Statutory Approvals Committee – minutes

Item 1

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Woodhouse-Sakati Syndrome, OMIM #241080

Thursday, 25 April 2019

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

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Emma Cave Tony Rutherford Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Secretary Licensing Manager (Observer)
Specialist Adviser	Professor Mary Porteous	
Legal Adviser	Graham Miles	Blake Morgan LLP
Observers	Amanda Evans Jennifer Rogerson	Research Manager (Induction) Research Manager (Induction)

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, Professor Mary Porteous, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Woodhouse-Sakati Syndrome, OMIM #241080, 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 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 that Woodhouse-Sakati Syndrome is 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 each parent has a relevant mutation.

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

1.9. Woodhouse-Sakati Syndrome is a genetic condition caused by a mutation in the DCAF17 gene. The condition is characterised by hormonal effects, including hypogonadism, hypothyroidism and type 2 diabetes. Patients also experience effects on the neurological system, including muscle spasms (which may affect the ability to walk and lead to wheelchair dependency), and difficulties with speaking and swallowing. Patients may also experience hearing loss and mild learning difficulties.

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

1.11. The committee noted the recommendation of the peer reviewer to consider the inclusion of multiple sub-types of neurodegeneration with Brain Iron Accumulation within this application. The committee decided that as there was insufficient information provided to make a decision, it was not appropriate to consider the additional sub-types under this application.

1.12. The committee noted the inspectorate's request to consider Woodhouse-Sakati Syndrome, OMIM #241080, 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, Woodhouse-Sakati Syndrome, OMIM #241080 is a severe, disabling and painful condition which can develop in childhood and impact on daily living due to the accumulation of symptoms. These include mobility, hearing, speech and cognitive development issues. There is no cure for the condition and the committee noted that affected individuals living with this condition will require lifelong medical treatment. The committee considered the adverse physical and psychological impact on the quality of life for patients living with this 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 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.

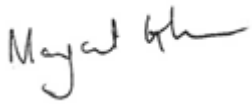
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Woodhouse- Sakati Syndrome, OMIM #241080

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

15 May 2019