Statutory Approvals Committee - minutes

Centre 0044 (The Centre for Reproductive and Genetic Health)

Preimplantation Genetic Diagnosis (PGD) application for Achromatopsia type 2, OMIM #216900

Date:	29 April 2021
Venue:	Microsoft Teams Meeting
Committee Members:	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde Jason Kasraie
Specialist Adviser:	Jenny Carmichael
Legal Adviser:	Darryn Hale – DAC Beachcroft LLP
Members of the Executive:	Moya Berry - Committee officer Catherine Burwood - Licensing Manager
Observers:	Julia Chain - HFEA Chair (Induction) Tim Child - HFEA Authority Member (Induction) Neil Ward - Mills & Reeve LLP (New Legal Adviser) (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 Jenny Carmichael, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Achromatopsia 2, (ACHM2), OMIM #216900, 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 Achromatopsia 2, (ACHM2), OMIM #216900, 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. Achromatopsia 2, (ACHM2), OMIM #216900, is a congenital disorder, characterised by poor visual acuity, nystagmus, loss of central vision, reduced or complete loss of colour discrimination, and extreme painful sensitivity to light (day blindness). Many of those affected will be registered as partially sighted or blind during childhood.

1.10. There is no cure for this condition. Treatments include the use of dark or special filter glasses or contact lenses to filter out the light that is uncomfortable, although over time such lenses may compromise vision further. Low vision aids can also be used along with occupational therapy adjustments such as preferential classroom seating for children.

1.11. The committee noted the executive's request to consider Achromatopsia 2, (ACHM2), OMIM #216900, for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

1.12. The committee noted the recommendation of the peer reviewer to consider a number of additional conditions for inclusion on the list for which PGD can be applied and agreed to consider the application on this basis. The conditions are Achromatopsia 3 (ACHM3), OMIM

#262300, Achromatopsia 4 (ACHM4), OMIM #613856, Achromatopsia 5 (ACHM5), OMIM #613093, Achromatopsia 6 (ACHM6), OMIM #610024 and Achromatopsia 7 (ACHM7), OMIM #616517.

1.13. Achromatopsia 3 (ACHM3), OMIM #262300, Achromatopsia 4 (ACHM4), OMIM #613856, Achromatopsia 5 (ACHM5), OMIM #613093 and Achromatopsia 7 (ACHM7), OMIM #616517, are inherited in an autosomal recessive manner, meaning the risk of inheriting the condition is 25% in each pregnancy if each parent carries a relevant mutation. The penetrance of the condition types is 100%. The condition types are considered by the peer reviewer to be equivalent to Achromatopsia 2 (ACHM2), OMIM #216900 in severity of symptoms, being characterised by poor visual acuity, nystagmus, loss of central vision, reduced or complete loss of colour discrimination, and extreme painful sensitivity to light.

1.14. The committee noted that Achromatopsia 6 (ACHM6), OMIM #610024, has been reported with both autosomal recessive and autosomal dominant inheritance. Based on the advice of its specialist adviser, the committee agreed that for the purpose of this application, only the recessive form of Achromatopsia 6 (ACHM6), OMIM #610024, should be considered for inclusion on the list for which PGD can be applied.

2. Decision

2.1. The committee considered that, in the worst-case scenario, Achromatopsia 2, (ACHM2), OMIM #216900, is a serious, incurable, and painful congenital condition that can severely affect visual function leading to some of those affected being registered as legally blind during childhood. The committee considered the serious implications for, and the physical and psychological impact on the quality of life of, those affected by the condition and the severe pain the condition may cause.

2.2. The committee also considered that the conditions Achromatopsia 3 (ACHM3), OMIM #262300, Achromatopsia 4 (ACHM4), OMIM #613856, Achromatopsia 5 (ACHM5), OMIM #613093, Achromatopsia 6 (ACHM6), OMIM #610024 and Achromatopsia 7 (ACHM7), OMIM #616517 are, in the worst-case scenario, serious painful congenital conditions resulting in many of those affected being registered as legally blind from childhood.

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 abnormality 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.

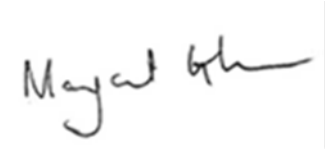
2.4. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Achromatopsia 2 (ACHM2), OMIM #216900
- Achromatopsia 3 (ACHM3), OMIM #262300
- Achromatopsia 4 (ACHM4), OMIM #613856
- Achromatopsia 5 (ACHM5), OMIM #613093
- Achromatopsia 6 (ACHM6), OMIM #610024 (autosomal recessive only)
- Achromatopsia 7 (ACHM7), OMIM #616517

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

1 June 2021