Statutory Approvals Committee – minutes

Centre 0101 (CARE Nottingham)

Pre-implantation Genetic Diagnosis (PGD) application for

Cerebro-oculo-facial-skeletal syndrome type 3, OMIM #616570

Monday, 23 January 2017

Church House Westminster, Dean's Yard, Westminster SW1P 3NZ

Committee members	Margaret Gilmore (Chair) Anne Lampe Ruth Wilde Tony Rutherford	
Members of the Executive	Dee Knoyle Siobhain Kelly	Secretary Interim Head of Corporate Governance
External adviser	Dr Ed Blair	
Legal Adviser	Tom Rider	Field Fisher
Observers	Bobby Farsides Bernice Ash	Member (Induction) Committee Secretary

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
- Email from the PR at centre O101 agreeing that COFS 1, 2 and 4 should also be considered for approval as conditions for which PGD can be applied.

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr Ed Blair, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Cerebro-oculo-facial-skeletal syndrome (COFS) type 3 is consistent with the Peer Review.

1.3. 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.

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

1.5. The committee noted that Cerebro-oculo-facial-skeletal syndrome type 3 is a severe, neuro-degenerative disorder, characterised by arthrogryposis (multiple joint contractures), microcephaly (small head size), growth failure, severe developmental delay, congenital cataracts, ultra violet light sensitivity of the skin and skeletal and craniofacial defects. Survival beyond six years of age is rare.

1.6. The committee noted that there is no curative treatment for the condition.

1.7. The committee noted that the peer reviewer had highlighted three other types of recessively inherited Cerebro-oculo-facial-skeletal syndrome (COFS) which have the same clinical features and level of severity as the condition applied for. These include:

- COFS 1 (OMIM #214150) caused by mutations in the ERCC6 gene (OMIM 609413);
- COFS 2 (OMIM #610756) caused by mutations in the ERCC2 gene (OMIM 126340)
- COFS 4 (OMIM #610758) caused by mutations in the ERCC1 gene (OMIM 126380).

1.8. The committee noted the inspectorate's recommendation to consider the approval of the four types of Cerebro-oculo-facial-skeletal syndrome (COFS) including Type 1, Type 2, Type 3 and Type 4 to be included on the PGD List and that the PR at centre 0101 had agreed to all four types being included as part of the application. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee had regard to its decision tree.

2.2. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph IZA(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'.

2.3. The committee noted that the condition is inherited in an autosomal recessive pattern and there is a 1 in 4 chance of an embryo being affected with the condition where both parent are carriers of relevant mutations.

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

2.5. The committee considered that the condition is serious due to the abnormalities children are born with, including a small head growth failure and multiple joint contractures and the severe developmental delay associated with this condition as well as its deterioration and reduced life expectancy.

2.6. 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 IZA(1)(b) and (2) of Schedule 2 of the Act.

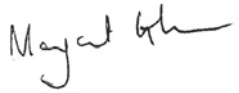
2.7. The committee agreed to authorise the testing of embryos for:

- Cerebro-oculo-facial-skeletal syndrome type 1 - COFS 1 (OMIM #214150)
- Cerebro-oculo-facial-skeletal syndrome type 2 - COFS 2 (OMIM #610756)
- Cerebro-oculo-facial-skeletal syndrome type 3 - COFS 3 (OMIM #616570)
- Cerebro-oculo-facial-skeletal syndrome type 4 - COFS 4 (OMIM #610758)

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

10 February 2017