HFEA Statutory Approvals Committee

25 July 2013

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

Minutes – Item 3

Centre 0119 (Birmingham Women's Hospital) – PGD application for Hereditary Multiple Exostoses Type II OMIM #133701

Members of the Committee:	Committee Secretary:
David Archard (lay) Chair	Lauren Crawford
Sue Price (professional)	
Rebekah Dundas (lay) (teleconference)	Legal Adviser:
Jane Dibblin (lay)	Tom Rider, Field Fisher
Hossam Abdalla (professional)	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

- Executive summary
- Application form
- Redacted peer review
- Genetic Alliance opinion
- Licence Committee minutes approving HME Type I - 22 June 2009

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 Hereditary Multiple Exostoses is caused by a mutation in the EXT1 and EXT2 genes. A mutation in the EXT1 gene is called Hereditary Multiple Exostoses Type I OMIM #133700 and a mutation in the EXT2 gene is called Hereditary Multiple Exostoses Type II OMIM #133701.

4. The Committee noted that Hereditary Multiple Exostoses Type II OMIM #133701 condition is inherited in an autosomal dominant pattern through an affected parent and there is a 50% chance of the embryo being affected by the genetic mutation.

5. The Committee noted that Hereditary Multiple Exostoses (HME) is a rare genetic condition in which people develop exostoses: multiple, benign tumours of the bone. It is a rare condition with a prevalence of 1 in 50,000 worldwide.

6. The Committee noted that large exostoses are often painful and/or uncomfortable. As exostoses grow, they may hinder growth; resulting in short stature as well as disruption in the development of arms, hands and legs. Since these problems may not affect the left and right sides equally, individuals can develop uneven limb length. Discrepancies in limb length, as well as abnormalities in joints, such as the ankles and hip joints can result in difficulties in walking and movement. The range of movement can be severely limited, resulting in extreme discomfort and frustration for the individual.

7. The Committee noted that there is no cure for Hereditary Multiple Exostoses. Severe bone deformities often need to be corrected surgically. This is usually required during childhood at specialist clinics. In the UK, only one such clinic exists. During childhood, in addition to surgeries, an annual review by an orthopaedic surgeon will be required and may continue into adulthood.

8. The Committee noted that PGD testing for Hereditary Multiple Exostoses Type I OMIM #133700 was previously approved by the HFEA on 22 June 2009.

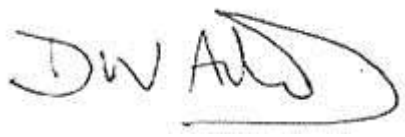
9. The Committee considered that the condition is serious. In its more severe form, the child or adult will have multiple exostoses requiring surgical removal and significant joint deformity. There is a low lifetime risk of malignant transformation of exostoses.

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

11. 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.

12. The Committee agreed to authorise the testing of embryos for Hereditary Multiple Exostoses Type II OMIM #133701. The Committee confirmed that this condition will be added to the published list of condition for which PGD may be carried out.

Signed: Date: 20/08/2013



David Archard (Chair)