HFEA Licence Committee Meeting

7 March 2013

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

Minutes – Item 2

Centre 0044 (The Centre for Reproductive and Genetic Health) – PGD for Maple Syrup Urine Disease OMIM # 248600

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford
Rebekah Dundas (lay)	
Debbie Barber (professional)	
Jane Dibblin (lay)	Legal Adviser:
Andy Greenfield (lay)	Juliet Oliver, Field Fisher
Bishop Lee Rayfield	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
- Redacted Peer Review

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 supporting documents with this application did not contain a Genetic Alliance Opinion. The Committee were satisfied that they had, in any event, sufficient information before them on which to make a decision.

3. 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'.

4. The Committee noted that Maple Syrup Urine Disease (MSUD), OMIM #248600 is inherited in an autosomal recessive manner. There is therefore a 1 in 4 chance of the embryo being affected by this condition where both parents are carriers..

5. The Committee noted the symptoms of MSUD include feeding difficulties, lethargy, seizures, coma, urine that smells like maple syrup and vomiting. Patients with MSUD cannot break down the amino acids leucine, isoleucine and valine, which leads to a build-up of these chemicals in the blood.

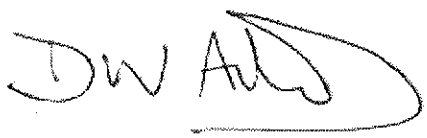
6. The Committee noted that treatment of MSUD requires a special diet which avoids consumption of leucine, isoleucine and valine. The diet is very low in natural protein, with supplements of all the other amino acids. Patients must remain on this diet permanently in order to avoid nervous system (neurological) damage. Frequent blood tests and close supervision by a registered dietician and physician are necessary. Due to the risk of acute illness (that could lead to coma and brain damage) during infections, no dietary protein is consumed at these times and an oral 'emergency regimen' is undertaken. If this is vomited or the patient deteriorates, hospital admission is needed for intravenous management and potentially for dialysis.

7. The Committee considered that the condition is serious because the age of onset for this condition is within two weeks of birth. If untreated the condition is fatal in early life. At times of physical stress such as not eating for a long time, or disease, including even minor infections, there is a risk of acute illness that could lead to coma, encephalopathy and brain damage. Most patients with MSUD have some learning difficulties and many have neurological problems affecting mobility. Even with dietary treatment (described above), stressful situations and illness can still cause high levels of certain amino acids, which may lead to death.

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 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.

10. The Committee agreed to authorise the testing of embryos for Maple Syrup Urine Disease (MSUD), OMIM #248600. The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed:  Date: 26/03/2013

David Archard (Chair)