HFEA Licence Committee Meeting

15 January 2012

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

Minutes – Item 3

Centre 0201 (Edinburgh Assisted Conception Unit) PGD for Nephrogenic Diabetes Insipidus (OMIM # 304800)

Members of the Committee: David Archard (lay) Chair Sue Price (professional) Debbie Barber (professional) Jane Dibblin (lay) Rebekah Dundas (lay)	Committee Secretary: Lauren Crawford Legal Adviser: Juliet Oliver, 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
- Redacted Peer Review

The Committee also had before it

1. HFEA Protocol for the Conduct of Licence Committee Meetings and Hearings
2. 8th edition of the HFEA Code of Practice
3. Human Fertilisation and Embryology Act 1990 (as amended)
4. Decision trees for granting and renewing licences and considering requests to vary a licence (including the PGD decision tree); and
5. Guidance for members of Authority and Committees on the handling of conflicts of interest approved by the Authority on 21 January 2009.
6. Guidance on periods for which new or renewed licences should be granted
7. Standing Orders and Instrument of Delegation
8. Indicative Sanctions Guidance
9. HFEA Directions 0000 – 0012
10. Guide to Licensing
11. Compliance and Enforcement Policy
12. Policy on Publication of Authority and Committee Papers

13. HFEA Pre-Implantation Diagnostic Testing ("PGD") Explanatory Note For Licence Committee

Background

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 considerable 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)(c) of schedule 2 of the Act, ie. 'to establish the sex of the embryo in case where there is a particular risk that any resulting child will have or develop a gender-related serious physical or mental disability, serious illness or serious medical condition' and also paragraph 1ZA(1)(b) of Schedule 2 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 the supporting documents with this application did not contain a Genetic Alliance Opinion. The Committee were satisfied that the application form and Peer Review contained sufficient information to enable them to make a decision.

4. The Committee noted that Nephrogenic Diabetes Insipidus (NDI) (OMIM # 304800) is an X-linked disorder that is inherited in a recessive manner. This means for male embryos there is a 1 in 2 chance of the embryo being affected and for female embryos there is a 1 in 2 chance of the embryo being a carrier.

5. The Committee noted that Nephrogenic Diabetes Insipidus (NDI) is usually congenital, or presents in the first few days or weeks of life. In utero presentation may also occur, and more rarely a diagnosis is not made until later infancy/childhood. Female carriers may have no symptoms or be very mildly affected. In rare instances females may present with severe manifestations of the disease.

6. The Committee noted that NDI causes an inability to concentrate urine normally, which usually presents as dehydration in the first few weeks of life. All individuals affected by NDI will have a high urine output and a resulting need to drink large volumes of fluid. If a high enough fluid intake is not maintained, the resulting dehydration can be life-threatening. NDI may be associated with short stature, perhaps as a result of chronic dehydration, or

because of reduced nutrition secondary to the high volume of water that is required. Rapid shifts in sodium level may lead to seizures, and chronic hypernatraemia (an excessive concentration of sodium in the blood plasma) can cause intellectual impairment, although this is now rare. Sleep can be disrupted and toilet training delayed because of the need to urinate during the night. Frequent urination (polyuria) may result in urinary tract complications, such as hydronephrosis (stretching of one or both kidneys due to excess build-up of urine), hydroureter (distension of the ureter with fluid) and megacystitis (an abnormally enlarged bladder). These complications may result in pain, infection and renal failure, which has been reported as early as the teenage years.

7. The Committee noted that ongoing management of NDI involves frequent monitoring of serum electrolytes and renal function. Polyuria and subsequent polydipsia (excessive thirst) may be reduced by a low-sodium diet and medications such as thiazide diuretics and non-steroidal anti-inflammatory drugs, but this does not effect a cure and typically has a maximum of 50% effect. Free access to water and toilet facilities for frequent urination are also required by individuals with NDI. The disorder has a significant effect on quality of life as well as long term health for some individuals. The Committee noted that those with the condition were particularly vulnerable in hot climates or if unwell or pyrexic.

8. The Committee considered that NDI is serious because renal failure may occur in a small number of individuals. However all affected individuals will have severe polydipsia and polyuria. Seizures and coma may also occur. Emergency treatment of dehydration is required to prevent serious complications.

9. The Committee noted that the purpose for the proposed testing is to establish whether an embryo has an x-linked condition. It 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 male 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 for one or both of the purposes specified under paragraph 1ZA(1)(b) and (c) of Schedule 2 to the Act.

10. The Committee agreed to authorise the testing of embryos for Nephrogenic Diabetes Insipidus (NDI) (OMIM # 304800). The Committee confirmed that this authorisation will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 15/02/2013

David Archard (Chair)