Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Mabry Syndrome (Hyperphosphatasia mental retardation syndrome-HPMRS), Type 1, OMIM #239300

Monday, 23 January 2017

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

Committee members	Margaret Gilmore (Chair) Anne Lampe Ruth Wilde Tony Rutherford	
Members of the Executive	Dee Knoyle Siobhain Kelly	Secretary Interim Head of Corporate Governance
External adviser	Dr Ed Blair	
Legal Adviser	Tom Rider	Field Fisher
Observers	Bobby Farsides Bernice Ash	Member (Induction) Committee Secretary

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
- Two additional papers submitted with the application
- Redacted peer review
- Genetic Alliance Opinion
- Minutes of the SAC on 28 August 2014 which approved hyperphosphasia with mental retardation syndrome 4 (HPMRS4) (OMIM #615716), as a condition for which PGD can be applied.

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr Ed Blair who confirmed that the condition is as described in the papers.

1.2. The committee noted that the description in the PGD application for Mabry Syndrome, Type 1 is consistent with the Peer Review and the Genetic Alliance opinion paper.

1.3. 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 condition being applied for is not on the list of approved PGD conditions.

1.5. Affected children present with intellectual disability, delay of physical development including major aspects such as walking, sitting, speaking and fine movements of hands and fingers and coordination. Children may also have epilepsy. Neurological symptoms range from severe hypotonia (floppiness) and delayed motor milestones in early life to epilepsy, which in some cases may not respond to treatment. In later childhood, a dystonic (stiffness) movement disorder may gradually set in which can worsen the neurodisability. The degree of developmental delay in most patients is severe. Some patients never develop expressive language.

1.6. The committee noted that there is no curative treatment for the condition.

1.7. The committee noted the inspectorate's recommendation to consider the approval of Mabry Syndrome, Type 1 to be included on the PGD List.

2. Decision

2.1. The committee had regard to its decision tree.

2.2. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph IZA(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'.

2.3. 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 parent are carriers of relevant mutations.

2.4. The committee noted that the condition demonstrates 100% penetrance and onset of symptoms is from one to two years of age.

2.5. The committee considered that the condition is serious due to learning disabilities, severe developmental delay, congenital malformations, poor speech development or no speech and epilepsy. The committee considered that affected children would have a limit to independent life.

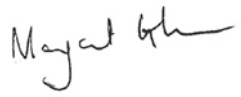
2.6. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition meets the criteria for testing under paragraph IZA(1)(b) and (2) of Schedule 2 of the Act.

2.7. The committee agreed to authorise the testing of embryos for Mabry Syndrome, Type 1 OMIM #239300.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

10 February 2017