HFEA Licence Committee Meeting

27 September 2012

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

Minutes – Item 4

Centre 0044 (Centre for Reproductive and Genetic Health) – PGD for Osteopetrosis with renal tubular acidosis (OPTB3) (OMIM#259730)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Sue Price (professional) Jane Dibblin (lay) Anna Carragher (lay) Mair Crouch (lay) Rebekah Dundas (lay) – videoconference	Lauren Crawford Legal Adviser: Graham Miles, Morgan Cole

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
- 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 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 Osteopetrosis with renal tubular acidosis (OPTB3) (OMIM#259730) is a disorder that is inherited in an autosomal recessive manner. There is a 1 in 4 chance of an embryo inheriting this condition where both parents are carriers.

4. The Committee noted that the mean age of diagnosis is 24 months. The condition begins very early in life, is fully penetrant and severe, most untreated children die in the first decade as a complication of bone marrow suppression. Diagnosis is largely based on clinical and radiographic evaluation, confirmed by gene testing.

5. The Committee noted that symptoms are physical and mental; primary neurodegeneration, mental retardation, skin and immune system involvement, renal tubular acidosis, anaemia, hepatosplenomegaly, failure to thrive, and recurrent infections are common. Associated congenital abnormalities are found in about 26% of affected individuals. Deafness and, hydrocephalus are relatively less common. Intrauterine growth is normal, but metabolic acidosis may already be evident in the neonatal period.

6. The Committee noted that haematopoietic stem cell transplantation is considered for the most severe forms associated with bone marrow failure and currently offers the best chance of longer-term survival in this group.

Further understanding of the molecular pathogenesis of these conditions will reveal new targets for pharmacotherapy.

7. The Committee noted that the condition is serious because quality of life is profoundly affected in most cases within the first 10 years. This is mainly due to bone disease, fractures and learning disability. Treatment with bone marrow transplantation may be undertaken. There is significant early mortality.

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

9. The Committee had regard to its explanatory note, in particular paragraph 5.5 '*Where a condition has variable symptoms, the Licence Committee will base its determination of how serious the disability, illness or condition is, on the worst possible symptoms'*, and noted that on the basis of the information presented, given the conditions worst symptoms, it was also 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.

10. The Committee agreed to authorise the testing of embryos for Osteopetrosis with renal tubular acidosis (OPTB3) (OMIM#259730). The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 09/10/2012

David Archard (Chair)