Statutory Approvals Committee - minutes

Centre 0044 (Centre for Reproductive and Genetic Health) – PGD application for Beckwith-Wiedemann Syndrome OMIM #130650

Thursday, 25 February 2015

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

Committee members	David Archard (Chair) Margaret Gilmore Anthony Rutherford Anne Lampe	
Members of the Executive	Trent Fisher	Secretary
External adviser	Professor John Walter	
Legal Adviser	Dawn Brathwaite	Mills & Reeve
Observers	None	

Declarations of interest:

- members of the committee declared that they had no conflicts of interest in relation to this item.

The committee had before it:

- 8th edition of the HFEA Code of Practice
- standard licensing and approvals pack for committee members

The following papers were considered by the committee:

- executive summary
- PGD application form
- redacted peer review
- Genetic Alliance opinion

1. Consideration of application

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

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

1.3. The committee noted that Beckwith-Wiedemann Syndrome OMIM #130650 can be caused by a number of different defects, both genetic and epigenetic. As the application is for PGD, the committee agreed that it would consider the application for Beckwith-Wiedemann Syndrome caused by a mutation on the maternally inherited copy of the CDKN1C gene or inheritance of a chromosome translocation, deletion or duplication affecting the chromosome 11 Beckwith Wiedemann syndrome region.

1.4. 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.

1.5. The committee noted that the condition being applied for is not on the approved PGD condition list.

1.6. 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'.

1.7. The committee noted that the condition is inherited in an autosomal dominant manner which means there is a 1 in 2 chance in each pregnancy of an embryo being affected with the condition where the mother carries the mutation in the CDKN1C gene or where a parent carries a chromosome translocation, deletion or duplication affecting the chromosome 11 Beckwith Wiedemann syndrome region.

1.8. The committee noted that the condition is an overgrowth syndrome usually diagnosed soon after birth. A child with this disorder has an increased risk of developing tumours, particularly Wilms tumour, neuroblastoma and adrenocortical tumours. Children with this condition will undergo an intensive regimen of cancer screening throughout childhood. In the worst case scenario a child will die from a related malignancy.

1.9. The committee noted that symptoms can include abdominal wall defects, persistent low blood sugar, body asymmetry and large tongue causing feeding problems.

1.10. The committee noted that the condition has a high penetrance in families where it is due to a CDKNIC mutation or inherited chromosome 11 rearrangement. Onset of symptoms is from infancy.

1.11. The committee noted that there is no curative treatment for the condition, but there is treatment for symptoms that present.

2. Decision

2.1. The committee considered that the condition is serious due to the increased risk of childhood malignancy which can be fatal.

2.2. 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

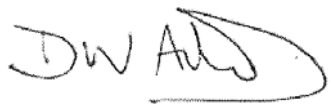
the condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 to the Act.

2.3. The committee agreed to authorise the testing of embryos for Beckwith-Wiedemann Syndrome caused by a mutation on the CDKN1C gene or inheritance of a chromosome translocation, deletion or duplication affecting the chromosome 11 Beckwith Wiedemann syndrome region.

3. Chair's signature

3.1. I confirm this is a true and accurate record of the meeting.

Signature



Name

David Archard

Date

10/03/2016