Statutory Approvals Committee - minutes

Centre 0035 (Oxford Fertility Unit) – PGD application for Joubert Syndrome 6 OMIM #610688

Thursday, 26 November 2015

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

Committee members	David Archard (Chair) Rebekah Dundas (Deputy Chair) Sue Price Bishop Lee Rayfield	
Members of the Executive	Trent Fisher Sam Hartley	Secretary Head of Governance and Licensing
External adviser	Professor Mary Porteous	
Legal Adviser	Jane Williams	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 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 can present in utero with abnormalities of the brain and enlarged cystic kidneys. Other medical problems associated with Joubert Syndrome 6 are renal failure due to abnormal kidneys, liver disease due to fibrosis of the liver and development of visual impairment due to developmental anomalies of the eyes or retinal degeneration

1.6. The committee noted that other symptoms associated with the condition are development delays with a variable degree of intellectual disability; extra digits may be present on the outside of the hands and feet. At birth infants may have low muscle tone, impaired feeding and panting respiration. Prognosis is particularly poor if there if there is encephalocele of the brain and these infants do not normally survive.

1.7. The committee noted that other medical problems associated with Joubert Syndrome 6 are renal failure due to abnormal kidneys, liver disease due to fibrosis of the liver and development of visual impairment due to developmental anomalies of the eyes or retinal degeneration.

1.8. The committee noted that the condition demonstrates a high penetrance.

1.9. The committee noted that there is no curative treatment for the condition; treatment is purely symptomatic and supportive.

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

1.11. The committee welcomed the advice of its specialist advisor, Professor Mary Porteous, who confirmed that the condition is as described in the papers.

2. Decision

2.1. The committee considered that the condition is serious due to the severity of the symptoms and medical problems associated with the condition and the lack of any curative treatment.

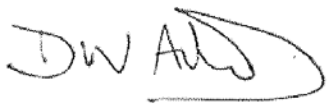
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious 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 Joubert Syndrome 6, OMIM #610688

3. Chair's signature

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

Signature



Name

David Archard

Date

10/12/2015