Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital) – PGD application for Joubert Syndrome 1-4, 7-9, 13-18 and 20-26 OMIM #213300, #608091, #608629, #609583, #611560, #612291, #612285, #614173, #614424, #614464, #614465, #614615, #614815, #614970, #615636, #615665, #616490, #616654, #616781 and #616784

Friday, 5 August 2016

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Rebekah Dundas (Chair) Margaret Gilmore Anthony Rutherford Anne Lampe	
Members of the Executive	Ian Brown Trent Fisher	Head of Corporate Governance Secretary
External adviser	Peter Turnpenny	
Legal Adviser	Sarah Ellson	Fieldfisher LLP
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
- three supporting research publications submitted with the application
- redacted peer review
- Genetic Alliance opinion

1. Consideration of application

1.1. The committee welcomed the advice of its specialist advisor, Professor Peter Turnpenny, who confirmed that the conditions are as described in the papers.

1.2. The committee noted that the application is consistent with the Peer Review and the Genetic Alliance opinion.

1.3. 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.

1.4. The committee noted that the conditions being applied for are not on the approved PGD condition list (although Joubert Syndrome types 5 and 6 are) and welcomed the centre's application to cover multiple appropriate forms of Joubert Syndrome.

1.5. 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'.

1.6. 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.

1.7. The committee noted that the condition affects the cilia which are hair-like structures found on cells in many different parts of the body. It is a multisystem disorder of the central nervous system. Symptoms can include abnormal development of the brain, cystic kidney and scarring of the kidneys which can lead to renal failure, liver abnormalities, poor muscle tone, a range of eye abnormalities including blindness, and breathing problems.

1.8. The committee noted further that some behavioural traits have also been described in children and adults with JS including; impulsive behaviour, temper problems and autistic features. There is a list of other rarer findings in JS that overlaps all clinical subtypes, including; endocrine abnormalities, obesity, laterality defects and other structural brain abnormalities.

1.9. The committee noted that the condition demonstrates complete penetrance although there may be some variability in expression. The committee noted that symptoms may be present at birth or develop during childhood.

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

2. Decision

2.1. The committee considered that the conditions are serious as each is a severe multisystem disorder which can have a devastating impact on an individual's life.

2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the conditions' 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 conditions meet 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 1-4, 7-9, 13-18 and 20-26 OMIM #213300, #608091, #608629, #609583, #611560, #612291, #612285, #614173, #614424, #614464, #614465, #614615, #614815, #614970, #615636, #615665, #616490, #616654, #616781 and #616784.

3. Chair's signature

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

Signature

Name

Rebekah Dundas

Date

19 August 2016