HFEA Statutory Approvals Committee

11 December 2014

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

Minutes – Item 2

Centre 0102 (Guys Hospital) – PGD application for non-Herlitz Junctional Epidermolysis Bullosa OMIM #226650

Members of the Committee: David Archard (lay) Chair Sue Price (professional) Debbie Barber (professional) Advisor: Dr Edward Blair	Committee Secretary: Trent Fisher Legal Adviser: Graham Miles (Blake Morgan) Also in attendance: Sam Hartley, Head of Governance and Licensing, HFEA

Declarations of Interest: The Members declared no conflicts in relation to this item

The following papers were considered by the Committee:

- Executive summary
- Application form
- Redacted peer review
- Genetic Alliance 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) ("the Act")
- 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 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 non-Herlitz Junctional Epidermolysis Bullosa (OMIM #226650) is inherited in an autosomal recessive pattern and there is a 1 in 4 chance of an embryo being affected with the condition where both parents are carriers.

4. The Committee noted that the primary symptom of this condition is deep scarring blisters which can be widespread across a person's skin. The blisters can be extremely painful, be slow to heal and become infected. Healing may lead to scarring causing contractures across joins. Infection, dehydration, respiratory problems, gastrointestinal and/or genitourinary tract involvement may lead to death in infants. Individuals also have an increased risk of developing skin cancer.

5. The Committee noted that other symptoms that individuals may develop are complete loss of scalp hair, areas of missing skin at birth, nail dystrophy and tooth pitting.

6. The Committee noted that the degree of penetrance is complete where a person has inherited two abnormal copies of the gene. The condition is normally present from or soon after birth.

7. The Committee noted that there is no curative treatment for non-Herlitz Junctional Epidermolysis Bullosa and the only treatment options available are those to manage the symptoms that arise from the condition.

8. The Committee noted that the application is consistent with the Peer Review.

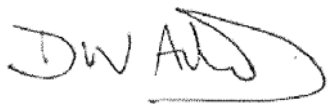
9. The Committee welcomed the advice of its Advisor, Dr Edward Blair, who confirmed that the condition was as described in the papers and added that it is a painfully disfiguring condition which causes mobility issues and can lead to recurring skin cancer.

10. The Committee considered that the condition is serious due to the complete penetrance, the deep scarring blisters causing chronic pain and mobility issues, and possibility of death during infancy.

11. 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) and (2) of Schedule 2 to the Act.

12. The Committee agreed to authorise the testing of embryos for non-Herlitz Junctional Epidermolysis Bullosa (OMIM #226650)

Signed: Date: 22/12/2014



David Archard (Chair)