Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital)

Preimplantation Genetic Diagnosis (PGD) application for Otodental Dysplasia, OMIM #166750

Date:	28 January 2021
Venue:	Microsoft Teams Meeting
Committee Members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde
Specialist Adviser	Peter Turnpenny
Legal Adviser	Gerard Hanratty- Browne Jacobson LLP
Members of the Executive	Moya Berry - Committee officer Catherine Burwood – Licensing Manager
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:

- 9th edition
- Standard Licencing and Approvals Pack

The following papers were considered by the committee:

- Executive Summary
- PGD Application form
- Redacted Peer review
- Supporting publication: Gregory-Evans 2007
- SAC minutes, 28/09/2017, PGD for Congenital Deafness with inner ear agenesis, microtia and microdontia, OMIM #610706.

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Professor Peter Turnpenny, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Otodental Dysplasia, OMIM #166750 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 condition Otodental Dysplasia, OMIM #166750, is also known as Oculootodental syndrome on the OMIM website. However, as the OMIM website entry lists Otodental Dysplasia as the primary name of the condition, the condition, for the purposes of this application, will be known as Otodental Dysplasia, OMIM #166750.

1.5. The committee noted that a Genetic Alliance (UK) statement had not been provided for this application.

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 Otodental Dysplasia, OMIM #166750 is inherited in an autosomal dominant manner, which means there is a 50% chance of an embryo being affected by the condition in each pregnancy if either parent has a relevant mutation.

1.9. The committee noted that the evidence for penetrance of the condition is limited but appears to be close to 100% penetrance for dental and oral anomalies. Hearing loss is also highly penetrant but presents later in life.

1.10. Otodental Dysplasia, OMIM #166750 consists of significant craniofacial abnormalities which may adversely affect quality of life due to the need for extensive medical and surgical intervention in many cases. It is characterised by dental and oral abnormalities and hearing loss with a variable age of onset.

1.11. There is no cure for the condition and those affected may require extensive medical and surgical intervention to help ameliorate the symptoms.

1.12. The committee noted the executive's request to consider Otodental Dysplasia, OMIM #166750, 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, Otodental Dysplasia, OMIM #166750, is a rare condition with significant craniofacial abnormalities, which may present from birth or early childhood. There is no cure for the condition and affected individuals can suffer with serious dental abnormalities requiring multiple surgeries, and potentially severe, bilateral

hearing loss by early adulthood. The committee considered the extensive medical and surgical interventions required for those living with the condition and the potentially serious psychological and physical impact this may have on their quality of life.

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.

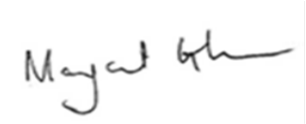
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Otodental Dysplasia, OMIM #166750

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

23 February 20121