Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Brooke Spiegler Syndrome (BSS), OMIM #605041

Thursday, 25 May 2017

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

Committee members	Margaret Gilmore (Chair) Anne Lampe Ruth Wilde Anthony Rutherford Bobbie Farsides	
Members of the Executive	Bernice Ash Paula Robinson	Secretary Head of Planning & Governance
External adviser	Dr Jenny Carmichael	
Legal Adviser	Sarah Ellson	Fieldfisher
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
- Content provided by Peer Reviewer
- 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 PGD application for Brooke-Spiegler Syndrome (BSS), OMIM #605041 is consistent with the Peer Review.

1.3. The committee noted that the Genetic Alliance opinion 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 the condition is inherited in an autosomal dominant manner which means there is 50% chance of having an affected child in each pregnancy, if either parent has a relevant mutation.

1.8. Brooke-Spiegler Syndrome (BSS) is an autosomal dominant disorder characterized by the appearance of multiple skin appendage tumours such as cylindroma, trichoepithelioma, and spiradenoma. These tumours are typically located in the head and neck region, appear in early adulthood, and gradually increase in size and number throughout life. There is an increased risk of skin and parotid gland tumours associated with the condition. BSS does not affect the intellect or generally shorten lifespan.

1.9. The penetrance is very high, close to 100%, but age dependent and variable. The condition in the severe forms can be very painful and disfiguring, and have a significant psychological impact. There is also an increased risk of skin and parotid tumour cancers in affected individuals.

1.10. The committee noted that treatment options for tumours include excision, dermabrasion, electrodessication, carbon dioxide (CO2) laser, cryotherapy, and radiotherapy. Despite treatment, the tumours are likely to reoccur.

1.11. The committee noted the inspectorate’s recommendation to consider the approval of Brooke-Spiegler Syndrome (BSS), OMIM #605041 to be included on the PGD List. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered Brooke-Spiegler Syndrome (BSS), OMIM #605041 is serious given the effects of this condition, including the painful, progressive development and appearance of the tumours, the risk of tumours becoming malignant and psychological impact. The committee considered the impact on the individuals' quality of life and the family caring for affected individuals.

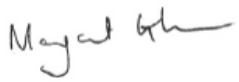
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition of Brooke-Spiegler Syndrome (BSS), OMIM #605041 does meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

2.3. The committee agreed to authorise the testing of embryos for Brooke-Spiegler Syndrome (BSS), OMIM #605041.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

8 June 2017