Statutory Approvals Committee - minutes

Centre 0102 (Guy's Hospital) – PGD application for Distal Renal Tubular Acidosis with progressive nerve deafness OMIM #602722 #267300

Thursday, 24 September 2015

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

Committee members	David Archard (Chair, lay) Rebekah Dundas (Deputy Chair, lay) Sue Price (professional) Margaret Gilmore (lay)	
Members of the Executive	Sam Hartley Trent Fisher	Head of Governance and Licensing Secretary
External adviser	Dr Anne Lampe	
Legal Adviser	Ros Foster	Browne Jacobson
Observers	Erin Barton	Projects, Inspections and Logistics Officer

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 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.

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

1.3. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of schedule 2 to the Human Fertilisation and Embryology Act 1990 (as amended) ("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'.

1.4. The committee noted that the condition applied for 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.

1.5. The committee noted that in its most severe form the condition is characterised by severe metabolic acidosis and progressive renal failure which both can be life limiting if unrecognized or poorly treated. This can cause hypertension, polyuria, polydipsia, osteopenia, osteomalcia and rickets, nephrolithiasis, and growth failure.

1.6. The committee noted that another aspect of this condition is progressive nerve deafness which is variable and can present at any stage.

1.7. The committee noted that the condition demonstrates complete penetrance and onset of symptoms can be from infancy.

1.8. The committee noted that there is no curative treatment for the condition, but that urgent and rapid correction of the underying metabolic abnormality may prevent some of the complications except deafness.

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

1.10. The committee welcomed the advice of its specialist advisor, Dr Anne Lampe, who confirmed that the condition is as described in the papers and that it is a severe and rare condition. Dr Lampe also pointed out that the description of the conditions contained at paragraph 4.3 of the Executive Summary was inaccurate insofar as it referred to 'asymptomatic patients' as the condition is fully penetrant and all those who inherit this condition will show some symptoms.

2. Decision

2.1. The committee considered that the condition is serious due to range of symptoms, lack of curative treatment and the early age of onset.

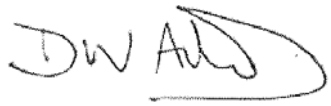
2.2. The committee had regard to its explanatory note and on the basis of the information presented, given the condition's worst symptoms, was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness 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 Distal Renal Tubular Acidosis with progressive nerve deafness OMIM #602722 #267300.

3. Chair's signature

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

Signature



Name

David Archard

Date

8 October 2015