Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Neonatal Inflammatory Skin and Bowel Disease 1 (NISBD1), OMIM #614328 and Inflammatory Skin and Bowel Disease 2 (NISBD2), OMIM #616069

Thursday, 31 October 2019

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

Committee members	Margaret Gilmore (Chair) Rachel Cutting Emma Cave	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Professor Mary Porteous	
Legal Adviser	Ros Foster	Browne Jacobson LLP
Observer	Alistair Robertson	DAC Beachcroft LLP

Declarations of interest:

- Members of the committee declared that they had no conflicts of interest in relation to this item.

Apologies:

- Apologies were received from Tony Rutherford and Bobbie Farsides

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 Mary Porteous, who confirmed that the conditions were as described in the papers.

1.2. The committee noted that the description in the PGD application for Neonatal Inflammatory Skin and Bowel Disease 1 (NISBD1), OMIM #614328 and Neonatal Inflammatory Skin and Bowel Disease 2 (NISBD2), OMIM #616069 is consistent with the peer review.

1.3. The committee noted that the conditions being applied for are 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 conditions 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 NISBD1, OMIM #614328 and NISBD2, OMIM #616069 are inherited in an autosomal recessive manner, which means there is a 25% chance of an embryo being affected by the conditions in each pregnancy if each parent has a relevant mutation.

1.8. The committee noted that the penetrance of both conditions is 100%.

1.9. Both condition types are characterised by inflammation of the skin and bowel leading to recurrent infections, bronchiolitis, dilated cardiomyopathy and viral myocarditis. Patients also experience chronic diarrhoea, malabsorption and a failure to thrive. Early death in childhood can occur in both conditions and onset occurs during the neonatal period (before one month).

1.10. There is no cure for NISBD1 and NISBD2, and treatment focuses on managing the symptoms.

1.11. The committee noted the executive's request to consider Neonatal Inflammatory Skin and Bowel Disease 1 (NISBD1), OMIM #614328 and Neonatal Inflammatory Skin and Bowel Disease 2 (NISBD2), OMIM #616069 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 Neonatal Inflammatory Skin and Bowel Disease 1 (NISBD1), OMIM #614328 and Neonatal Inflammatory Skin and Bowel Disease 2 (NISBD2), OMIM #616069 are serious debilitating multi-system conditions. There is no cure for the conditions which present during the neonatal period with death occurring in early childhood.

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 conditions' worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

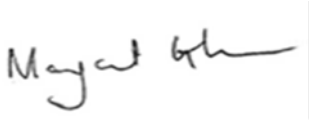
2.3. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for

- Neonatal Inflammatory Skin and Bowel Disease 1 (NISBD1), OMIM #614328
- Neonatal Inflammatory Skin and Bowel Disease 2 (NISBD2), OMIM #616069

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
18 November 2019