Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

RELN-associated Lissencephaly, OMIM #257320

Thursday, 30 March 2017

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Anne Lampe Anthony Rutherford	
Members of the Executive	Dee Knoyle Erin Barton	Secretary Governance Manager
External adviser	Dr Alison Male	
Legal Adviser	Sarah Ellson	Fieldfisher
Observers		

Declarations of interest

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

The committee had before it:

- 8th 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 Opinion
- Additional paper submitted with the application: 'American Journal of Medical Genetics Part C (Seminars in Medical Genetics) 166C:198–210.'

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 RELN-associated Lissencephaly, OMIM #257320 is consistent with the Peer Review.

1.3. The committee noted that the Genetic Alliance opinion provided a patient's perspective and supported the application.

1.4. 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.

1.5. The committee noted that the condition being applied for is not on the list of approved PGD conditions.

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 RELN-associated Lissencephaly is inherited in an autosomal recessive manner which means there is a 25% chance of having an affected child in each pregnancy, if each parent has a relevant mutation.

1.8. The committee noted that the condition demonstrates 100% penetrance and the onset of symptoms is from birth.

1.9. The committee noted that Lissencephaly (literally meaning ‘smooth brain’) is a congenital brain problem leading to absence or incomplete folding in the outer layers of the brain tissue (the cerebral cortex). This part of the brain is responsible for some movement and thought, as well as language skills and ‘higher functioning’. Lissencephaly is present from birth and is a life limiting condition.

1.10. Lissencephaly is associated with severe to profound developmental delay from birth including impaired motor and cognitive abilities, feeding difficulties and growth retardation. Children may not be able to speak or learn to sit or walk. Seizures occur in some cases and are often resistant to medication. Reported cases also have severe intellectual disability.

1.11. There is no curative or disease altering treatment for this condition. Supportive nursing care to help with feeding and comfort is needed. The range of symptoms impact significantly on the quality of life for the affected child and their family.

1.12. The committee noted the inspectorate’s recommendation to consider the approval of RELN-associated Lissencephaly to be included on the PGD List. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that the condition is serious. Babies are born with a brain problem, profound developmental delay including impaired motor and cognitive abilities, feeding difficulties and growth retardation. Children may not be able to speak or learn to sit or walk and may have severe intellectual disability and seizures which are often resistant to medication. Lissencephaly is a life limiting condition and the range of symptoms impact significantly on the quality of life for the affected child and their family.

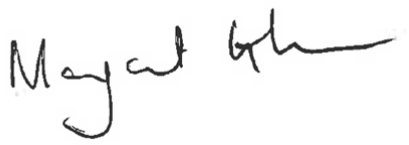
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 to the Act.

2.3. The committee agreed to authorise the testing of embryos for PGD for RELN-associated Lissencephaly, OMIM #257320. The committee agreed that the other name for this condition, Lissencephaly 2 (Norman-Roberts type) (LIS-2), OMIM #257320 should also be added to the list of approved PGD conditions.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

30 March 2017