

HFEA Licence Committee Meeting

25 November 2010

21 Bloomsbury Street London WC1B 3HF

Minutes – Item 6

Centre 0102 (Guys Hospital) – PGD for Methylmalonic Aciduria and Homocystinuria OMIM# 277400

Members of the Committee:	Committee Secretary:
David Archard (lay) – Chair	Terence Dourado
Debbie Barber (professional)	
Anna Carragher (lay)	Legal Adviser:
Sally Cheshire (lay)	Graham Miles, Morgan Cole
Mair Crouch (lay)	
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
- Genetic Alliance opinion

The Committee also had before it:

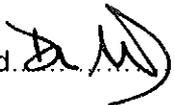
- HFEA Protocol for the Conduct of Licence Committee Meetings and Hearings
- 8th edition of the HFEA Code of Practice
- Human Fertilisation and Embryology Act 1990 (as amended)
- Decision trees for granting and renewing licences and considering requests to vary a licence (including the PGD decision tree); 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 and was satisfied that the Centre has an established PGD programme and has considerable experience of carrying out PGD.
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 Methylmalonic Aciduria and Homocystinuria is inherited in an autosomal recessive pattern. If an embryo inherits a copy of the faulty gene from both parents it will develop the disease, ie. there is a 25% chance of the embryo having the abnormality.
4. The Committee noted that there is a significant risk that a person with the abnormality will develop a serious medical condition because it is fully penetrant.
5. The Committee was satisfied that the condition is serious because although the age of onset may be variable, many individuals with the condition present at the severest end of the spectrum in infancy. Symptoms include failure to thrive, neurological problems (developmental delay/regression, seizures, hypotonia (abnormal loss of muscle tone), coma), and anaemia. Some develop multisystem pathology, including renal, liver and lung disease, cardiomyopathy (heart muscle disease), retinopathy and the haemolytic uremic syndrome. Later onset variants may present at anytime in childhood or adulthood with varying degrees of confusion, disorientation, psychiatric symptoms, developmental regression, gait abnormalities and incontinence.
6. The condition is partially treatable; primarily with intramuscular injections every 1-3 days for life. Most patients show some biochemical improvement, however, patients with the severest form of the condition may require additional treatment with oral betaine and/or dietary protein restriction, and may have a limited life span despite treatment. The degree of responsiveness to therapy depends upon the clinical severity of the condition and the degree of permanent brain damage prior to

commencement of treatment. Most patients survive long-term and neurological progression can be slowed or halted with treatment, although most have mild, moderate or severe physical and intellectual impairment despite optimal therapy in the longer term.

7. The Committee noted the variability of the phenotype. However it was minded to make its decision in respect of the most serious form of the condition.
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, Methylmalonic Aciduria and Homocystinuria OMIM# 277400 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... 16/12/2016

As approved by David Archard (Chair)