Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Combined Oxidative Phosphorylation Deficiency MRPS22, OMIM #611719

Thursday, 27 April 2017

Church House Westminster, Dean's Yard, Westminster SW1P 3NZ

Committee members	Margaret Gilmore (Chair) Anne Lampe Ruth Wilde Anthony Rutherford Bobbie Farsides	
Members of the Executive	Dee Knoyle Paula Robinson Bernice Ash	Secretary Head of Planning & Governance Committee Officer
External adviser	Professor Peter Turnpenny	
Legal Adviser	Graham Miles	Blake Morgan
Observers		

Declarations of interest

- Members of the panel 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 adviser, Professor Peter Turnpenny, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Combined Oxidative Phosphorylation Deficiency, MRPS22 is consistent with the Peer Review.

1.3. The committee noted that the Genetic Alliance opinion provided a patient's perspective and supported the application.

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 list of approved PGD conditions.

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 recessive pattern which means there is a 25% chance of having an affected child in each pregnancy, if each parent has a relevant mutation.

1.8. Combined Oxidative Phosphorylation Deficiency, MRPS22 is a rare disorder which is often fatal within the first few months of life, or results in very severe disability. There have only been a few cases reported but it appears to be fully penetrant.

1.9. Reported symptoms include muscle hypotonia, lactic acidosis, cardiomyopathy, tubulopathy and cerebral abnormalities including small cysts, micro-calcifications and absent or underdeveloped corpus callosum.

1.10. There is currently no curative treatment available for Combined Oxidative Phosphorylation Deficiency, MRPS22.

1.11. The committee noted the inspectorate’s recommendation to consider the approval of Combined Oxidative Phosphorylation Deficiency, MRPS22 to be included on the PGD List named as Combined Oxidative Phosphorylation Deficiency 5, OMIM #611719, which is in line with the information published on the OMIM website. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that this is a serious mitochondrial condition which is often fatal within the first few months of life, or results in severe disability. Babies affected by the condition are likely to have heart and kidney problems, low muscle tone in the limbs and trunk of the body making them floppy, with poor growth and very little development. Affected individuals may experience progressive metabolic brain disease (leukoencephalopathy), seizures and a severe build-up of acid in the body (metabolic acidosis) which can lead to coma and death.

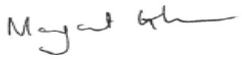
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition Combined Oxidative Phosphorylation Deficiency 5, (COXPD5) OMIM #611719 does meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

2.3. The committee agreed to authorise the testing of embryos for Combined Oxidative Phosphorylation Deficiency, MRPS22 and agreed that the condition should be included on the PGD List on the HFEA website named as Combined Oxidative Phosphorylation Deficiency 5, (COXPD5) OMIM #611719.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

15 May 2017