Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Atrial Septal Defect (ASD) 7 with or without Conduction Defects, OMIM #108900.

Thursday, 25 May 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	Bernice Ash Paula Robinson	Secretary Head of Planning & Governance
External adviser	Dr Jenny Carmichael	
Legal Adviser	Sarah Ellson	Fieldfisher
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, Dr Jenny Carmichael, who confirmed that the condition was as described in the papers.

1.2. The committee noted the description in the application for Atrial Septal Defect (ASD) 7 with or without motor conduction defects, OMIM #108900 is consistent with the Peer Review.

1.3. The committee noted that the Genetic Alliance opinion provided a patient 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 dominant pattern which means there is a 50% chance of having an affected child in each pregnancy, if either parent has a relevant mutation.

1.8. The condition is due to mutations in the NKX2.5 gene (OMIM 600584) that result typically in babies being born with a "hole in the heart", usually an atrial septal defect (ASD), often combined with disturbances of the electrical conduction within the heart. Heart muscle disease (cardiomyopathy) and sometimes more complex congenital heart disease (CHD) and sudden cardiac death can occur. Penetrance is thought to be very high but probably not complete.

1.9. The committee noted that treatment does not modify the disease process per se but is aimed at preventing complications. Thus, this includes surgical repair of CHD if present and monitoring for heart block and rhythm disorders with the insertion of a pacemaker or an implantable defibrillator as necessary. The risk of sudden death remains, even after surgery.

1.10. The committee noted the inspectorate's recommendation to consider approval of Atrial Septal Defect (ASD) 7 with or without motor conduction defects, OMIM #108900 to be included in the PGD List, named as Atrial Septal Defect (ASD) 7 with or without motor conduction defects, OMIM #108900 which is in line with the information published by OMIM. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered Atrial Septal Defect (ASD) 7 with or without motor conduction defects, OMIM #108900 is serious given the early onset and the impact on quality of life of this condition, including heart murmurs, strokes, development of cardiomyothapy, risk of sudden death and the lack of a cure. The committee considered the impact on the individuals' quality of life and the family caring for affected individuals.

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

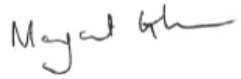
serious illness or any other serious medical condition. The committee was therefore satisfied that the condition of Atrial Septal Defect (ASD) 7 with or without motor conduction defects OMIM #108900 does meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

2.3. The committee agreed to authorise the testing for Atrial Septal Defect (ASD) 7 with or without motor conduction defects, OMIM #108900.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

8 June 2017