Statutory Approvals Committee – minutes

Item 5

Centre 0201 (Edinburgh Assisted Conception Unit)

Pre-implantation Genetic Diagnosis (PGD) application for Bare Lymphocyte Syndrome, Type II, complementation group A-E OMIM #209920

Thursday, 28 March 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	
Members of the Executive	Moya Berry Catherine Burwood	Committee Secretary Licensing Manager (Observer)
Specialist Adviser	Dr Alison Male	
Legal Adviser	Tom Rider	Fieldfisher LLP
Observers	Hannah Carpenter	Policy Officer

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

- SAC minutes 27 Feb 2014, PGD approval for SCID Adenosine Deaminase (ADA) deficient, OMIM #102700
- Licence Committee minutes, 30 August 2012 - PGD for SCID, OMIM #601457
- Licence Committee minutes, 14 June 2012 - PGD for SCID autosomal recessive, OMIM #600802

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 Bare Lymphocyte Syndrome, Type II, complementation groups A-E, OMIM #209920, 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 opinion provided a patient perspective and supported the application.

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 Bare Lymphocyte Syndrome, Type II, complementation groups A-E are 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. The condition is characterised by severe primary immunodeficiency. Patients are at an increased risk of developing serious and life-threatening infections. The damage caused by repeated and persistent infections of the respiratory, gastrointestinal and urinary tracts may lead to organ failure and most patients do not survive beyond early childhood.

1.10. Those affected by Bare Lymphocyte Syndrome, type II, complementation groups A – E, may experience severe and persistent diarrhoea, mucocutaneous candidiasis (infections of the skin, mucus membrane and nails), pneumonia and various recurrent bacterial infections. Repeated lung infections lead to bronchiectasis (widened airways within the lungs) and permanent damage to the lung tissue. Affected children have difficulty absorbing nutrients from food and as a result grow more slowly than their peers. Persistent infections may lead to organ failure and, without treatment, patients will not survive past early childhood.

1.11. There is no cure for the condition. Children with Bare Lymphocyte Syndrome, type II complementation groups A – E can be treated using bone marrow or stem cells transplants which may enable affected children to develop a degree of immunity to infection, but treatment may fail, or children may die of complications of the treatment. Some children also require a lung transplant if their lungs have become irreversibly damaged by recurrent severe infections.

2. Decision

2.1. The committee considered that in the worst-case scenario, Bare Lymphocyte Syndrome, Type II, complementation groups A-E is a serious condition which is characterised by severe primary immunodeficiency in infancy/early childhood. Patients are at an increased risk of developing serious and life-threatening infections. The damage caused by repeated and persistent infections of the respiratory, gastrointestinal and urinary tracts may lead to organ failure and most patients do not survive beyond early childhood. There is no cure for the 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 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.

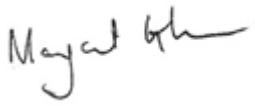
The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act and agreed to authorise testing for:

- Bare Lymphocyte Syndrome, Type II, complementation groups A-E, OMIM #209920

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

24 April 2019