

HFEA Licence Committee Meeting

27 January 2011

21 Bloomsbury Street London WC1B 3HF

Minutes – Item 2

Centre 0102 (Guy's Hospital) – PGD for Elastin (ELN) – Related Supravalvular Aortic Stenosis OMIM #185500

Members of the Committee:	Committee Secretary:
David Archard (lay) – Chair	Terence Dourado
Debbie Barber (Professional)	
Sally Cheshire (lay)	Legal Adviser:
Mair Crouch (lay)	Mary Timms
Jane Dibblin (lay)	
Rebekah Dundas (lay)	
Sue Price (Professional)	

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
- Joint opinion by Genetic Alliance and British Heart Foundation

The Committee also had before it:

- PGD Explanatory Note
- 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

1. The Committee had regard to its Decision Tree. It was aware that the Centre has considerable experience of carrying out PGD and it was satisfied that it has a large PGD programme.
2. The Committee was satisfied 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, ie. '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 Elastin (ELN) – related Supravalvular Aortic Stenosis OMIM #185500 is an autosomal dominant disorder. Only one copy of the affected gene is sufficient to cause the disorder, ie. there is a 50% chance of the embryo having the abnormality in a family where one parent is affected and the other is unaffected.
4. The Committee noted that there is a significant risk that a person with the abnormality will develop a serious medical condition because the condition can be as high as 100% penetrant.
5. Although the Committee recognised that the condition has variable symptoms, it based its determination of the seriousness of the condition on the worst possible symptoms (as stated in the PGD Explanatory note for Licence Committee). The Committee noted that at its worst the condition presents in the newborn period and requires immediate and invasive cardiac surgery and is potentially fatal. The Committee was satisfied that the condition is serious.
6. The Committee noted that there is a marked variability even within a family with the condition. It noted that centres will need to assess whether a particular request for PGD for the condition is appropriate on an individual family basis. It noted that the HFEA Code of Practice provides guidance on how such decisions should be made.
7. The Committee noted the British Heart Foundation and Genetic Alliance UK's joint opinion that states the impact upon individuals carrying an alteration in elastin varies widely; it can be a very mild condition with little or no impact upon patients, but can also lead to sudden infant death, heart attacks or strokes. The Committee was mindful of the significant possible impact upon a family of a child having the condition in the latter form.

8. 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 to the Act.

9. The Committee agreed that the licence should be varied to authorise the testing of embryos for Elastin (ELN) – related Supravalvular Aortic Stenosis - OMIM #185500, and that no conditions should be put on the licence in relation to the variation. The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed  Date 18 February 2011

David Archard (Chair)