Statutory Approvals Committee – minutes

Centre 0044 (The Centre for Reproductive and Genetic Health)

Pre-implantation Genetic Diagnosis (PGD) application for Nephropathy due to CFHR5 Deficiency, OMIM #614809

Thursday, 25 July 2019

HFEA Foyle Meeting Room, Level 1, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Emma Cave Tony Rutherford	
Members of the Executive	Moya Berry Catherine Burwood	Committee Secretary Licensing Manager (Observer)
Specialist Adviser	Peter Turnpenny	
Legal Adviser	Jane Williams	Mills & Reeve LLP

Declarations of interest

- Members of the committee declared that they had no conflicts of interest in relation to this item.
- Apologies were noted from Rachel Cutting (Committee Member)

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 UK statement

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 Nephropathy due to CFHR5 deficiency, OMIM #614809 was consistent with the peer review. The committee also noted that the condition appears on the OMIM website as CFHR5 deficiency, OMIM #614809 and agreed that going forward the condition would be known as this.

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 CFHR5 deficiency is inherited in an autosomal dominant pattern, which means there is a 50% chance of an embryo being affected with the condition in each pregnancy if either parent has a relevant mutation.

1.8. The committee noted the penetrance of the condition is very high and thought to be close to 100%.

1.9. CFHR5 deficiency is characterised by kidney damage and progressive dysfunction, with high blood pressure and raised cholesterol. The condition often progresses to end-stage renal disease (ESRD) in middle age but occasionally presents in childhood or adolescence. Once ESRD develops patients will require regular dialysis and /or kidney transplant, without which the condition is fatal.

1.10. There is no cure for the condition but treatments can mitigate the symptoms.

1.11. The committee noted the inspectorate's request to consider CFHR5 deficiency, OMIM #614809 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 CFHR5 deficiency, OMIM #614809 is a very serious and potentially fatal condition. Patients with the condition, which has the potential to develop in childhood, may require dialysis and /or kidney transplantation which is not without risk and patients may die whilst waiting on the transplant list. The committee considered the severe effects on the quality of life of those affected by this 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 abnormalities 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.

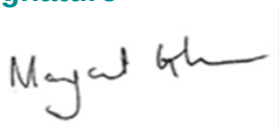
2.3. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- CFHR5 deficiency OMIM #614809

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
14 August 2019