HFEA Statutory Approvals Committee

30 January 2014

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

Minutes – Item 3

Centre 0206 (Reproductive Genetics Institute (RGI)) – PGD application for Congenital Myasthenic Syndrome (COLQ gene 603033) (Type Ic) OMIM #603034

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
- 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) ("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 Congenital Myasthenic Syndrome (COLQ gene 603033) (Type Ic) OMIM #603034 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 typically, affected patients suffer from a severe muscle weakness, which is progressive with onset in childhood. Most patients are severely disabled from by the second decade with difficulty breathing and severe scoliosis. The disorder is caused by mutations in the COLQ gene. Disruption of the gene leads to a reduction in the amount of acetylcholinesterase, an enzyme needed at neuromuscular junctions. As a result, acetylcholine is not broken down so signalling between nerve and muscle cells is prolonged. This signalling overload can damage muscle cells, leading to the characteristic muscle weakness. There is some variability ranging from exercise intolerance to severe disability. Mutations of the COLQ gene are typically associated with significant physical impairment due to extreme muscle weakness, respiratory problems and scoliosis.

5. The Committee noted that symptoms may present at birth, although later onset forms have been described. Most patients are severely disabled by late childhood or adolescence, and symptoms are progressive.

6. The Committee noted that there are no known treatments for Congenital Myasthenic Syndrome (COLQ gene 603033) (Type Ic).

7. The Committee noted that the application was supported by the Peer Reviewer.

8. The Committee noted that the application did not contain a Genetic Alliance Opinion but were satisfied and agreed that the information in paperwork was sufficient for them to consider the item in its absence.

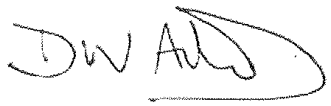
9. The Committee welcomed the advice of its Advisor who confirmed that the condition was as described in the papers.

10. The Committee considered that the condition is serious because affected individuals have significant physical impairment due to extreme muscle weakness. Respiratory problems and scoliosis.

11. 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.

12. The Committee agreed to authorise the testing of embryos for Congenital Myasthenic Syndrome (COLQ gene 603033) (Type Ic) OMIM #603034.

Signed: Date: 13/02/2014



David Archard (Chair)