Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Anderson-Tawil Syndrome

OMIM #170390

Thursday, 31 October 2019

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

Committee members	Margaret Gilmore (Chair) Rachel Cutting Emma Cave	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Professor Mary Porteous	
Legal Adviser	Ros Foster	Browne Jacobson LLP
Observer	Alistair Robertson	DAC Beachcroft LLP

Declarations of interest

- Members of the committee declared that they had no conflicts of interest in relation to this item.
- The Specialist Adviser explained she was also responsible for the peer review of the application. This was found not to pose a conflict.

Apologies:

- Apologies were received from Tony Rutherford and Bobbie Farsides.

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
- Licence Committee Minutes, 29 March 2012 - PGD for Long QT syndrome types 1, 2, 3, 5 & 6

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Professor Mary Porteous, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application Anderson-Tawil Syndrome, OMIM #170390, 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 Anderson-Tawil Syndrome is referred to as to Andersen Cardiodysrhythmic Periodic Paralysis, #170390 on the OMIM website. The committee therefore agreed at the request of the executive, to change the name of the condition for the purposes of this application to Andersen Cardiodysrhythmic Periodic Paralysis, #170390 to ensure consistency with the OMIM website.

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

1.6. 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.

1.7. 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'.

1.8. The committee noted that Andersen Cardiodysrhythmic Periodic Paralysis, #170390 is inherited in an autosomal dominant pattern, which means there is a 50% chance of an embryo being affected by the condition in each pregnancy if either parent has a relevant mutation.

1.9. The committee noted the penetrance of the condition is less than 100%. 80-94% of those affected may have a chance of developing symptoms, with approximately 75% exhibiting the three main features of heart rhythm abnormalities, muscle weakness, and skeletal abnormalities.

1.10. Andersen Cardiodysrhythmic Periodic Paralysis, #170390 is characterised by three main features; heart rhythm abnormalities which may lead to sudden death if left untreated, scoliosis, episodes of muscle weakness and paralysis, and physical abnormalities of the bones. Skeletal abnormalities are present from birth and cardiac arrythmia and muscle weakness present from young childhood.

1.11. There is no cure for this condition and treatment is directed to the management of specific symptoms which may include surgery, the possible need for implantable defibrillators and /or medication. The committee noted that there was the potential for heart and muscle treatments to counteract each other and that there was the potential for sudden death notwithstanding the treatment options available.

1.12. The committee noted the executive's request to consider Andersen Cardiodysrhythmic Periodic Paralysis, #170390 for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

1.13. The committee noted the executive's request for the peer reviewer to consider other types of long QT syndrome for approval alongside this application. The peer reviewer did not recommend that any other types of long QT syndrome should be considered within this application given the specific association of Andersen Cardiodysrhythmic Periodic Paralysis with episodic muscle paralysis, facial anomalies and in some cases mild learning difficulties. The committee therefore agreed that it would not consider other types of long QT syndrome on this occasion.

2. Decision

2.1. The committee considered that, in the worst-case scenario Andersen Cardiodysrhythmic Periodic Paralysis, #170390 is a progressive, debilitating and painful condition with symptoms present from birth and early childhood, and which in some cases may result in sudden unexpected death. There is no cure for the condition and the committee noted that some treatments may counteract each other due to the conflicting symptoms associated with the disease. The committee considered the possible adverse physical and psychological impact on the quality of life for patients living with this condition.

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 conditions' worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

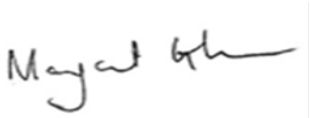
2.3. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Andersen Cardiodysrhythmic Periodic Paralysis, OMIM #170390

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
18 November 2019