HFEA Licence Committee Meeting

26 August 2010

21 Bloomsbury Street London WC1B 3HF

Minutes – Item 3

Centre 0102 (Guys Hospital) –Application for Variation to include PGD for Pachyonychia Congenita Type 1 OMIM# 167200

Members of the Committee: Anna Carragher (lay) – (Chair) Jane Dibblin (lay) Sally Cheshire (lay) Sue Price (Professional) Debbie Barber (Professional) Mair Crouch (lay) Apologies: Rebekah Dundas (lay)	Committee Secretary: Joanne McAlpine Legal Advisers: Sarah Ellson – Field Fisher

Declarations of Interest: members of the Committee declared that they had no conflicts of interest in relation to this item, Debbie Barber noted that she works in a licensed centre.

The following papers were considered by the Committee:

- Executive summary
- Application form with supporting statement from genetics counsellor
- Lay summary of condition
- Redacted peer review
- Genetics Alliance UK 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)
- HFEA (Licence Committees and Appeals) Regulations 1991 (SI 1991/1889)
- Decision trees for granting and renewing licences and considering requests to vary a licence (including the PGD decision tree)
- Guidance for members of Authority and Committees on the handling of conflicts of interest approved by the Authority on 21 January 2009.
- 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

1. The Committee had regard to its Decision Tree. The Committee was satisfied that the Centre has been licensed to perform PGD for a number of years and had considerable experience.

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, i.e. ' 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 Pachyonychia Congenita Type 1 is a rare genetic condition that is autosomal dominant with a 50% chance of an embryo inheriting the mutation.

4. The Committee noted that the condition is 100% penetrant and mostly presents in the first few days of life.

5. The Committee considered that the condition is serious. The condition Pachyonychia Congenita Type 1 is a rare genetic condition with variable presentation, involving the skin, nails, hair, teeth and some other structures. The most striking feature tends to be thickened skin (keratoderma) of the hands and feet, and often blistering of the palms and soles. The keratoderma and blistering of feet can be painful, making it difficult for some patients to walk, sometimes to the point of being wheelchair-dependent. Cysts and painful nail infections are other causes of pain. The keratoderma has a tendency to become infected, requiring extensive treatment such as drug therapy or surgical excision of the keratoderma. However, treatment is not effective at preventing the symptoms, which can cause prolonged pain and disability.

6. Alongside the daily pain and frequent cleaning and treating of infections, and cracked calluses and blisters; the combination of tongue discolouration, fingernail abnormalities, and patients' distinctive gait create an unusual appearance that can damage the self-esteem of patients throughout their lives, and especially during their childhood and teenage years. In adulthood patients may be socially isolated or stigmatised and have difficulty in forming relationships.

7. The Committee noted the letter from the Genetics Counsellor in particular paragraph four which states ' In the interim and foreseeable future, patients with pachyonychia congenita are going to suffer from prolonged pain associated with this often disabling and life disrupting condition.'

The Committee's Decision

8. The Committee agreed that on the basis of the information presented, the Committee 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 1ZA(1)(b) of Schedule 2 of the Act.

9. The Committee agreed that the licence should be varied to authorise the testing of embryos for Pachyonychia Congenita Type 1 OMIM# 167200, and that no conditions should be put on the licence in relation to this variation. The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Anna Carragher Date: 15.9.2010

Anna Carragher (Chair)