Statutory Approvals Committee – minutes

Centre 0119 (Birmingham Women's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Allan-Herndon-Dudley Syndrome

OMIM #300523

Thursday, 26 September 2019

HFEA, Spey Meeting Room, Level 2, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Anne Lampe Emma Cave Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr. Jenny Carmichael	
Legal Adviser	Tom Rider	FieldFisher LLP
Observer	Bernice Ash	Committee Officer

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:

- 9th 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 PGD application for Allan-Herndon-Dudley Syndrome, OMIM #300523 is consistent with the peer review.

1.3. The committee noted that the condition being applied for is not on the list of approved PGD conditions.

1.4. The committee noted that the Genetic Alliance UK statement provided a perspective on the impact of the condition on patients, their families and carers.

1.5. 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.

1.6. 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 Allan-Herndon-Dudley Syndrome, OMIM #300523 is inherited in an X-linked recessive manner, which means there is a 25% chance of having an affected male child and a 25% risk of a carrier female child in each pregnancy, if the mother has a relevant mutation.

1.8. The committee noted the penetrance of the condition is 100%.

1.9. Allan-Herndon-Dudley Syndrome, OMIM #300523 is characterised by physical and cognitive developmental delays leading to moderate to severe intellectual disability, difficulty with speech, joint contractures and muscle weakness which may result in severe pain, which cannot be communicated. Some children never gain the ability to hold their heads up or walk and most affected males will be wheelchair bound by the time they reach adulthood. Although rare, early deaths in childhood have been reported. As the condition is X-linked it will affect males, although girls, if carriers of the condition, can be affected with thyroid problems and intellectual disability.

1.10. There is no cure for this condition and limited options are available to manage symptoms.

1.11. The committee noted the executive's request to consider Allan-Herndon-Dudley Syndrome, OMIM #300523 for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that, in the worst-case scenario Allan-Herndon-Dudley Syndrome, OMIM #300523 is a devastating and intensely painful progressive condition resulting in profound physical and intellectual disability. Individuals are likely to have limited movement and develop little or no speech. The condition presents during infancy and there is no cure or treatment that can mitigate against this disease. Death may occur in early childhood.

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 abnormalities in question and that there is a significant risk, that a person with such an abnormality will, given the condition's worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

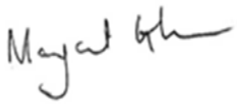
2.3. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Allan-Herndon-Dudley Syndrome, OMIM #300523

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
28 October 2019