Statutory Approvals Committee - minutes

Centre 0102 (Guys Hospital)

Preimplantation Genetic Diagnosis (PGD) application for Chudley-McCullough Syndrome (CMCS), OMIM #604213

Date:	28 January 2021
Venue:	Microsoft Team Meeting
Committee Members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde
Specialist Adviser	Peter Turnpenny
Legal Adviser	Gerard Hanratty - Browne Jacobson LLP
Members of the Executive	Moya Berry - Committee officer Catherine Burwood - Licensing Manager
Apologies:	No apologies were received for the meeting
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
- Standard Licencing and Approvals Pack

The following papers were considered by the committee:

- Executive Summary
- PGD Application form
- Redacted Peer review
- Genetic Alliance (UK) Statement
- Academic paper: Doherty et al (2014)

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 in the PGD application for Chudley-McCullough Syndrome (CMCS), OMIM #604213 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 that Chudley-McCullough Syndrome (CMCS), OMIM #604213, is inherited in an autosomal recessive manner, 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 that the penetrance of the condition is uncertain but that there is a high degree of penetrance for deafness.

1.9. Chudley-McCullough Syndrome (CMCS), OMIM #604213 is characterised by severe to profound bilateral sensorineural hearing loss at a young age. This is commonly associated with communication and speech delay. The condition is also associated with brain abnormalities including hydrocephalus, which can cause seizures and intellectual disability.

1.10. There is no cure for this disease, but symptoms can sometimes be managed with bilateral cochlear implants for hearing loss, surgery to relieve intracranial pressure, and anti-epileptic medication to treat seizures.

1.11. The committee noted the executive's request to consider Chudley-McCullough Syndrome (CMCS), OMIM #604213, 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, Chudley-McCullough Syndrome (CMCS), OMIM #604213, is a rare, serious condition that presents in newborn babies/early infancy. Those affected can suffer from the rapid progression of profound, bilateral deafness usually by the age of three years. The condition may also be associated with specific structural brain abnormalities such as hydrocephalus and seizures. There is no cure for the condition and cochlear implant surgery, and surgery to relieve intracranial pressure from hydrocephalus, are not always suitable and are not without risk of serious complications. The committee considered the possible serious physical and psychological impact of those living 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 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.

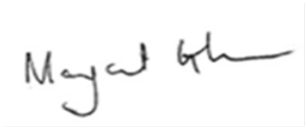
2.3. The committee was therefore satisfied that the following condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

- Chudley-McCullough Syndrome (CMCS), OMIM #604213

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

23 February 2021