

Statutory Approvals Committee – minutes

Centre 0035 (Oxford Fertility)

Pre-implantation Genetic Diagnosis (PGD) application for Isolated Sulfite Oxidase Deficiency (ISOD), OMIM #272300

Thursday, 26 November 2020

HFEA, 10 Spring Gardens, London, SW1A 2BU via Teams Meeting

Committee members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Professor Mary Porteous	
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 Statement
- 2018-06-28 SAC minutes, PGD for Molybdenum Cofactor Deficiency Type B (MOCODB), OMIM #252160

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 Isolated Sulfite Oxidase Deficiency (ISOD), OMIM #272300, 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 that Isolated Sulfite Oxidase Deficiency (ISOD), OMIM #272300, is inherited in an autosomal recessive manner, 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 that the penetrance of the condition is 100%.
 - 1.9.** Isolated Sulfite Oxidase Deficiency (ISOD), OMIM #272300, is characterised by brain dysfunction that gets progressively worse. Isolated Sulfite Oxidase Deficiency can appear within the first few days after birth and presents with seizures that are difficult to treat and feeding difficulties. Brain development is impaired, motor skills do not develop, and death usually follows within a few months. If Isolated Sulfite Oxidase Deficiency is diagnosed between 6-18 months, the condition can cause severe mental and physical disability.
 - 1.10.** There is no cure for this condition and treatment focuses on managing the symptoms.
 - 1.11.** The committee noted the executive's request to consider Isolated Sulfite Oxidase Deficiency (ISOD), OMIM #272300, for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.
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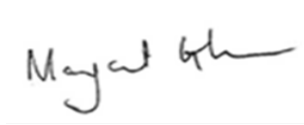
2. Decision

- 2.1.** The committee considered that, in the worst-case scenario, Isolated Sulfite Oxidase Deficiency (ISOD), OMIM #272300 is a severe, multi-system and fatal condition which presents in the first few days after birth. Babies diagnosed with the condition usually only live for a few months. Those children diagnosed between 6-18 months suffer severe physical and mental disability requiring long-term care and support. The committee considered the potentially devastating physical and psychological impact on the quality of life of those affected 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 abnormality 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.
- 2.3.** 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:
 - Isolated Sulfite Oxidase Deficiency (ISOD), OMIM #272300

3. Chairs signature

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

Signature

A handwritten signature in black ink, appearing to read "Margaret Gilmore", written on a white rectangular background.

Name

Margaret Gilmore

Date

23 December 2020