HFEA Statutory Approvals Committee

27 February 2014

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

Minutes – Item 9

Centre 0006 (The Lister Fertility Clinic) – PGD application for Cohen Syndrome OMIM #216550

Members of the Committee: David Archard (lay) Chair Rebekah Dundas (lay) Jane Dibblin (lay) Debbie Barber (professional) Advisor: Dr Mary Porteous	Committee Secretary: Lauren Crawford Legal Adviser: Stephen Hocking, DAC Beachcroft Also in attendance: Sam Hartley, Head of Governance and Licensing, HFEA

Declarations of Interest: Hossam Abdalla left the meeting before this item as he is the PR (Person Responsible) at this centre. The Members present declared no conflicts in relation to this item.

The following papers were considered by the Committee

- Executive summary
- Application form
- Redacted peer review
- Tabled Genetic Alliance Opinion

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 Cohen Syndrome (OMIM #216550) 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 the first symptom of Cohen syndrome is often difficulty feeding for the newborn. This occasionally can result in the necessity for a tube to be surgically inserted directly in the baby's stomach to deliver food. (This is called percutaneous endoscopic gastrostomy (PEG) feeding.) Children with Cohen syndrome have global developmental delay. They are slow to reach milestones of development such as crawling and walking. Children with Cohen syndrome are slow to learn to talk. Some do not achieve speech at all, and some do not progress past single words. All children with Cohen syndrome have learning difficulties, often needing to attend specialist schools.

5. The Committee noted that nearly all children with Cohen syndrome have sight problems that start with short-sightedness at a young age and become increasingly severe. Though complete blindness is unlikely, people with Cohen syndrome frequently develop profound myopia, retinal degeneration, and weakness of muscles around and in the eye. These impairments tend to

result in a major sight disability. Children and adults with Cohen syndrome have particular behavioural characteristics, the most prominent of which being their cheery positive disposition. The other characteristics are similar to autism spectrum disorder.

6. The Committee noted that there is no curative treatment for this disorder. There are a few things that can be done, but these can only have a small impact on the main features of the disorder. Treatment includes: spectacle correction of refractive errors, low-vision training for the visually impaired. Early intervention and physical, occupational and speech therapy help address developmental delay, hypotonia, joint hyperextensibility, and motor clumsiness. Recurrent infections are treated per standard therapy.

7. The Committee noted that the application is supported by the Peer Reviewer and the Genetic Alliance.

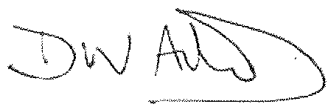
8. The Committee welcomed the advice of its Advisor, Mary Porteous, who confirmed that the condition was as described in the papers.

9. The Committee considered that the condition is serious because severely impacts the quality of life for affected individuals and they will require lifelong support.

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) and (2) of Schedule 2 to the Act.

11. The Committee agreed to authorise the testing of embryos for Cohen Syndrome (OMIM #216550).

Signed: Date: 13/03/2014



David Archard (Chair)