Statutory Approvals Committee – minutes

Item 3

Centre 0035 (Oxford Fertility)

Pre-implantation Genetic Diagnosis (PGD) application for Bartsocas – Papas Syndrome, OMIM #263650

Thursday, 25 April 2019

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

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Emma Cave Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Secretary Licensing Manager (Observer)
Specialist Adviser	Professor Mary Porteous	
Legal Adviser	Graham Miles	Blake Morgan LLP
Observers	Jennifer Rogerson Amanda Evans	Research Manager (induction) Research Manager (induction)

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

- 25 May 2017 Statutory Approvals Committee Minutes – PGD for Multiple Pterygium Syndrome, Escobar Variant, EVMPS, OMIM #265000
- 25 July 2013 Statutory Approvals Committee - PGD for Lethal Multiple Pterygium Syndrome, OMIM #253290
- 25 March 2010 Statutory Approvals Committee – PGD for Popliteal Pterygium Syndrome, OMIM #119500

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Professor Mary Porteous, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Bartsocas-Papas syndrome (Popliteal Pterygium syndrome, lethal type), OMIM #263650 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 Bartsocas-Papas syndrome 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 penetrance of the condition is 100%.

1.9. Bartsocas-Papas syndrome is a rare genetic condition caused by a mutation in the RIPK4 gene. During development in the womb, foetuses affected by Bartsocas-Papas syndrome have restricted growth. The condition causes malformation of the face, skin and genitals. The condition is usually fatal either before birth (intrauterine death) or during the neonatal period. Occasional survival into childhood has been reported.

1.10. There are no treatments available for Bartsocas-Papas syndrome. If a patient survives the initial neonatal period there may be major management issues around mobility and feeding.

1.11. The committee noted the inspectorate's request to consider Bartsocas-Papas syndrome (Popliteal Pterygium syndrome, lethal type), OMIM #263650 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 Bartsocas-Papas syndrome (Popliteal Pterygium syndrome, lethal type), OMIM #263650 is a very severe lethal condition. There is no cure for the condition and the majority of those affected die before birth or shortly after.

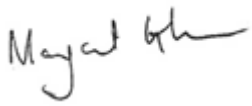
2.2. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, that a person with the abnormality will, given the conditions' worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition. 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. The committee agreed to authorise testing for:

- Bartsocas-Papas syndrome (Popliteal Pterygium syndrome, lethal type), OMIM #263650

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

15 May 2019