Statutory Approvals Committee – minutes

Centre 0044 The Centre for Reproductive and Genetic Health

Application to perform PGD for Stargardt disease, type 1, OMIM #248200

Thursday, 26 July 2018

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe Ruth Wilde	
Members of the Executive	Richard Chamberlain Dee Knoyle Bernice Ash Paula Robinson Catherine Burwood	Temporary Committee Clerk Committee Secretary (Observer) Committee Secretary (Observer) Head of Planning and Governance (Observer) Senior Governance Manager (Observer)
External Adviser	Professor Peter Turnpenny	
Legal Adviser	Graham Miles	Blake Morgan LLP
Apologies	Anthony Rutherford	

Declarations of interest

- The Specialist Adviser explained he was also responsible for the peer review of the application. It was found not to invalidate either contribution. Other members of the committee declared that they had no conflicts of interest in relation to this item.

The committee had before it:

- 8th 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 UK statement
- 2018-01-25 SAC minutes, PGD consideration, Best Vitelliform Macular Dystrophy
- 2011-02-24 LC minutes, PGD consideration, Macular Dystrophy Retinal 2
- 2009-02-23 LC minutes, PGD consideration, X-linked childhood-onset Macular Dystrophy

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 application to perform PGD for Stargardt disease, Type 1, OMIM #248200 is consistent with the peer review.

1.3. The committee noted that Centre 0044 - The Centre for Reproductive and Genetic Health Centre, is licensed to provide embryo testing.

1.4. The application was supported by Genetic Alliance UK. They consider the use of PGD to avoid the birth of a child affected by Stargardt disease, Type 1 to be appropriate.

1.5. The committee noted that the condition is inherited in an autosomal recessive pattern which means there is a 25% chance of an embryo being affected with the condition in each pregnancy, if both parents have a relevant mutation

1.6. Stargardt disease, type 1, is the most common form of inherited juvenile macular degeneration. The condition causes the death of photoreceptor cells in the central portion of the retina (the macula) and atrophy of the underlying retinal pigment epithelium, resulting in progressive central vision loss. People with Stargardt disease, type 1, may have impaired colour vision. The signs and symptoms of Stargardt disease, type 1, typically appear in late childhood to early adulthood and worsen over time. Onset is typically between the ages of 7 and 12 years of age. Central vision loss is the first symptom of the condition expressed in most of those affected and characteristic yellowish flecks (lipofuscin) appear under the macula. Impairment of colour vision develops.

1.7. The condition is progressive, so vision deteriorates with time. Eventually, most individuals with Stargardt disease, type 1, have a visual acuity in the range of 20/200 to 20/400. The condition has 100% penetrance

1.8. There is no curative treatment available and central vision loss limits the patient's ability to perform many visual tasks including reading. The vision loss is not correctable with prescription eyeglasses.

1.9. The purpose of testing by PGD is to establish whether the embryo has an abnormality causing Stargardt disease, type 1.

1.10. The Executive considers that the centre has provided all information on the relevant form to the HFEA as required by General Direction 0008.

1.11. The committee noted that the peer reviewer supported the application and pointed out the condition had an adverse effect on a young person's education, career options and lifestyle.

1.12. The peer reviewer, after consideration, felt it was not appropriate to include Stargardt disease types 3 and 4 (OMIMs #600110 and #603786) because they are linked to different genes and the pattern of inheritance is different.

1.13. The committee noted that Stargardt disease affected central vision and that it had a severe impact on education and career opportunities and if onset was late presenting in the patient's 20s, it could effectively terminate a career but has no direct effect on cognitive functions.

1.14. The committee noted that the disease could appear to level off in its progression but could progress later and was 100% penetrant.

1.15. 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.

2. Decision

2.1. The committee considered that given the onset of this disease is usually in childhood between seven and twelve years of age it has a debilitating effect on a child's and young adult's education and lifestyle and can have psychological effects. The disease is 100% penetrant leading in the worst-case scenario to legal blindness, just sufficient to move around a room. The application was fully supported by Genetic Alliance UK.

2.2. The committee considered the executive's request to consider other types of Stargardt disease, notably types 3 and 4, alongside this application, but noted that this had not been requested by the centre's PR and had not been recommended by the peer reviewer who had explained that differing genes were involved, and the pattern of inheritance was different (autosomal dominant).

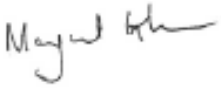
2.3. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, given the condition's worst symptoms, that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition as its penetrance was 100%. The committee was therefore satisfied that the condition Stargardt disease, type 1, OMIM #248200, 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 the Stargardt disease, type 1, OMIM #248200.

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

30 August 2018