HFEA Licence Committee Meeting

25 February 2010

21 Bloomsbury Street London WC1B 3HF

Minutes – Item 4

Centre 0101 (CARE Nottingham) – Variation application to perform PGD for Niemann Pick disease Type A - OMIM# 257200

Members of the Committee: David Archard (lay) – Chair Anna Carragher (lay) Rebekah Dundas (lay) Sue Price (Professional) Apologies: Jane Dibblin (lay)	Committee Secretary: Terence Dourado Legal Advisers: Rosalind Bedward

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
- Application Form
- Anonymised peer review
- Email from JI, a Clinical Nurse Specialist in Niemann Pick Diseases

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. The Committee was satisfied that the Centre already has considerable experience of carrying out PGD and has conducted 37 PGD cycles between 1 January 2009 and an interim inspection at the Centre on 5 November 2009.

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 Niemann Pick disease Type A 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 baby with the abnormality will develop a serious medical condition because it is fully penetrant and one would expect each affected child to be affected to the same severe and fatal degree.

5. The Committee noted that the condition is serious because it is due to an inherited deficiency of an enzyme acid sphingomyelinase (ASM). It has been categorised as either neuronopathic (type A) (i.e. damaging the nervous system, with death in early childhood), or non-neuronopathic (type B). It is a very rare disease but mutations causing the type A form are more prevalent in the Ashkenazi Jewish population (1:80 and 1:100 people in that population carry one of three common mutations). The first symptom in type A is enlargement of the liver and spleen, usually noted by age three months. Over time the liver and spleen become massive; feeding problems, failure to thrive, gastrointestinal complaints (e.g. constipation, diarrhoea, and vomiting) occur. After a brief period of normal development, deterioration is relentless so that neurological development progresses no further than the twelve-month level for any domain, and skills are lost with disease progression. Developmental age usually does not progress beyond age nine months for gross motor skills; ten months

for fine motor skills, adaptive behavior; and twelve months for expressive language. Lung disease results in frequent respiratory infections and often respiratory failure. Most children die before age three. Whilst there is a report of successful bone marrow transplantation in most cases there is currently no intervention to alter the natural course of the disease.

6. 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.

7. The Committee agreed that the licence should be varied to authorise the testing of embryos for Niemann Pick disease Type A - OMIM# 257200, 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..5/3/2010..

David Archard (Chair)