Statutory Approvals Committee – minutes

Item 2

Centre 0327 (Boston Place)

Fanconi Anaemia, Complementation Group J, OMIM #609054

Thursday, 27 June 2019

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

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe Rachel Cutting Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Secretary Licensing Manager (Observer)
Specialist Adviser	Jenny Carmichael	
Legal Adviser	Sarah Ellson	Fieldfisher LLP
Observers	Dee Knoyle Vicky Brown	Committee Secretary Inspector

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 UK statement
- Statutory Approvals Committee Minutes- November 2018, PGD for Fanconi Anaemia Complementation Group N - OMIM #610832
- Licence Committee Minutes- July 2008, HLA-PGD for Fanconi Anaemia Complementation Group A - OMIM #227650
- Licence Committee Minutes- July 2008, HLA-PGD for Fanconi Anaemia Complementation Group C - OMIM #227645

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 Fanconi Anaemia Complementation Group J, OMIM #609054 was 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 Fanconi Anaemia Complementation Group J 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 each parent has a relevant mutation.

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

1.9. Fanconi Anaemia Complementation Group J is a rare genetic condition. It is characterised by congenital abnormalities in major organ systems, notably the heart and kidneys, skeletal defects, early onset bone marrow failure (average age between 2 and 6 years) and a significant risk of cancer, especially leukaemias, developing in childhood.

1.10. Fanconi Anaemia Complementation Group J can be fatal, and death can occur in utero, at birth or in early childhood. There is no cure for the condition and treatment focuses on managing the symptoms.

1.11. The committee noted the inspectorate's request to consider Fanconi Anaemia Complementation Group J, OMIM #609054 for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

1.12. The committee also noted the recommendation of the peer reviewer to consider other Fanconi Anaemia Complementation Groups for approval as conditions for which PGD can be applied. The committee noted that Fanconi Anaemia Complementation Group A, OMIM #227650, and Fanconi Anaemia Complementation Group C, OMIM #227645, have been approved for PGD since July 2008. Fanconi Anaemia Complementation Group N, OMIM #610832, has also been on the list of conditions for which PGD can be applied since November 2018. Clinically the other complementation groups are indistinguishable from each other and from Fanconi Anaemia Complementation Group J and follow an autosomal recessive inheritance, except for Group B, which is an X-linked recessive condition. There are no additional treatment options available for any of these conditions which are serious and life threatening. The complementation Groups are:

- Fanconi Anaemia Complementation Group B, OMIM #300514
- Fanconi Anaemia, Complementation Group D1 OMIM #605724
- Fanconi Anaemia, Complementation Group D2 OMIM #227646
- Fanconi Anaemia, Complementation Group E OMIM #600901

- Fanconi Anaemia, Complementation Group F OMIM #603467
- Fanconi Anaemia, Complementation Group G OMIM #614082
- Fanconi Anaemia, Complementation Group I OMIM #609053
- Fanconi Anaemia, Complementation Group L OMIM #614083
- Fanconi Anaemia, Complementation Group O OMIM #613390
- Fanconi Anaemia, Complementation Group P OMIM #613951
- Fanconi Anaemia, Complementation Group Q OMIM #615272
- Fanconi Anaemia, Complementation Group R OMIM #617244
- Fanconi Anaemia, Complementation Group S OMIM #617883
- Fanconi Anaemia, Complementation Group T OMIM #616435

1.13. The committee noted the risk of inheriting these conditions is 25% in each pregnancy if each parent carries a relative mutation. In the worst-case scenario, these conditions are serious and life threatening with no additional treatment options.

2. Decision

2.1. The committee considered that, in the worst-case scenario Fanconi Anaemia Complementation Group J, OMIM #609054 is a very serious multi-system condition that can be fatal in very early childhood, or even before birth. There is no cure for the condition and there is a serious risk of children with this condition developing leukaemia. The committee considered the severe effects on the quality of life of those and the families affected by this condition.

2.2. The committee considered that, in the worst-case scenario Fanconi Anaemia Complementation Groups B, D1, D2, E, F, G, I, L, O, P, Q, R, S, T are conditions of similar severity and clinical presentation to Fanconi Anaemia Complementation Group J.

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 abnormalities 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:

- Fanconi Anaemia, Complementation Group J, OMIM #609054
- Fanconi Anaemia, Complementation Group B OMIM #300514
- Fanconi Anaemia, Complementation Group D1 OMIM #605724
- Fanconi Anaemia, Complementation Group D2 OMIM #227646
- Fanconi Anaemia, Complementation Group E OMIM #600901
- Fanconi Anaemia, Complementation Group F OMIM #603467
- Fanconi Anaemia, Complementation Group G OMIM #614082
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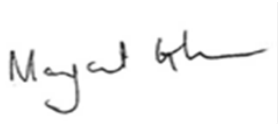
- Fanconi Anaemia, Complementation Group T OMIM #616435

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
22 July 2019