Statutory Approvals Committee – minutes

Item 3

Centre 0101 (CARE Fertility Nottingham)

Pre-implantation Genetic Diagnosis (PGD) application for:

Rhizomelic Chondrodysplasia Punctata (RCDP1) Type 1, OMIM #215100
Rhizomelic Chondrodysplasia Punctata (RCDP2) Type 2, OMIM #222765
Rhizomelic Chondrodysplasia Punctata (RCDP3) Type 3, OMIM #600121
Rhizomelic Chondrodysplasia Punctata (RCDP5) Type 5, OMIM #616716

Thursday, 28 February 2019

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

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Ruth Wilde Rachel Cutting Emma Cave	
Members of the Executive	Dee Knoyle Moya Berry Catherine Burwood	Committee Secretary Committee Secretary (Observer) Senior Governance Manager (Observer)
Legal Adviser	Sarah Ellson	Fieldfisher LLP
External adviser	Dr Alan Fryer	
Observers		

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

1. Consideration of application

1.1. The committee welcomed the advice of its Specialist Adviser, Dr Alan Fryer who confirmed that the condition was as described in the papers.

1.2. The committee noted that the descriptions in the application for PGD for Rhizomelic Chondrodysplasia Punctata (RCDP) Types 1, 2, 3 and 5 are consistent with the peer review.

1.3. The committee noted that the conditions being applied for are not on the list of conditions approved for PGD.

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 Rhizomelic Chondrodysplasia Punctata Types 1, 2, 3 and 5 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 both parents have a relevant mutation. The conditions are 100% penetrant from birth.

1.8. The committee noted that Rhizomelic Chondrodysplasia Punctata is a form of skeletal dysplasia. Physical symptoms of RCDP are dwarfism and microcephaly (small head). The condition is characterised by shortening of the thighs and upper arms and joint deformities. Children diagnosed with RCDP have a specific bone abnormality called Chondrodysplasia Punctata, an unusual stippling at the end of the long bones visible on x-ray. Affected individuals have congenital contractures and limited movement, facial malformations, progressive cataracts, breathing difficulties, significant developmental delay and severe intellectual disability. Other symptoms may include seizures, ichthyosis (dry, thickened, scaly skin) and alopecia. Death occurs during the first decade of life in typical (classical) cases, mainly due to respiratory complications.

1.9. The committee noted that there is no curative treatment for this condition. Those affected have multiple medical problems, some of which can be treated surgically. The condition is associated with a poor quality of life and premature death is inevitable.

1.10. The Specialist Adviser informed the committee that Rhizomelic Chondrodysplasia Punctata types 1, 2, 3 and 5 are phenotypically very similar. The committee accepted this advice.

1.11. The committee noted the inspectorate’s request to consider whether Rhizomelic Chondrodysplasia Punctata Types 1, 2, 3 and 5 should be approved 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 Rhizomelic Chondrodysplasia Punctata Types 1, 2, 3 and 5 are serious conditions which are diagnosed at birth, and life threatening with death occurring in the first decade of life. Symptoms include seizures, respiratory complications and severe physical and intellectual disabilities. There is no cure. Treatment may include invasive surgery.

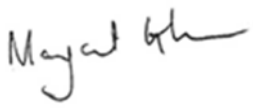
2.2. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, given the condition's worst symptoms, that a 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 following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act and agreed to authorise testing:

- Rhizomelic Chondrodysplasia Punctata (RCDP1) Type 1, OMIM #215100
- Rhizomelic Chondrodysplasia Punctata (RCDP2) Type 2, OMIM #222765
- Rhizomelic Chondrodysplasia Punctata (RCDP3) Type 3, OMIM #600121
- Rhizomelic Chondrodysplasia Punctata (RCDP5) Type 5, OMIM #616716.

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

14 March 2019