HFEA Licence Committee Meeting

1 March 2012

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

Minutes – Item 5

Centre 0201 (Assisted Conception Centre, Edinburgh) – PGD for Renal Coloboma Syndrome (OMIM #120330)

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 1519
- 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 that Renal Coloboma Syndrome (OMIM #120330) 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 Renal Coloboma Syndrome is a disorder caused by a mutation in the PAX2 gene. It primarily affects kidney and eye development. The condition is highly variable with high variation in penetrance, severity, age of onset and rate of progression. The application states that 33% of those persons with a PAX2 mutation exhibit hypoplastic kidneys and 100% of those with hypoplastic kidneys go on to develop renal insufficiency and failure. Visual acuity is reduced in one or both eyes in 75% of individuals, which can be serious, including blindness.

5. The Committee noted that there is no curative treatment for Renal Coloboma Syndrome. Management is focused on preventing complications of end stage renal failure and/or vision loss. End stage renal disease requires treatment with dialysis and/or renal transplantation.

6. The Committee considered that the condition is serious. Affected individuals have congenital renal problems and 30% develop renal failure

which would require dialysis or transplant. There is no curative treatment available, only renal replacement therapies.

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, which states that where a condition has variable symptoms, the Committee will base its determination of the seriousness of the condition on the worst possible symptoms. On this basis, 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. 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 Renal Coloboma Syndrome (OMIM #120330). 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)