HFEA Statutory Approvals Committee

27 June 2013

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

Minutes – Item 2

Centre 0102 (Guy's Hospital) – PGD application for Galactosialidosis (early infantile and adult/ juvenile types) OMIM #256540

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford
Sue Price (professional)	
Rebekah Dundas (lay) (Videoconference)	Legal Adviser:
Debbie Barber (lay)	Stephen Hocking, DAC Beachcroft

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
- 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
- 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 Galactosialidosis (early infantile and adult/ juvenile types) OMIM #256540 is inherited in an autosomal recessive pattern. There is therefore a 1 in 4 chance of the embryo being affected by this condition where both parents are carriers.

4. The Committee noted that Galactosialidosis can manifest in three different ways and that all are covered by OMIM #256540. The three forms are distinguished by the age at which symptoms develop and the pattern of features.

5. The Committee noted that the disorder is fully penetrant.

6. Babies with the early infantile form of the condition are born with a characteristic swelling and excess fluid between their organs or under their skin (hydrops fetalis and oedema), blood cells with characteristic empty spaces (vacuolated lymphocytes), an enlarged liver and spleen (hepatosplenomegaly). Babies fail to thrive, and do not develop physically or mentally. Damage occurs to many organs, and babies show symptoms of anaemia. Death usually occurs after a few months.

7. The Committee noted that children develop the late infantile form of the condition towards the end of their first year of life. The late infantile form of galactosialidosis is a milder form of the early infantile form. It is highly variable, and life expectancy varies accordingly. Children are also affected by short stature, eye problems, intellectual disability and heart valve problems.

8. The Committee noted that symptoms of the juvenile/adult form of the condition begin to develop around the age of 16. The primary symptoms are seizures, difficulty coordinating movements, and progressive mental impairment. Secondary symptoms are spinal abnormalities, eye problems and hearing loss. Life expectancy is normal.

9. The Committee noted that there is currently no cure to stop the progression of the symptoms of Galactosialidosis. Treatment is therefore directed at supporting specific problems as they arise.

10. The Committee considered that the condition is serious in all three formations because this is a serious metabolic genetic condition resulting in infantile death or significant life-long medical problems associated with severe learning disabilities.

11. The Committee noted that the application is supported by the Genetic Alliance UK.

12. 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.

13. The Committee agreed to authorise the testing of embryos for Galactosialidosis (early infantile and adult/ juvenile types) OMIM #256540. The Committee confirmed that this condition will be added to the published list of condition for which PGD may be carried out.

Signed: Date: 17/07/2013

David Archard (Chair)