HFEA Statutory Approvals Committee

25 April 2013

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

Minutes – Item 1

Centre 0102 (Guys Hospital) – PGD Application Carnitine Acylcarnitine Translocase Deficiency (CACT) OMIM #212138

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford
Sue Price (professional)	Rebecca Loveys (observing)
Rebekah Dundas (lay) (Videoconference)	
Jane Dibblin (lay)	Legal Adviser:
	Juliet Oliver, Field Fisher Waterhouse

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

- Cover sheet
- Executive Summary
- PGD Application form
- Redacted Peer Review

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
- 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 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 Carnitine Acylcarnitine Translocase Deficiency (CACT, OMIM #212138) is inherited in an autosomal recessive pattern. There is therefore a 1 in 4 chance of the embryo being affected by this condition if both parents are carriers.

4. The Committee noted that CACT deficiency is a rare genetic condition which causes problems with the way that the body uses fat to produce energy. This is more of a problem during fasting (e.g. during illness) when our bodies switch from using carbohydrates to fats as the main energy source. Fatty acid metabolism is particularly important in heart and skeletal muscles (during exercise) and in other tissues during fasting. The gene involved in this disorder is called SLC25A20, and it is the instruction which tells our bodies how to make an enzyme called Carnitine Acylcarnitine Translocase. This enzyme is essential in the process where 'fatty acids' are metabolized and used for energy. Alterations or mutations in this gene mean that the enzyme is not produced correctly and so the fatty acids cannot be transported around the cell to locations where it is broken-down. This leads to a build up of the fatty acids and other chemicals in cells and also to low blood sugar. This imbalance in body chemistry means that babies usually develop symptoms soon after birth.

5. The Committee noted that symptoms often present at birth in the condition's severe form and include a combination of heart muscle weakness and rhythm abnormalities, breathing problems and liver failure which could all lead to sudden death or coma. A small number of patients have a milder form of the condition which develops in infancy but the symptoms are still severe and

include life-threatening encephalopathy (a brain disorder affecting consciousness) and hypoglycaemia.

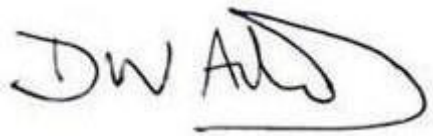
6. The Committee considered that the condition is serious as it presents soon after birth, and the symptoms are life threatening and in some cases fatal. There is no cure for Carnitine Acylcarnitine Translocase Deficiency in its severe form. The condition can be managed for children diagnosed in infancy with intravenous glucose and a restricted diet which is high in carbohydrate and low in fats. Infants receiving this supportive treatment can still be at risk of coma and sudden death due to the build-up of toxins and heart problems.

7. The Committee noted that the application is supported by the Peer Reviewer and the Genetic Alliance UK.

8. 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) of Schedule 2 to the Act.

9. The Committee agreed to authorise the testing of embryos for Carnitine Acylcarnitine Translocase Deficiency (OMIM #212138). The Committee confirmed that this condition will be added to the published list of condition for which PGD may be carried out.

Signed: Date: 09/05/2012



David Archard (Chair)