HFEA Statutory Approvals Committee

30 January 2014

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

Minutes – Item 5

Centre 0044 (The Centre for Reproductive and Genetic Health (CRGH)) – PGD application for (PIGN gene) Multiple Congenital Anomalies Hypotonia – Seizures Syndrome 1 OMIM #614080

Members of the Committee: David Archard (lay) Chair Rebekah Dundas (lay) Sue Price (professional) Hossam Abdalla (professional) Jane Dibblin (lay) Debbie Barber (professional) Advisor: Dr Peter Turnpenny	Committee Secretary: Lauren Crawford Legal Adviser: Graham Miles, Morgan Cole Observing: Sam Hartley, Head of Governance and Licensing, HFEA

Declarations of Interest: The Members declared no conflicts in relation to this item.

The following papers were considered by the Committee:

- Executive summary
- Application form

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) ("the Act")
- 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 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 Multiple congenital anomalies hypotonia – seizures syndrome 1 (PIGN gene) (OMIM #614080) is inherited in an autosomal recessive pattern and there is a 1 in 4 chance of an embryo being affected with the condition where both parents are carriers.

4. The Committee noted that Multiple congenital anomalies hypotonia – seizures syndrome 1 (PIGN gene) is a rare condition caused by a mutation in the PIGN (phosphatidylinositol glycan anchor biosynthesis, class N) protein-coding gene. Affected children have multiple, variable congenital abnormalities, epileptic seizures and severe development delay.

5. The Committee noted that symptoms of the condition are apparent at birth and can include blindness, deafness and seizures. Babies can be born with severe hypotonia, a loss of muscle tone which results in babies having little control of their neck muscles and being 'floppy' in appearance. The condition affects a child's ability to walk, eat, speak and swallow. Children may have missing teeth, toe and finger nails and structural heart abnormalities.

6. The Committee noted that there is no cure for Multiple congenital anomalies hypotonia – seizures syndrome 1 (PIGN gene) and no remedial treatments available.

7. The Committee noted that the application did not contain a Peer Review or Genetic Alliance Opinion but were satisfied agreed that the information in paperwork was sufficient for them to consider the item in their absence.

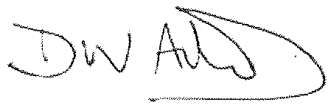
8. The Committee welcomed the advice of its Advisor, Dr Peter Turnpenny, who confirmed that the condition was as described in the papers.

9. The Committee considered that the condition is serious because this is a multi-system disorder, having profound effects on physical and mental function. Affected children will need special care and there are no available treatments.

10. The Committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it 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. The Committee was therefore satisfied that the condition meets the criteria for testing under paragraph 1ZA(1)(b) of Schedule 2 to the Act.

11. The Committee agreed to authorise the testing of embryos for Multiple congenital anomalies hypotonia – seizures syndrome 1 (PIGN gene) (OMIM #614080).

Signed: Date: 13/02/2014



David Archard (Chair)