HFEA Licence Committee Meeting

30 August 2012

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

Minutes – Item 1

Centre 0102 (Guy's Hospital) – PGD for X-Linked Emery-Dreifuss Muscular Dystrophy - OMIM #310100

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair Sue Price (professional) Debbie Barber (professional) Jane Dibblin (lay) Anna Carragher (lay) Mair Crouch (lay) – VC Rebekah Dundas (lay) – VC	Lauren Crawford Legal Adviser: Sarah Ellson, 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
- Additional Information - (referenced in App 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'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 X-Linked Emery-Dreifuss Muscular Dystrophy – (OMIM #310100) is a disorder that is inherited in an X-linked manner. Couples with a family history of the condition have a 1 in 4 risk in each pregnancy of conceiving an affected child (half of all male children).

4. The Committee noted that the centre's application was to test for two purposes under Schedule 2, both to establish the sex of the embryo and for thefamilial genetic change. It also noted the application states "Our PGD analysis will determine which embryos are affected or unaffected males and which are carrier and non-carrier females. All embryos except affected males will be considered for embryo transfer."

5. The Committee considered that there is a significant risk that a male with the abnormality will develop a serious medical condition because it is fully penetrant in affected males.

6. The Committee noted that Emery-Dreifuss Muscular Dystrophy (EDMD) is an inherited condition affecting boys. Cardiac problems can sometimes be the first presentation of the condition, but generally occur from the second decade, and surveillance may begin at 10-12 years of age, with most requiring a permanent pacemaker by the age of 30. Patients will need lifelong surveillance for cardiac involvement with the risk of sudden death.

7. The Committee noted EDMD is characterised by a combination of joint contractures, especially of the elbow and ankles, which start in early childhood, slowly progressive muscle weakness and wasting, mainly affecting the upper arms and lower legs, which can later extend to the shoulder girdle and pelvis muscles in addition to the, potentially lethal cardiac involvement cited above which manifests as palpitations, irregular heart beat and congestive heart failure.

8. The Committee considered that EDMD is serious because the age of onset is usually early childhood. There is inter and intra-familial variation with some cases presenting later and progressing more slowly. Symptoms may be helped by surgery for the contractures and scoliosis, and various anti-arrhythmic drugs or an implantable cardiac pacemaker but there is no known cure for this condition. This is a degenerative condition with usually slow progression in the first 3 decades after which it becomes more rapid. Severe contractures may prevent walking in time. Respiratory failure and sudden cardiac death are also well reported.

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 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 satisfied that this X-linked condition affects males significantly more than females accordingly, it was appropriate to grant the application under paragraph 1ZA(1)(b) and (c) of Schedule 2 to the Act.

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

11. The Committee agreed to authorise the testing of male embryos for X-Linked Emery-Dreifuss Muscular Dystrophy – (OMIM #310100). The Committee confirmed that this authorisation will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 13/09/2012

David Archard (Chair)