HFEA Licence Committee Meeting

1 March 2012

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

Minutes – Item 3

Centre 0078 (IVF Hammersmith) – PGD for Seathre-Chotzen (OMIM #101400)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Anna Carragher (lay) Rebekah Dundas (lay) (videoconference) Mair Crouch (lay) (videoconference) Sue Price (professional) Jane Dibblin (lay)	Lauren Crawford Legal Adviser: Tom Rider, Field Fisher Waterhouse

Declarations of Interest: members of the Committee declared that they had no conflicts of interest in relation to this item.

The following papers were considered by the Committee

- Cover sheet
- Executive Summary
- PGD Application form: Ref 1467
- 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)
- 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 much 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 Centre's proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of schedule 2 of the Act, ie. '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 Seathre-Chotzen (OMIM #101400) is a disorder that is inherited in an autosomal dominant manner. There is a 1 in 2 chance of an embryo being affected by this condition.

4. The Committee noted that this condition is fully penetrant but has considerable variation in expression. Seathre-Chotzen is caused by a mutation in the TWIST gene. It is characterised by a premature fusion of skull bones. This early fusion prevents the skull from growing normally, which leads to irregular head shapes and progressive distortion of the face. Other commonly observed symptoms include deviated septum, vertebral abnormalities, fusion of the skin between the fingers and jaw malformation and dental problems. In some cases sufferers can present with moderate mental retardation, short stature, hearing loss, heart defects, intestinal malformation, eye malformation and sight problems.

5. The Committee noted that there is no curative treatment for Seathre-Chotzen. Surgical interventions are required to correct the various malformations caused by the condition, such as cranioplasty, mid facial and othognathic surgery. Many of these are major procedures that are not without risk. As some of the malformations are progressive and occur with growth, surgery is required during the patient's childhood and teenage

years. They may also require ophthalmologic evaluation, orthodontic treatment and treatment for hearing loss.

6. The Committee considered that the condition is serious because the disease can have a significant effect on the quality of life due to the prolonged hospital admissions for treatment, physical appearance and functional limitations if the hands are significantly affected or of there is intellectual disability. Whilst many complications are amenable to surgery, the treatment may have to be staged involving multiple and prolonged hospital admissions and the outcome can be sub-optimal with continuing cosmetic deformity.

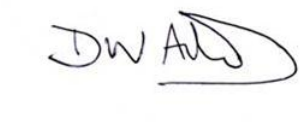
7. The Committee noted that the application is supported by the Peer Reviewer as well as by Genetic Alliance UK.

8. The Committee had regard to its explanatory note and noted that on the basis of the information presented, given the conditions 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. Accordingly, it was appropriate to grant the application under paragraph 1ZA(1)(b) of Schedule 2 to the Act.

Decision

9. The Committee agreed to authorise the testing of embryos for Seathre-Chotzen (OMIM #101400). The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 13/03/2012



David Archard (Chair)