Statutory Approvals Committee – minutes

Centre 0037 (Glasgow Royal Infirmary)

Pre-implantation Genetic Diagnosis (PGD) application for ST3GAL3 Intellectual Disability, Autosomal Recessive 12, OMIM #611090

Thursday, 22 March 2018

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe Ruth Wilde Anthony Rutherford	
Members of the Executive	Bernice Ash Dee Knoyle Paula Robinson Catherine Burwood	Committee Secretary Committee Secretary (Observer) Head of Planning and Governance (Observer) Senior Governance Manager (Observer)
Specialist Adviser	Dr Jenny Carmichael	
Legal Adviser	Sarah Ellson	FieldFisher LLP
Observers		

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 UK Statement

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 that the description in the application for ST3GAL3 Mental Retardation, Autosomal Recessive 12, OMIM #611090 is consistent with the peer review. The committee noted the Executive's recommendation that the application is considered as Mental Retardation, Autosomal Recessive 12, OMIM #611090, to match the condition name used by the Peer Reviewer and the OMIM website. However, the committee decided that the condition should be named as Intellectual Disability Autosomal Recessive 12, OMIM #611090.

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 recessive pattern which means there is a 25% chance of an embryo being affected with the condition, if each parent has a relevant mutation.

1.8. The committee noted that Intellectual Disability, Autosomal Recessive 12 is a recently described genetically inherited condition resulting in moderate to severe intellectual disability present from birth. In severe cases, the person affected will have an IQ of as low as 25 or 30, resulting in limited language development, the inability to make autonomous decisions and live independently. Constant supervision will be required to assist in daily tasks, including dressing and eating.

1.9. The committee noted the condition is fully penetrant.

1.10. There is no curative treatment for the condition; appropriate supportive care and therapies are available. Affected children can attend schools for children with special educational needs and have very limited language development. Adults with the condition are unable to live independently. This condition will affect the quality of life of the person affected as well as the whole family.

1.11. The committee noted the inspectorate's request to consider whether Intellectual Disability, Autosomal Recessive 12, OMIM #611090, should be approved for inclusion on the PGD List. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that Intellectual Disability, Autosomal Recessive 12, OMIM #611090, is serious as it results in a severe intellectual disability, highly impacting on the ability to communicate and interact in all aspects of daily life, requiring constant supervision and resulting in the incapability to sustain any level of independence. The committee considered that this condition would have a severe impact on the life of the affected individual and their family.

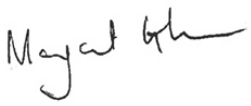
2.2. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, given the condition's worst symptoms, that a 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 Intellectual Disability, Autosomal Recessive 12, OMIM #611090, meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

2.3. The committee agreed to authorise testing for Intellectual Disability, Autosomal Recessive 12, OMIM #611090.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

9 April 2018