

Human Fertilisation and Embryology Authority

Minutes of the Statutory Approvals Committee

Meeting held at Finsbury Tower, 103-105 Bunhill Row, London, EC1Y 8HF on
26 February 2015

Minutes – Item 2

Centre 0102 (Guys Hospital) – PGD application for ichthyosis with confetti OMIM #609165

Members of the Committee:	David Archard (Chair, lay) Jane Dibblin (lay) Sue Price (professional) Tony Rutherford (professional)
Legal Adviser:	Philip Grey, Mills and Reeve
Specialist Attending:	Dr Anne Lampe
Members of the Executive:	Sam Hartley – Head of Governance and Licensing Trent Fisher – Secretary

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:

- Executive Summary
- PGD application form
- Redacted peer review
- Genetic alliance opinion
- Further information provided by the centre

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)
- 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

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 condition being applied for is not on the approved PGD conditions list.
3. 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'.
4. The committee noted that ichthyosis with confetti OMIM #609165 is inherited in an autosomal dominant pattern and that there is a 1 in 2 chance of an embryo being affected with the condition where one parent is affected.
5. The committee noted that individuals affected by this condition suffer from erythroderma where skin is red, inflamed and scaly. The skin can be itchy and sore and is prone to colonisation or infection with bacteria. Eyelids can become thick and everted causing significant dryness, pain and risk of corneal damage. There is also an increased risk of skin cancer. Some infants with this condition present as a collodion baby, encased in tight shiny film. This is associated with a high risk of infection, dehydration and electrolyte imbalance and can cause breathing difficulties. Some babies do not survive the neonatal period.
6. The committee noted that other symptoms that may develop are thickness of skin, difficulties in perspiring causing trouble managing body temperature, clogged ears from skin debris and malodour caused by yeast and bacteria infections.

7. The committee noted that the condition demonstrates complete penetrance and that symptoms can be present from birth.
8. The committee noted that there is no curative treatment for the condition and the only treatment options available are those to manage the symptoms that arise from the condition.
9. The committee noted that the application is consistent with the Peer Review.
10. The committee welcomed the advice of its advisor, Dr Anne Lampe, who confirmed that the condition was as described in the papers and added that the tight skin can produce secondary contractures in joints causing limitations on movement which can affect mobility. She highlighted that most reported cases to date are de novo so there is no penetrance data but agreed with peer reviewer that penetrance is likely to be full.
11. The committee considered that the condition is serious due to the complete penetrance, the absence of curative treatment, the severity of symptoms and the effect they have on an individual's quality of life.
12. 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.
13. The committee agreed to authorise the testing of embryos for ichthyosis with confetti OMIM #609165

Signed:

Date: 12 February 2015

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

David Archard(Chair)