# Statutory Approvals Committee – minutes

## Centre 0102 (Guys Hospital)

## Pre-implantation Genetic Diagnosis (PGD) application for Aarskog-Scott Syndrome, OMIM #305400

Thursday, 25 June 2020

HFEA, 10 Spring Gardens, London, SW1A 2BU via Teleconference

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| Committee members | Margaret Gilmore (Chair)<br>Emma Cave<br>Anne Lampe<br>Ruth Wilde | |
| Members of the Executive | Moya Berry<br>Catherine Burwood | Committee Officer<br>Licensing Manager |
| Specialist Adviser | Professor Mary Porteous | |
| Legal Adviser | Ros Foster | Browne Jacobson - LLP |

## Declarations of interest:

- Members of the committee declared that they had no conflicts of interest in relation to this item.

## Apologies:

- Apologies were received from Tony Rutherford.

## 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 from centre 0102, submitted 31 December 2019
- Supporting document 1- Letter from the Consultant Clinical Geneticist at centre 0102
- Supporting document 2- Letter from patients
- Supporting document 3- Supporting letter from the patients' Consultant Clinical Geneticist
- Supporting document 4- Supporting letter from the patients' health visitor
- Papers considered by SAC in November 2017 for the last application for Aarskog-Scott Syndrome by centre 0035 including
  - Executive Summary
  - Application from centre 0035
  - Redacted peer review

  - Genetic Alliance UK statement
- Minutes of previous considerations of Aarskog-Scott Syndrome by the Statutory Approvals Committee on:
  - 30 November 2017
  - 28 August 2014
  - 29 May 2014
- Licence Committee minutes (24 June 2010) of the approval of Pseudovaginal perineoscrotal hypospadias, OMIM #264600, due to 5 alpha reductase deficiency

## 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 for Aarskog-Scott syndrome due to a mutation in the FGD1 gene on chromosome Xp11, OMIM #305400, 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, produced for the November 2017 consideration of this condition, provided a perspective on the impact of the condition on patients, their families and carers.

**1.5.** The committee noted that an application to provide PGD for Aarskog-Scott Syndrome had been submitted by the centre on the 31 December 2019. The committee also noted that an application had also been considered previously in both 2014 and 2017 but had not been approved.

**1.6.** The committee noted the additional supporting information provided by the centre to support the application. This included opinions from two consultant clinical geneticists, a patient letter, and a letter from the health visitor.

**1.7.** The committee noted the request from the Aarskog Foundation to re-name the condition to Aarskog Syndrome, OMIM #305400 as this is the name most commonly used within the UK. However, the committee considered that it was not within its remit to change the condition's name when determining the application. Therefore, the condition being applied for will be known as Aarskog-Scott Syndrome, OMIM #305400.

**1.8.** 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.

**1.9.** 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'.

**1.10.** The committee noted that Aarskog-Scott Syndrome, OMIM #305400, is inherited in an autosomal recessive manner. Males have a 50% chance of inheriting the gene change and females equally have a 50% chance of inheriting the gene change.

**1.11.** The committee noted that the penetrance of the condition is not known.

**1.12.** Aarskog-Scott Syndrome, OMIM #305400, is characterised by skeletal malformations, development delay and may be associated with mild learning problems. Patients can also

exhibit umbilical hernia, shawl scrotum and undescended testes. The impact of the condition varies between those affected, even within the same family. While symptoms in some cases are mild, more severe cases can occur in which early surgical intervention is necessary, along with lifelong treatment from a multidisciplinary team which can significantly compromise quality of life.

**1.13.** There is no cure for this condition and treatment focuses on managing the symptoms of individual patients.

**1.14.** The committee noted the executive's request to consider Aarskog-Scott Syndrome, OMIM #305400, 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 agreed that, in the worst-case scenario Aarskog- Scott Syndrome, OMIM #305400, is a rare condition that presents from birth. Those affected with the condition may have a number of anomalies that require significant interventions and/or complex multiple corrective surgeries, which may result in a high degree of pain and suffering. The committee considered the serious physical, developmental and psychological implications, and impact on the quality of life for those worst affected with the condition.

**2.2.** The committee received advice from its specialist adviser and considered the new, up to date, additional information and supporting evidence provided by the centre. The committee noted letters from two consultant clinical geneticists in support of the application, including advice that while some patients have mild symptoms, there is a significant risk of a more severe phenotype.

**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 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.

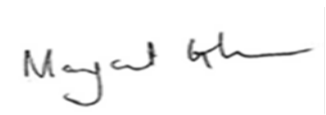
**2.4.** The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Aarskog-Scott Syndrome, OMIM #305400

## 3. Chairs signature

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

**Signature**



**Name**
Margaret Gilmore

**Date**
23 July 2020