Statutory Approvals Committee – minutes

Centre 0044 (The Centre for Reproductive and Genetic Health)

Pre-implantation Genetic Diagnosis (PGD) application for Jalili Syndrome, OMIM #217080

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
- 2019-03-28 Statutory Approvals Committee Minutes, Cone Rod Dystrophy, OMIM #601777
- 2015-05-28 Statutory Approvals Committee Minutes, Leber Congenital Amaurosis types 3 -17
- 2012-11-01 Licence Committee Minutes, Leber Congenital Amaurosis, OMIM #204000/204100

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 Jalili syndrome, OMIM #217080 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 Jalili syndrome, OMIM #217080, 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. Jalili syndrome, OMIM #217080, is characterised by progressive visual loss as well as severe dental abnormalities. Visual impairment usually manifests in early infancy or childhood with nystagmus (jerky eye movements), photophobia (discomfort due to bright light), reduced visual acuity and colour vision defects. Nyctalopia (poor night vision) and visual field defects may develop later which may indicate involvement of the other type of light receptor cells, the rods. The dental abnormalities result in discolouration, sensitivity and brittleness of teeth, with premature loss of both primary and secondary dentition.

1.10. There are no treatments available to cure this disease, but some supportive therapies are available.

1.11. The committee noted the executive's request to consider Jalili syndrome, OMIM #217080, 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, Jalili syndrome, OMIM #217080, is a rare condition, which is progressive and incurable and can lead to severe visual impairment with some of those affected being registered as legally blind during childhood or in early adult life. The condition can also result in severe dental abnormalities requiring painful interventions and frequent hospital visits. The condition can significantly impact on the quality of life for patients and their families and may also have a serious psychological effect as a result of its progressive nature. 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.

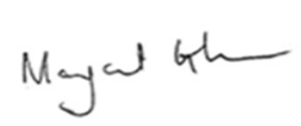
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Jalili syndrome, OMIM #217080,

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
23 December 2020