HFEA Licence Committee Meeting

21 February 2013

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

Minutes – Item 3 (taken as last item due to conflicts)

Centre 0035 (Oxford Fertility Unit) PGD for Conradi-Hunermann-Happle Syndrome (OMIM # 302960)

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Rebekah Dundas (lay) Jane Dibblin (lay) Andy Greenfield (lay) Bishop Lee Rayfield	Lauren Crawford Legal Adviser: Juliet Oliver, Field Fisher Waterhouse

Declarations of Interest: Debbie Barber declared an interest as she works part time at the Oxford Fertility Unit. The Chair noted this and Ms Barber recused herself from hearing this item.

The following papers were considered by the Committee

- Cover sheet
- Executive Summary
- PGD Application form
    - Additional Information
    - Patient Information Sheet
    - Patient Consent Form
- Redacted Peer Review
- Genetic Alliance Opinion

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 proposed three purposes for testing the embryos: (1) 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'; (2) as set out in 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'; and (3) as set out in paragraph 1ZA(1)(a) of Schedule 2 ie 'whether the embryo has a gene, chromosome or mitochondrion abnormality that may affect its capacity to result in a live birth'.

3. The Committee noted that Conradi-Hunermann-Happle Syndrome (OMIM #302960) is inherited in a dominant X-linked manner and results from mutation of the emopamil binding protein (EBP) gene on the short arm of the X chromosome.

4. The Committee noted that most patients with Conradi-Hunermann-Happle Syndrome are girls with a heterozygous EBP mutation. The EBP mutations that underlie Conradi-Hunermann-Happle Syndrome are generally lethal in male foetuses, which have only one X chromosome and thus no compensatory activity from a normal EBP gene on a second X chromosome. The implication of this inheritance pattern is that the woman in any couple requesting Conradi-Hunermann-Happle Syndrome testing for PGD will usually have the condition. The couple have a 25% chance of conceiving an affected male fetus, which will miscarry, and a 25% chance of conceiving an affected female, who will have the condition.

5. The Committee considered that Conradi-Hunermann-Happle Syndrome is serious. CHHS causes growth deficiency, and asymmetric short limbs. Girls are born with short limbs, and grow slowly. The majority of girls with CHHS have or develop cataracts or other eye problems such as underdeveloped eyes. In the vast majority of cases, male fetuses with this condition are not viable.

6. The Committee noted that symptoms include skin abnormalities (ichthyosis, unusual pigmentation, erythroderma, alopecia), skeletal malformations (limb abnormalities, discrepant length, vertebral anomalies, scoliosis, rhizomelic short stature, abnormal calcification, joint contractures), ophthalmological abnormalities (cataracts, microphthalmia, visual defects) and mildly dysmorphic features.

7. The Committee noted that the severity of symptoms cannot be reliably predicted and symptoms can vary between affected families and even within affected individuals in the same family. The onset of the condition is usually in infancy, but some babies in-utero may display signs of anaemia.

8. The Committee noted that there is no curative treatment for Conradi-Hunermann-Happle Syndrome. Management is supportive; e.g. surgery for limb abnormalities, cataracts and scoliosis and for management of skin and hair abnormalities. Visual problems are progressive and limb anomalies and skin manifestations can be difficult to manage.

9. The Committee noted that the purpose for the proposed testing is to establish whether the embryo has an x-linked condition. The Committee noted that Conradi-Hünnermann-Happle syndrome (CHHS) is an inherited condition causing miscarriage of male fetuses and therefore affects the embryos capacity to result in a live birth. It was satisfied that the condition in males was extremely serious.

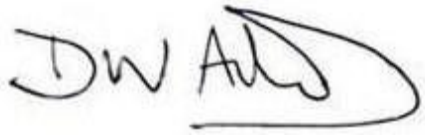
10. It noted that the condition affects the male sex significantly more than the female sex. It noted that it causes a multisystem condition in girls which may manifest with a number of symptoms and abnormalities which, cumulatively, can have significant impact on quality of life. The Committee had regard 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 female person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition.

11. The Committee was therefore satisfied that the condition meets the criteria for testing for one or more of the purposes specified under paragraph 1ZA(1)(a) to (c) of Schedule 2 to the Act.

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

13. The Committee agreed to authorise the testing of embryos for Conradi-Hunermann-Happle Syndrome (OMIM #302960). The Committee confirmed that this authorisation will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 06/03/2013



David Archard (Chair)