development for up to 6 months.

6. The Committee noted that there are many long term problems for those who survive beyond the neonatal period. Even children with milder variants of GCE have an impaired developmental quotient. Feeding difficulties and gastro-oesophageal reflux are common with many requiring gastrostomy tubes for feeding. Skeletal problems include hip dislocation, scoliosis and osteoporosis. Treatment options are limited; management with antiepileptic drugs is complicated. Sodium valporate is contraindicated as it raises blood and CSF glycine which can increase seizure frequency. If adequate seizure control is achieved this tends to favour developmental progress. Swallowing dysfunction is often seen, as is gastro-oesophageal reflux. Gastrostomy tube placement and a Nissens procedure are often required early on in management.

The Committee's Decision

7. The Committee agreed that 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 of the Act.

8. The Committee agreed that the licence should be varied to authorise the testing of embryos for Non-Ketotic Hyperglycinaemia (NKH)/ Glycine Encephalopathy OMIM# 605899, and that no conditions should be put on the licence in relation to this variation. The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Anna Carragher Date: 15.9.2010.

Anna Carragher (Chair)