HFEA Statutory Approvals Committee

28 November 2013

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

Minutes – Item 2

Centre 0102 (Guy's Hospital) – PGD application for Treacher Collins Syndrome Type 2 OMIM #613717

Members of the Committee: David Archard (lay) Chair Sue Price (professional) Hossam Abdalla (professional) Debbie Barber (professional) Rebekah Dundas (lay) Apologies: Jane Dibblin (lay) Advisor: Mary Porteous	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
- Email from Genetic Counsellor
- Redacted peer review
- 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 Treacher Collins Type 2 (TCS2) (OMIM #613717) is inherited in an autosomal dominant pattern and there is a 1 in 2 chance of an embryo being affected with the condition where one parent is affected.

4. The Committee noted that TCS2 is a craniofacial disorder where the affected patient is born with a malformed skull, cheekbones, chin, nose and jaw. The ears may be malformed or completely missing and eyelids may slant downwards making the eyes seem unsupported. In a worst case scenario, the affected patient can have breathing problems caused by tissue blocking the back of the nasal passage and bilateral hearing loss due to the abnormalities with the external auditory canal.

5. The Committee noted that TCS2 is almost 100% penetrant, but that the condition is variable, where some patients with the inherited mutation causing the condition are only mildly affected, with no obvious signs of the condition. Other patients may be more severely affected with serious facial abnormalities and life threatening breathing problems.

6. The Committee noted that the variability of the condition means that treatment types are tailored to the individual patient but usually involve management by a multidisciplinary team including geneticists, plastic surgeons, head and neck

surgeons and orthodontists. If a patient is born with the condition, the blockage or narrowing of the airway can cause neonatal death and airway management is required. Between the ages of 3 and 12 years, speech therapy takes place and from 13 to 18 years of age, craniofacial reconstruction takes place which could include ear reconstruction.

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

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

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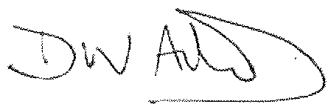
9. The Committee considered that the condition is serious because affected individuals may need a number of invasive operations to correct or alleviate congenital abnormalities.

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 Treacher Collins Type 2 (OMIM #613717).

Signed: Date: 09/12/2013



David Archard (Chair)