Statutory Approvals Committee - minutes

Centre 0102 (Guy's Hospital) – PGD application for Bartter Syndrome types 1, 2, 3, 4a and 4b, OMIM #601678, #241200, #607364, #602522 and #613090

Thursday, 27 August 2015

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

Committee members	David Archard (Chair, lay) Rebekah Dundas (Deputy Chair, lay) Margaret Gilmore (lay) Sue Price (professional)	
Members of the Executive	Jo McAlpine	Secretary
External advisor	Professor Peter Turnpenny	
Legal Advisor	Tom Rider	Fieldfisher
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 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 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'.

1.4. 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.

1.5. The committee noted that the condition affects the kidneys, resulting in the inability for the kidneys to reabsorb salt normally, so salt is lost in the urine.

1.6. The committee noted that symptoms that may develop are raised blood pressure, palpitations and heart arrhythmias, hardening (calcification) of the kidneys, recurrent vomiting, dehydration, muscle cramps, numbness or tingling of the extremities, abdominal pain, diarrhoea, fatigue and depression. Loss of excess amounts of salt in the urine leads to constipation and increased urine production and the loss of excess calcium causes weakening of the bones. Type 4 causes hearing loss because of abnormalities in the inner ear.

1.7. The committee noted that the condition demonstrates complete penetrance with symptoms depending on the gene involved.

1.8. The committee noted that there is no curative treatment for the condition; however, there are treatment options available to treat some of the symptoms. The patient would need lifelong monitoring.

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, Professor Peter Turnpenny, who confirmed that the condition is as described in the papers.

2. Decision

2.1. The committee considered that the condition is serious in its worst case due to the possibility of major early complications in childhood, including failure to thrive, progressive renal failure, deafness and drug-induced side effects. Quality of life can be significantly impaired in the more severe presentations despite the use of pharmacotherapy. In undiagnosed/untreated cases it can still be fatal.

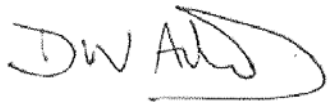
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious 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 Bartter Syndrome types 1, 2, 3, 4a and 4b, OMIM #601678, #241200, #607364, #602522 and #613090.

3. Chair's signature

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

Signature



Name

David Archard

Date

10 September 2015