Statutory Approvals Committee - minutes

Centre 0037 (Glasgow Royal Infirmary)

Preimplantation Genetic Diagnosis (PGD) application for Spondyloepiphyseal dysplasia tarda (SEDT), OMIM #313400

Date:	27 May 2021
Venue:	Microsoft Teams Meeting
Committee Members:	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde
Specialist Adviser:	Alison Male
Legal Adviser:	Graham Miles – Blake Morgan LLP
Members of the Executive:	Moya Berry - Committee officer Catherine Burwood - Licensing Manager
Observers:	Dina Halai- Senior Scientific Policy Manager HFEA India Hickey – Research Officer (Induction)
Apologies:	No apologies were received for the meeting
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:

- HFEA Code of Practice 9th edition
- Standard Licensing and Approvals Pack

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 Alison Male, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Spondyloepiphyseal dysplasia tarda (SEDT), OMIM #313400, 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 PGD application for Spondyloepiphyseal dysplasia tarda (SEDT), OMIM #313400 is known as Spondyloepiphyseal dysplasia tarda, X-linked (SEDT), OMIM #313400 on the OMIM website and to ensure consistency the condition will be known as Spondyloepiphyseal dysplasia tarda, X-linked (SEDT), OMIM #313400.

1.5. The committee noted that the Genetic Alliance (UK) statement provided a perspective on the impact of the condition on patients, their families, and carers.

1.6. 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.

1.7. 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'.

1.8. The committee noted that Spondyloepiphyseal dysplasia tarda, X-linked (SEDT), OMIM #313400, is inherited in an X-linked recessive manner, which means that for female carriers there is a 25% chance of having an affected male and a 25% chance of having a carrier female in any pregnancy. For an affected male, all males will be unaffected, and all females will be carriers.

1.9. The committee noted that the penetrance of the condition is 100% in males. Female carriers tend to be asymptomatic, although the literature suggests that some female carriers may display mild osteoarthritis.

1.10. Spondyloepiphyseal dysplasia tarda, X-linked (SEDT), OMIM #313400, is characterised by moderate-to-severe spinal deformities (scoliosis and kyphosis), and premature osteoarthritis. Symptoms are seen from childhood onwards. Those affected experience progressive joint and back pain with osteoarthritis commonly seen in hip, knee, and shoulder joints in early adulthood.

1.11. There is no cure for this condition. Osteoarthritis and spinal problems can require long term analgesia for pain management and surgical intervention, e.g. hip replacement, and spinal surgery.

1.12. The committee noted the executive's request to consider Spondyloepiphyseal dysplasia tarda, X-linked (SEDT), OMIM #313400, for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

1.13. The committee noted the executive's request to consider the risks and severity of inheritance with regard to the symptoms in female carriers of the condition. The committee, when considering this application, agreed that any testing should only be carried out for the

‘condition’, under the S2 1ZA (1) (b) (abnormality) of Schedule 2 of the Act only and not for the purposes of sex testing S2 1ZA (1) (b) of Schedule 2 of the Act.

2. Decision

2.1. The committee considered that, in the worst-case scenario, Spondyloepiphyseal dysplasia tarda (SEDT), OMIM #313400, is a severely painful, and debilitating condition that presents in infancy. There is no cure for the condition and many affected individuals require joint replacement surgeries from an early age, which can result in a high degree of pain and the requirement for long-term chronic pain management. The committee considered the potential serious implications for, and the physical and psychological impact on the quality of life of, those affected by 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.

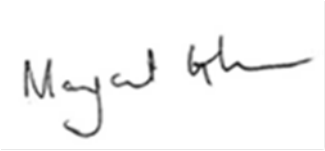
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Spondyloepiphyseal dysplasia tarda, X-linked (SEDT), OMIM #313400

3. Chair’s signature

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

Signature



Name

Margaret Gilmore

Date

21 June 2021