Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Doyne honeycomb retinal dystrophy (DHRD), OMIM #126600

Thursday, 17 December 2020

HFEA, 2nd Floor, 2 Redman Place, London E20 1JQ 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	Dr. Alison Male	
Legal Adviser	Tom Rider	Fieldfisher - 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
- 2018-01-25 Statutory Approvals Committee minutes, PGD application for Best Macular dystrophy (Best vitelliform macular dystrophy), OMIM #153700
- 2011-02-24 Licence Committee Minutes, PGD application for Macular Dystrophy Retinal 2, OMIM #608051
- Supporting Document- Symptomatic abnormalities of dark adaptation with EFEMP1 retinal dystrophy (Malattia Leventinese/ Doyne honeycomb retinal dystrophy), R Haimovici et al,

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 Doyne honeycomb retinal dystrophy (DHRD), OMIM #126600, 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 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 Doyne honeycomb retinal dystrophy (DHRD), OMIM #126600, 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.

1.8. The committee noted the penetrance of the condition is close to 100%.

1.9. Doyne honeycomb retinal dystrophy (DHRD), OMIM #126600, is a rare form of retinal dystrophy that usually causes symptoms from early adult life but can present in late teenage years. Patients may notice problems seeing detail, distortion (for example, straight lines looking wobbly), gaps in the centre of their vision, and difficulty adapting to changes in light level. Once the signs are detectable at the back of the eye, patients gradually lose their central vision. The progression of symptoms is usually relatively slow (becoming significant by the 40s-50s) but in some patients a more rapid visual loss occurs.

1.10. There is no cure for the condition and the focus is on symptom management and providing training and advice to individuals who experience sight loss so that they can learn and develop skills to enable them to get around safely and continue to do everyday activities.

1.11. The committee noted the executive's request to consider Doyne honeycomb retinal dystrophy (DHRD), OMIM #126600, 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, Doyne honeycomb retinal dystrophy (DHRD), OMIM #126600, is a rare and progressive condition that can present from the teenage years and may eventually lead to severe visual impairment, with some of those affected being registered as legally blind in early adult life. The progression of symptoms may affect the quality of life and independent living. There is no cure for the condition and the committee considered the potential physical and serious psychological impact on the quality

of life of those affected with the condition and the associated uncertainty of its progressive nature.

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.

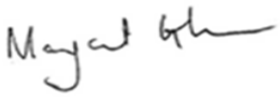
2.4. The committee agreed to authorise testing for:

- Doyne honeycomb retinal dystrophy (DHRD), OMIM #126600

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
14 January 2021