Statutory Approvals Committee – minutes

Centre 0102 (Guy's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Glycogen Storage Disorder Type 3 (GSD3), OMIM #232400

Thursday, 24 May 2018

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe	
Members of the Executive	Bernice Ash Dee Knoyle Paula Robinson Catherine Burwood Mhairi West	Committee Secretary Committee Secretary (Observer) Head of Planning and Governance (Observer) Senior Governance Manager (Observer) Inspector (Observer - Induction)
Specialist Adviser	Dr Ed Blair	
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
- Comment from centre regarding peer review
- Genetic Alliance UK Statement

1. Consideration of application

1.1. The committee welcomed the advice of its Specialist Adviser, Dr Ed Blair who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the application for Glycogen Storage Disorder Type 3 (GSD3), OMIM #232400 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 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 GSD3 is an inherited metabolic disorder caused by a build-up of a complex sugar in the body called glycogen. This impairs the function of some organs and tissues in the body, especially the liver and muscles. Symptoms are likely to develop in infancy or early childhood. The penetrance of GSD3 is very high (>90%) the phenotype is quite variable.

1.9. Symptoms of GSD3 are hypoglycaemia (low blood sugar), immunodeficiency, hypertriglyceridemia (high triglyceride blood levels), hepatomegaly (enlarged liver), hepatic failure and short stature. In a small percentage of people benign tumours may form in the liver. GSD3 is subdivided into 4 types which are mainly distinguished by the tissues which they affect. GSD types 3a and 3c typically affect both the liver and muscles, while types 3b and 3d typically affect only the liver. Individuals with type 3a may develop myopathy in both the heart and skeletal muscles later in life, (>50%).

1.10. The panel noted there is no cure for GSD3. In some cases, diet therapy is helpful. Liver transplantation may be indicated for patients with hepatic cancers. In infancy there is an increased risk of death due to seizures caused by low blood sugar. Many people with GSD3 live into adulthood. However, liver disease and muscle weakness may contribute to death.

1.11. The committee noted the inspectorate’s request to consider whether Glycogen Storage Disorder Type 3 (GSD3), OMIM #232400 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, in the worst case scenario, Glycogen Storage Disorder Type 3 (GSD3), OMIM #232400 is serious, with symptoms often developing in early infancy, causing an array of associated health problems, particularly affecting the muscles and liver. The committee noted this is a rare condition which can cause death in infancy. Treatment can adversely affect quality of life. The committee considered the impact on the individual's quality of life and the family.

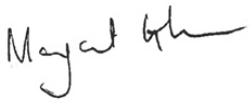
2.2. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, given the condition's worst symptoms, that a 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 Glycogen Storage Disorder Type 3 (GSD3), OMIM #232400 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 Glycogen Storage Disorder Type 3 (GSD3), OMIM #232400.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

19 June 2018