Statutory Approvals Committee – minutes

Centre 0119 (Birmingham Women's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Campomelic Dysplasia, OMIM #114290

Thursday, 30 August 2018

HFEA, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe Ruth Wilde Anthony Rutherford	
Members of the Executive	Catherine Burwood Bernice Ash Paula Robinson Stevan Cirkovic	Committee Secretary Committee Secretary (Observer) Head of Planning and Governance (Observer) Policy Officer (Observer)
Specialist Adviser	Professor Peter Turnpenny	
Legal Adviser	Jane Williams	Mills & Reeve LLP
Observers		

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:

- 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 UK statement

1. Consideration of application

1.1. The committee welcomed the advice of its Specialist Adviser, Professor Peter Turnpenny, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description of Campomelic Dysplasia, OMIM #114290 in the application for PGD is consistent with the peer review.

1.3. The committee noted that the Genetic Alliance opinion provided a patient perspective and supported the application.

1.4. 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.

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

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. Campomelic Dysplasia is inherited in an autosomal dominant manner, 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 condition is caused by a mutation in a gene known as SOX9 that is key in the early stages of foetal development. Therefore, the condition develops before birth. The majority of babies affected with Campomelic Dysplasia will have severe skeletal problems, including prenatal dwarfism and small ribcage - often resulting in death in the first year of life due to restricted breathing. Sometimes children born with Campomelic Dysplasia also have what is known as 46XY sex reversal, which means that, although they are genetically a male, they are born with female external genitalia and variable gonads that may comprise mixed male and female elements. This occurs in about 75% of cases that are genetically male.

1.9. Other symptoms include: cleft palate; hydrocephalus; absent olfactory bulbs; hypotonia and congenital heart disease. It is possible to reach adult life with this condition; however, most of those affected by the condition will die in the first year of life. Penetrance appears to be 100%.

1.10. There is no curative treatment for Campomelic Dysplasia, only supportive treatment, which comprises respiratory support including mechanical ventilation. Surgery may be required for scoliosis or cervical instability in survivors. Hearing aids may help hearing impairment. Sex reversal may affect the mental health of sufferers.

2. Decision

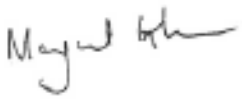
2.1. The committee considered that, in the worst-case scenario, Campomelic Dysplasia OMIM #114290, is a serious condition which affects the skeleton before birth and often leads to death in the first year of life. It is a severe multi-system disorder and management of symptoms is difficult. There is no curative treatment. The quality of life for affected individuals is severely impacted.

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, given the condition's worst symptoms, 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 Campomelic Dysplasia, OMIM #114290 meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act and agreed to authorise testing.

3. Chairs signature

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

Signature



Name

Margaret Gilmore

Date

26 September 2018