Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Schwannomatosis-type 1, OMIM #162091 and Schwannomatosis-type 2, OMIM #615670

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
Observers	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
- Licence Committee Minutes, 4 September 2006 – PGD for Neurofibromatosis type 2

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr Jenny Carmichael, who confirmed that the conditions were as described in the papers.

1.2. The committee noted that the description in the PGD application for Schwannomatosis type 1, OMIM #162091 and Schwannomatosis type 2, OMIM #615670. 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 Schwannomatosis type 1, OMIM #162091 and Schwannomatosis type 2, OMIM #615670 are inherited in an autosomal dominant pattern, which means there is a 50% chance of an embryo being affected by the conditions in each pregnancy if either parent has a relevant mutation.

1.8. The committee noted the penetrance of both the conditions is high but thought to be less than 100%.

1.9. The conditions are characterised by the development of multiple non-cancerous tumours (schwannomas) in different areas of the body but typically within the head, spine, arms and legs. Tumours can lead to significant pain which can be difficult to control, hearing loss, loss of balance, numbness, weakness, tingling, and headaches. In type 1 Schwannomatosis, meningiomas occur in approximately 5% of cases, which can cause changes in vision, hearing loss, headaches, memory loss, muscle weakness and seizures.

1.10. There is no cure for these conditions. Surgery can but does not always alleviate the symptoms and sometimes the tumours are not surgically operable due to their location and frequency. Tumours can be treated with radiotherapy and chemotherapy but this can increase the propensity for tumours to become malignant. Medication options available to control the severe pain are limited.

1.11. The committee noted the executive's request to consider Schwannomatosis type 1, OMIM #162091 and Schwannomatosis type 2, OMIM #615670 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 Schwannomatosis type 1, OMIM #162091 and Schwannomatosis type 2, OMIM #615670 are devastating, debilitating and severely painful conditions which can present in childhood. There is no cure for the conditions and treatment options to manage the pain are limited. The committee considered the possible adverse physical and psychological impact on the quality of life for patients living with the 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.

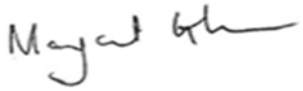
2.3. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Schwannomatosis type 1, OMIM #162091
- Schwannomatosis type 2, OMIM #615670

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