Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Ichthyosis, spastic quadriplegia, and mental retardation (ISQMR) syndrome, OMIM #614457

Thursday, 28 November 2019

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

Committee members	Margaret Gilmore (Chair) Anne Lampe Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr Alan Fryer	
Legal Adviser	Eve Piffaretti	Blake Morgan LLP

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

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr Alan Fryer, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Ichthyosis, spastic quadriplegia, and mental retardation (ISQMR) syndrome, OMIM #614457 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 Ichthyosis, spastic quadriplegia, and mental retardation (ISQMR) syndrome, OMIM #614457 is inherited in an autosomal recessive pattern, which means there is a 25% chance of an embryo being affected by the condition in each pregnancy if each parent has a relevant mutation.

1.8. The committee noted the penetrance of the condition is unknown but thought to be 100%.

1.9. Patients with the condition are likely to have significant developmental delay, spastic quadriplegia, and seizures within the first few months of life. Seizures are often resistant to treatment. Patients are unlikely to be able to live independently. Those people affected by this condition also have ichthyosis and skin infections associated with this are common and may cause severe pain. Death may occur in childhood.

1.10. There is no cure for this condition, so treatment focuses on managing the symptoms.

1.11. The committee noted the executive's request to consider Ichthyosis, spastic quadriplegia, and mental retardation (ISQMR) syndrome, OMIM #614457 for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

1.12. The committee also noted the peer reviewer's recommendation to consider Sjogren Larsson syndrome (SLS), OMIM #270200, alongside this application. SLS is a genetic condition which is phenotypically similar to ISQMR and is also characterised by ichthyosis, intellectual disability, spastic paresis and premature death.

2. Decision

2.1. The committee considered that, in the worst-case scenario Ichthyosis, spastic quadriplegia, and mental retardation (ISQMR) syndrome, OMIM #614457 is a rare, severe and devastating multi-system condition that presents at birth. There is no cure for the condition and those affected may die during childhood.

2.2. The committee considered in the worst-case scenario Sjogren Larsson syndrome (SLS), OMIM #270200 was of a similar clinical presentation and phenotype to ISQMR. The condition is also characterised by ichthyosis, intellectual disability, spastic quadriparesis and possible premature death.

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

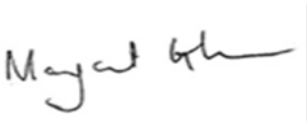
2.4. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Ichthyosis, spastic quadriplegia, and mental retardation (ISQMR) syndrome, OMIM #614457
- Sjogren Larsson syndrome (SLS), OMIM #270200

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
24 December 2019