Statutory Approvals Committee – minutes

Centre 0035 (Oxford Fertility)

Pre-implantation Genetic Diagnosis (PGD) application for

Congenital Disorder of Glycosylation type 1D (CDG1D), OMIM #601110

Thursday, 26 October 2017

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

Committee members	Margaret Gilmore (Chair) Anne Lampe Anthony Rutherford Bobbie Farsides	
Members of the Executive	Dee Knoyle Bernice Ash Susanna Nyarko-Parkin	Committee Secretary Committee Secretary (Observing) Governance Officer (Observing)
External adviser	Mary Porteous	
Legal Adviser	Jane Williams	Mills & Reeve LLP
Observers	Gerard Hanratty	Browne Jacobson LLP

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 opinion

1. Consideration of application

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

1.2. The committee noted that the description in the application for Congenital Disorder of Glycosylation type 1D (CDG1D), OMIM #601110 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 onset of CDG1D is from early infancy. This is a multi-system disorder, often with significant involvement of the central nervous system leading to severe neurological disability. Symptoms of the condition include poor suckling and children can fail to thrive with death occurring during infancy.

1.9. There is a delay in neuromotor and physical development and episodes of lethargy and hypothermia. Limb-wasting due to truncal floppiness and intractable seizures (convulsions) are a prominent manifestation early in life. Affected individuals may have organ failure requiring urgent hospitalisation, including cardiac failure with pericardial effusion (fluid around the heart) and/or hepatic (liver) insufficiency with abnormal blood clotting and multiple hospital admissions due to intractable seizures The condition can be associated with anomalies of the skeleton, eye (may include cataract, corneal opacities, iris coloboma and optic atrophy) and brain structure. Affected individuals may have moderate to severe intellectual impairment and those who survive beyond childhood often have problems with speech and mobility.

1.10. The condition is fully penetrant and there is no curative treatment. The treatment of CDG1D is mainly palliative. Anticonvulsant medications are given for seizure control. Physiotherapy and rehabilitative mobility is used though progression through developmental milestones is usually absent or reverses with time.

1.11. The condition is progressive and individuals develop visual impairment, become wheelchair bound and dependent. The condition is life limiting and often leads to early death before teenage years.

1.12. The committee noted the inspectorate's request to consider whether Congenital Disorder of Glycosylation type 1D (CDG1D), OMIM #601110 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 Congenital Disorder of Glycosylation type 1D (CDG1D), is serious, given the significant risk that a child could have a multi-system disorder affecting the central nervous system, eventually causing severe neurological disability and limb-wasting, visual impairment, seizures and organ failure. The onset of the condition is as early as infancy. The condition is progressive and debilitating and individuals may have a severe intellectual disability. The committee found the personal statements submitted by Genetic Alliance compelling and considered that the level of medical intervention could have a severe impact on an individual's quality of life. The committee also considered the psychological impact of living with the uncertainty of the progression of the condition which is life limiting.

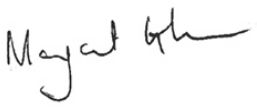
2.2. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, given the condition's worst symptoms, that a 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 Congenital Disorder of Glycosylation type 1D (CDG1D), OMIM #601110 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 Congenital Disorder of Glycosylation type 1D (CDG1D), OMIM #601110.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

16 November 2017