Human Fertilisation and Embryology Authority

Minutes of the Statutory Approvals Committee

Meeting held at Finsbury Tower, 103-105 Bunhill Row, London, EC1Y 8HF on
30 July 2015

Minutes – item 1

Centre 0102 (Guys Hospital) – PGD application for muscular dystrophy–dystroglycanopathy (MDD) type A1-A8 and A10-A14 OMIM #236670 #613150 #253280 #253800 #613153 #613154 #614643 #614830 #615041 #615181 #615249 #615287 #615350

Members of the Committee	David Archard (Chair, lay) Rebekah Dundas (Deputy Chair, lay) Sue Price (professional) Margaret Gilmore (lay) Bishop Lee Rayfield (lay)
Legal Adviser	Jane Williams, Mills & Reeve
Specialist Attending	Professor John Walter
Members of the Executive	Trent Fisher, Secretary

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)
- HFEA decision trees

- 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

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 is applying in respect of a cluster of 13 subtypes of Muscular Dystrophy-dystroglycanopathy Type A (ie: A1 – A8; A10 – A14). Of the 13 subtypes in question, testing for sub-types A1, A3 and A5 has previously been authorised. These three sub-types are currently listed on the HFEA PGD condition list.

3. The committee noted that Muscular Dystrophy-dystroglycanopathy type A1 is listed on the HFEA PGD condition list as 'Walker Warburg Syndrome (Muscular dystrophy dystroglycanopathy)', type A3 is listed as 'Muscle-Eye-Brain Disease' and type A5 is listed as 'Muscular Dystrophy-dystroglycanopathy Type A5'.

4. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of Schedule 2 of the Act, i.e. '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'.

5. The committee noted that the condition is inherited in an autosomal recessive pattern and there is a 1 in 4 chance of an embryo being affected with the condition where both parents are carriers.

6. The committee noted that the condition affects the early development of brain, eyes and muscles. Individuals present with an abnormal brain structure causing intellectual disabilities and delayed or absent motor development. They may develop water on the brain and seizures. Abnormal eye structure usually leads to visual impairment.

7. The committee noted further that infants are born with weak muscle tone which deteriorates over time. Even in its least severe forms, any level of

mobility and speech is rare. While there is variability within the spectrum of Type A, presentation is always severe and associated with early death, often within the first year of life.

8. The committee noted that the condition demonstrates complete penetrance and symptoms will be present from birth and infancy.

9. The committee noted that there is no curative treatment for the condition and the only treatment options available are those to manage the symptoms that arise from the condition.

10. The committee noted that the application is consistent with the Peer Review.

11. The committee welcomed the advice of its Advisor, Professor John Walter, who confirmed that the condition is as described in the papers.

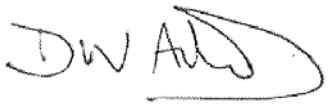
12. The committee considered the condition to be serious due to the early onset of symptoms, the number of extremely severe symptoms and the likelihood of early childhood death.

13. 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) and (2) of Schedule 2 to the Act.

14. The committee agreed to authorise the testing of embryos for Muscular Dystrophy-Dystroglycanopathy types A1, A2, A4, A6-A8 and A10-A14 OMIM #613150 #253800 #613154 #614643 #614830 #615041 #615181 #615249 #615287 #615350.

15. The committee agreed to the centre’s request to rename those conditions previously authorised under the names Walker Warburg Syndrome and muscle-Eye-Brain disease, as muscular dystrophy-dystroglycanopathy type A1 (Walker Warburg syndrome) OMIM #236670 and muscular dystrophy-dystroglycanopathy type A3 (muscle-Eye-Brain Disease) OMIM #253280. The committee considered the request carefully and concluded that it provided clarity to move away from eponymous terminology to a more uniform umbrella term that better describes the condition’s subtypes.

Signed: Date: 12 August 2015



David Archard(Chair)