# Statutory Approvals Committee – minutes

## Centre 0037 (Glasgow Royal Infirmary)

## Pre-implantation Genetic Diagnosis (PGD) application for

## Myoclonus Dystonia, OMIM #159900

Thursday, 27 April 2017

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

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| Committee members | Margaret Gilmore (Chair)<br>Anne Lampe<br>Ruth Wilde<br>Anthony Rutherford<br>Bobbie Farsides | |
| Members of the Executive | Dee Knoyle<br>Paula Robinson<br>Bernice Ash | Secretary<br>Head of Planning & Governance<br>Committee Officer |
| External adviser | Professor Peter Turnpenny | |
| Legal Adviser | Graham Miles | Blake Morgan |
| Observers | | |

## Declarations of interest

- Members of the panel 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, Professor Peter Turnpenny, who confirmed that the condition was as described in the papers.

**1.2.** The committee noted that the description in the PGD application for Myoclonus Dystonia, OMIM #159900 is consistent with the Peer Review.

**1.3.** The committee noted that the Genetic Alliance opinion provided a patient's 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 dominant pattern which means there is up to a 50% chance of having an affected child in each pregnancy, if either parent has a relevant mutation.

**1.8.** The committee noted that an individual who carries a mutation in the SGCE gene has a 50% risk of transmitting the mutation to their offspring. Importantly the gene is imprinted. If the transmitting parent is male the inheriting child is highly likely to develop the disorder. If the transmitting parent is female only about 5% of children inheriting the mutation go on to develop symptoms.

**1.9.** Myoclonus Dystonia is a rare neurological movement disorder in which incorrect signals between the brain and the muscles result in unwanted and uncontrollable muscle activity or spasms. These, sometimes painful, movements generally occur in the arms, trunk and neck, and force the body into twisting, repetitive movements or abnormal postures. Symptoms generally start at a young age, often within either the first or second decade of life.

**1.10.** Symptoms may be accompanied by dystonic tremor. The involuntary movements can be triggered or worsened by active movements of the affected parts of the body, and by stress or anxiety. Affected individuals may also have psychological symptoms such as obsessive-compulsive disorder (OCD), anxiety or depression.

**1.11.** There is currently no curative treatment available for Myoclonus Dystonia. Drugs may reduce symptoms in some patients and affected individuals may have regular botulinum toxin injections into affected muscles. Deep Brain Stimulation (DBS), which involves neurosurgery, may be used in severe cases but response is not consistent in all affected individuals.

**1.12.** Myoclonus Dystonia can seriously impact on quality of life. The dystonic element of the condition can spread to involve many segments of the body and the tremor can be disabling. Many affected people find going out in public uncomfortable resulting in social exclusion. The involuntary movements may improve when alcohol is taken but this does increase the risk of alcohol abuse and resulting long-term health effects. The psychological symptoms of this condition may have a significant impact on everyday life, affecting the ability to remain in employment and maintain relationships.

**1.13.** The committee noted the inspectorate's recommendation to consider the approval of Myoclonus Dystonia, OMIM #159900 to be included on the PGD List. The committee agreed to consider the application on this basis.

## 2. Decision

**2.1.** The committee considered whether Myoclonus Dystonia, OMIM #159900 is a serious condition. The committee noted the graphic description of first-hand experience of the condition and considered the worst case scenario. Individuals have problems with movement, uncontrollable muscle contractures or spasms in the upper body, forcing the body into twisting, repetitive movements or abnormal postures which can be painful.

**2.2.** The committee considered the individual's limit to independent life and employment opportunities. Affected individuals could be wheelchair bound and have their quality of life severely affected. Individuals may suffer prolonged pain and may have physical and mental health problems, including depression, anxiety and suicidal tendencies. Treatment to manage the condition is invasive and uncomfortable and there is no cure.

**2.3.** In making its decision the committee also took the advice of the specialist adviser, Professor Peter Turnpenny and noted the comments of the Peer Reviewer in relation to im-printing and the sex of the transmitting parent.

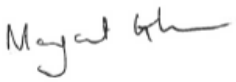
**2.4.** The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition Myoclonus Dystonia, OMIM #159900 does meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

**2.5.** The committee agreed to authorise the testing of embryos for Myoclonus Dystonia, OMIM #159900.

## 3. Chair's signature

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

### Signature



### Name

Margaret Gilmore

### Date

15 May 2017