Statutory Approvals Committee – minutes

Item 3

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Cataract 4 (CTRCT4), #115700

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 Anne Lampe Tony Rutherford	
Members of the Executive	Moya Berry Catherine Burwood	Committee Secretary Licensing Manager (Observer)
Specialist Adviser	Dr Alison Male (PGD)	
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
- August 2018 – SAC Minutes PGD for Congenital Cataracts Type 10 OMIM #600881
- January 2016 – SAC Minutes PGD for Congenital Cataracts OMIM #601885

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 Cataract 4 (CTRCT4), OMIM # 115700, 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 Cataract 4 is inherited in an autosomal dominant pattern, which means there is a 50% chance of an embryo being affected with the condition in each pregnancy if either parent has a relevant mutation.

1.8. The committee noted the penetrance of the condition is 100%; however, there is a great degree of variability in Cataract 4, such as the site and appearance of cataracts, even within the same family.

1.9. The committee noted the condition is a visual disorder presenting with dense congenital cataracts which need to be removed soon after birth as they impair vision by their presence and interfere with the normal development of vision which is occurring in the early part of life.

1.10. The committee noted that in its most severe form, congenital cataracts can cause complete blindness from birth or early childhood due to impairment of vision. This is particularly true where there have been secondary complications such as glaucoma, an increase of pressure within the eye. Glaucoma is painful and disabling for children and may be difficult to manage, requiring lifelong treatment with drugs or multiple surgical procedures.

1.11. The committee noted that congenital cataracts can be removed surgically, following which, children will need either an artificial lens implant or to be fitted with lenses or glasses. In cases where surgery is undertaken early and without complications, the outcome is not always guaranteed to be good and visual impairment may still result.

1.12. The committee noted the recommendation of the peer reviewer to consider a number of other forms of isolated hereditary cataract condition types, where the causative gene has been identified, for inclusion on the list for which PGD can be applied. All are individually rare but have a very similar course and are considered clinically similar to Cataract 4. These condition types are Cataract 1, OMIM #116200; Cataract 2, OMIM #604307; Cataract 3, OMIM #601547; Cataract 5, OMIM #116800; Cataract 13, OMIM #116700; Cataract 20, OMIM #116100; Cataract 31, OMIM #605387; Cataract 36, OMIM #613887; Cataract 46, OMIM #212500.

2. Decision

2.1. The committee considered that, in the worst-case scenario Cataract 4 #OMIM 115700 is a severe condition. A baby born with the condition may require repeated surgery , which may not be successful and may have future associated risks. Surgery may increase the risk of early onset glaucoma which in an infant or child may be extremely painful and require repeated , lifelong surgery. Treatments to mitigate the symptoms of the condition may not work and the patient may be registered blind at an early age . this can severely affect development . In the worst-case scenario the condition will have a severe impact on quality of life for the patient and their family.

2.2. The committee proceeded to consider the peer reviewer's request to approve Cataract 1, Cataract 2, Cataract 3, Cataract 5, Cataract 13, Cataract 20 Cataract 31, Cataract 36 and Cataract 46.

2.3. The committee agreed only to licence Cataract 1 and Cataract 2 as these conditions have a similar phenotype to Cataract 4 where the cataracts are congenital in their nature and present at birth. With regard to Cataract, 3, 5, 13, 20, 31, 36 and 46, the committee agreed with the advice of the specialist adviser who felt there was insufficient evidence to confirm the conditions were of a similar phenotype to Cataracts 1, 2 and 4 (i.e. cataracts are present at birth) and therefore did not meet the criteria for testing under this application.

2.4. 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.

2.5. 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. The committee agreed to authorise testing for :

- Cataract 4, OMIM #115700

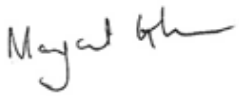
The committee also agreed to authorise testing for the following:

- Cataract 1, OMIM #116200
- Cataract 2, OMIM #604307

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