Statutory Approvals Committee – minutes

Centre 0006 (Lister Fertility Clinic)

Pre-implantation Genetic Diagnosis (PGD) application for

UBE2A - Intellectual Disability type Nascimento - X linked,

OMIM #300860

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	Dee Knoyle Bernice Ash Paula Robinson Catherine Burwood	Committee Secretary Committee Secretary (Observer) Head of Planning & Governance (Observer) Senior Governance Manager (Observer)
External 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 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 that the description in the application for UBE2A - Intellectual Disability type Nascimento - X linked, OMIM #300860 is consistent with the peer review.

1.3. The committee noted that Genetic Alliance 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 UBE2A - Intellectual Disability type Nascimento is an X-linked condition, which means there is a 50% chance of a child inheriting the affected X chromosome. 50% of males will be affected and 50% of females will be carriers. Males who inherit the mutation will be affected (100% penetrance). Female carriers are generally unaffected although some can show subtle facial features of the condition.

1.8. The committee noted that this is a rare condition. All affected males described to date have had moderate to severe intellectual disability with severe being far more common. Affected individuals have impaired or even absent speech and significant motor delay. Seizures are a common feature. Cardiac defects, particularly ventricular septal defects, are also common. Some affected individuals have recurrent infections. In addition, there are distinctive craniofacial dysmorphisms, hypospadias and undescended testes and underdevelopment of male genitalia has been described.

1.9. The committee noted that there is no curative treatment for this condition.

1.10. The committee noted the inspectorate’s request to consider whether UBE2A - Intellectual Disability type Nascimento - X linked, OMIM #300860 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 noted that this is a rare condition with very few cases reported in the literature. Given the limited data, but also what is known about the possibility of x-linked conditions potentially giving rise to serious disability, illness, or other medical conditions in worst case scenarios in females as well as in males, the committee decided that any decision to authorise testing should apply to testing embryos regardless of sex.

2.2. The committee considered that, in the worst case scenario, UBE2A - Intellectual Disability type Nascimento - X linked, OMIM #300860 is a serious condition. The age of onset is from birth and babies born with the condition may have cardiac defects, seizures, facial malformations and skin and nail abnormalities. Affected males may also have genital abnormalities. Children may have severe intellectual disability. This can severely affect the quality of life of affected individuals. Children may need special schooling and lifelong care and may never be able to live independently as adults.

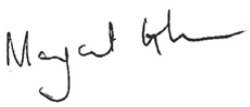
2.3. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, given the condition's worst symptoms, that a 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 UBE2A - Intellectual Disability type Nascimento - X linked, OMIM #300860 meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

2.4. The committee agreed to authorise testing for UBE2A - Intellectual Disability type Nascimento - X linked, OMIM #300860.

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