

HFEA Statutory Approvals Committee

20 November 2014

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

Minutes – Item 2

Centre 0201 (Edinburgh Assisted Conception Unit) – PGD application for Nail-Patella Syndrome OMIM #161200

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Trent Fisher
Rebekah Dundas (lay)	
Sue Price (professional)	Legal Adviser:
Jane Dibblin (lay)	Ros Foster, Browne Jacobson
	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

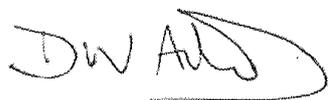
1. The Committee had regard to its Decision Tree. The Committee noted that the Centre is licensed to carry out PGD. The Committee was also satisfied that the Centre has experience of carrying out PGD and that generic patient information about its PGD programme and associated consent forms had previously 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 Nail-Patella Syndrome (OMIM #161200) is inherited in an autosomal dominant pattern and there is a 1 in 2 chance of an embryo being affected with the condition where one parent is affected.
4. The Committee noted that the symptoms of Nail-Patella Syndrome can vary from nail, knee and elbow abnormalities to scoliosis which can cause dislocation, instability and joint locking. Dental problems may include weak, crumbling teeth and thin enamel, and affected individuals are at increased risk of glaucoma, which can cause blindness.
5. The Committee noted that severe kidney problems develop in 30-50% of affected individuals causing proteinuria (proteins leaking into the urine), with or without haematuria (blood leaking into the urine), and may be static or progress to kidney failure. Approximately 5% of patients develop end-stage renal disease.
6. The Committee noted that Nail-Patella syndrome is extremely variable in both the severity and frequency of its manifestations, and it is impossible to predict how severely an affected embryo will become affected.
7. The Committee noted that there is no known cure for Nail-Patella Syndrome, however the symptoms can be treated with surgery for glaucoma and

orthopaedic problems as well as painkillers, physiotherapy and splinting. Transplants may be needed for severe kidney disease.

8. The Committee noted that the application is consistent with the Peer Review.
9. The Committee considered that the condition is serious because of the range of symptoms that may develop. In particular, the Committee noted that in the worst case scenarios severe impairment to mobility, pain in joints, glaucoma and end-stage renal disease are possibilities. The Committee also noted the extreme unpredictability of a patient affected by the condition developing symptoms.
10. 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.
11. The Committee agreed to authorise the testing of embryos for Nail-Patella Syndrome (OMIM #161200)

Signed:

Date: 3 December 2014

A handwritten signature in black ink, appearing to read 'DWA' followed by a stylized flourish.

David Archard (Chair)