

Human Fertilisation and Embryology Authority

Minutes of the Statutory Approvals Committee

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

Minutes – item 2

Centre 0102 (Guys Hospital) – PGD application for blepharophimosis, ptosis, and epicanthus inversus 1 & 2 (BPES) OMIM #110100

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| Members of the Committee | David Archard (Chair, lay) Rebekah Dundas (Deputy Chair, lay) Sue Price (professional) Margaret Gilmore (lay) Bishop Lee Rayfield (lay) |
| Legal Adviser | Jane Williams, Mills & Reeve |
| Specialist Attending | Professor John Walter |
| Members of the Executive | 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

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 decision trees
- 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 condition 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 BPES 1 & 2 OMIM #110100 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.
5. The committee noted that the condition affects the development of the eyelids. The four main features of the condition are: fused eyelids or narrowing of the eye opening, droopy eyelids, an upward fold of the skin of the lower eyelid near the inner corner of the eye and an increased distance between the inner corners of the eye.
6. The committee noted that other symptoms that may develop are impaired vision, lazy eye, misalignment of the eye, short or long sightedness, dry eyes due to decreased tear production and, in women with BPES 1, early menopause due to premature ovarian failure.
7. The committee noted that the condition is virtually fully penetrant.
8. The committee discussed at some length the seriousness of the condition, in particular the variability of the symptoms, both functional and cosmetic, and the extent of treatment available. The committee noted that with early surgical

intervention the symptoms affecting an individual's eyelids can be alleviated to allow their vision to be maintained.

9. The committee, although sensitive to the fact that the condition is associated with altered facial appearance, did not regard this aspect of the condition as meeting the threshold for the statutory test of seriousness.
10. The committee noted the reference to the potential for early menopause and the treatment available for this and concluded that this aspect of the condition was not in itself of such importance to have any additional weight in the committee's deliberations.
11. The committee welcomed the advice from its legal advisor who reminded the committee that, when assessing a condition with variable symptoms, the committee will base its determination on how serious the disability or condition is on the worst case scenario.
12. The committee, in considering the worst case scenario of the symptoms associated with the condition and taking into account the treatment options available, concluded that the threshold for the statutory test of seriousness, which would allow the committee to licence this condition for PGD, was not met.
13. The committee accordingly refused to authorise the testing of embryos for blepharophimosis, ptosis and epicanthus inversus 1 & 2 OMIM #110100

Signed:

Date: 12 August 2015

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

David Archard(Chair)