Statutory Approvals Committee – minutes

Centre 0005 (Fertility Exeter)

Pre-implantation Genetic Diagnosis (PGD) application for BAP1 tumour predisposition syndrome (BAP1-TPDS), OMIM #614327

Thursday, 17 December 2020

HFEA, 2nd Floor, 2 Redman Place, London E20 1JQ via Teams Meeting

Committee members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr. Alison Male	
Legal Adviser	Tom Rider	Fieldfisher - LLP

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
- Comprehensive Study of the Clinical Phenotype of Germline BAP1 Variant -Carrying Families Worldwide (2018), Walpole et al

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 BAP1 tumour predisposition syndrome (BAP1-TPDS), OMIM #614327, 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 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 not licensed to carry out PGD and that the PGD cycle will be undertaken at Guys Hospital, centre 0102. The committee noted that it is acceptable for centres which are not licensed for embryo testing, to make applications for a condition to be approved for PGD by the HFEA. The committee noted that when a condition is approved for PGD by the HFEA, any centre licensed to carry out PGD can test for it. Although the centre making the application does not have a PGD licence, it had presented relevant information to enable the committee to evaluate the application.

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 BAP1 tumour predisposition syndrome (BAP1-TPDS), OMIM #614327, is inherited in an autosomal dominant manner, which means there is a 50% chance of an embryo being affected by the condition in each pregnancy if either parent has a relevant mutation.

1.8. The committee noted the penetrance for the condition is high and may approach 100% for some tumours.

1.9. BAP1 tumour predisposition syndrome (BAP1-TPDS), OMIM #614327, is characterised by a predisposition to develop a number of different cancers, notably melanomas of the eye and skin and malignant mesothelioma, which are often incurable and lead to early death.

1.10. Treatment is not always curative and focuses on early detection and surgical removal of the tumour at the earliest possible stage. Treatment for advanced cancers can include major surgery, chemotherapy, and radiotherapy.

1.11. The committee noted the executive's request to consider BAP1 tumour predisposition syndrome (BAP1-TPDS), OMIM #614327, 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, BAP1 tumour predisposition syndrome (BAP1-TPDS), OMIM #614327, is a very serious and potentially lethal condition, and given the increased risk of developing a number of different cancers, can result in death if not detected and treated at an early stage. Cancers have been reported in patients in their teenage years. Treatments for the cancers are often aggressive and include chemotherapy and radiotherapy and may not always be successful, particularly with regard to malignant mesothelioma. The committee considered the potentially devastating physical and psychological impact on the quality of life of those affected with the condition, the lifetime of surveillance they will require, and the associated uncertainty of these cancers developing.

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 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.

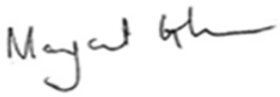
2.4. The committee agreed to authorise testing for:

- BAP1 tumour predisposition syndrome (BAP1-TPDS), OMIM #614327

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
14 January 2021