Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Hyaline Fibromatosis Syndrome (HFS), OMIM #228600

Thursday, 27 February 2020

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

Committee members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr Alan Fryer	
Legal Adviser	Tom Rider	FieldFisher - LLP
Observer	Emily Tiemann (Induction)	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 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 description in the PGD application Hyaline Fibromatosis Syndrome (HFS), OMIM #228600, is 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 UK statement provided a perspective on the impact of the condition 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 Hyaline Fibromatosis Syndrome (HFS), OMIM #228600, is inherited in an autosomal recessive pattern, which means there is a 25% chance of an embryo being affected by the condition in each pregnancy if each parent has a relevant mutation.

1.8. The committee noted the penetrance of the condition is close to 100%.

1.9. Hyaline Fibromatosis Syndrome (HFS), OMIM #228600, is characterised by deposition of abnormal fibrous tissue in tissues such as the skin, leading to thickened and pigmented skin over joints and skin nodules which may enlarge, increase in number and ulcerate. Thickened gums can cause feeding difficulties. It is also associated with progressive joint deformities that limit movement. In some cases, this condition includes chronic diarrhoea with a protein-losing enteropathy, and recurrent infections, which are the leading cause of death. Bones can become thin and fracture easily. The condition is also associated with severe pain in those affected. Those presenting in infancy are the most severely affected and fail to thrive and have skin involvement, congenital joint contractures and visceral involvement with persistent diarrhoea, recurrent infections, and death before 2 years of age. The later-onset forms (onset 3 months to 4 years) have milder skin and joint involvement and pain is less severe, but most teenagers and adults become bed-ridden and their quality of life is severely impaired.

1.10. There is no cure for this condition and treatment focuses on managing the symptoms.

1.11. The committee noted the executive's request to consider Hyaline Fibromatosis Syndrome (HFS), OMIM #228600, 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 Hyaline Fibromatosis Syndrome (HFS), OMIM #228600, is a rare, severely painful and life-limiting condition with symptoms present at birth. In severe cases death will occur before the age of 2 years. There is no cure for the condition and those who do survive into adulthood may be severely debilitated, requiring a lifetime of medical care. The committee noted the serious implications and impact on the quality of life for those affected with the condition.

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

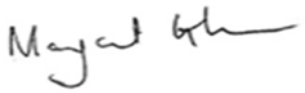
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Hyaline Fibromatosis Syndrome (HFS), OMIM #228600

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
17 March 2020