Statutory Approvals Committee – minutes

Centre 0102 (Guy's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Congenital Deafness with inner ear agenesis, microtia and microdontia, OMIM #610706.

Thursday, 28 September 2017

Church House Westminster, Dean's Yard, Westminster SW1P 3NZ

Committee members	Margaret Gilmore (Chair) Anne Lampe Ruth Wilde Bobbie Farsides Anthony Rutherford	
Members of the Executive	Dee Knoyle	Committee Secretary
External adviser	Dr Alan Fryer	
Legal Adviser	Tom Rider	Fieldfisher 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 opinion
- Minutes of the Licence Committee on 10 October 2001 which approved Autosomal recessive non-syndromic sensorineural deafness, OMIM #220290, as a condition for which PGD can be performed

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 application for Congenital Deafness with inner ear agenesis, microtia and microdontia, OMIM #610706 is consistent with the Peer Review.

1.3. The committee noted that Genetic Alliance supported the application and provided information gathered in relation to the types of symptoms seen in congenital deafness with inner ear agenesis, microtia and microdontia and responses to a survey from patient groups.

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. The committee noted that the condition is inherited in an autosomal recessive pattern and there is a 25% chance of an embryo being affected with the condition if each parent has a relevant mutation.

1.8. Congenital Deafness with inner ear agenesis, microtia and microdontia (also known as LAMM) is a rare genetic condition which causes profound sensorineural deafness caused by absence of the structures of the inner ear. Symptoms are present from birth. This condition appears to be fully penetrant but is very rare with less than 100 cases reported in the medical literature so it is difficult to be definitive.

1.9. Affected individuals often have abnormally small, malformed external ears and can also have unusually small and widely spaced teeth. This condition can affect the development of motor skills, such as sitting, crawling and walking because the inner ear is important for balance as well as hearing.

1.10. There is no cure for this condition, but children will have specialist treatment from audiology, ophthalmology and dental specialists for an initial assessment and regular reviews and will be enrolled on early intervention educational programmes for hearing impaired children. Some may need additional educational support for any special needs which occur as a result of the balance problems. Despite early intervention to maximise hearing potential there are limits to how far this can be corrected. Children are at increased risk of accidents given hearing and balance disabilities and some normal childhood activities will not be easily available.

1.11. The committee noted the inspectorate's request to consider whether Congenital Deafness with inner ear agenesis, microtia and microdontia, OMIM #610706 should be approved for inclusion on the PGD List. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that Congenital Deafness with inner ear agenesis, microtia and microdontia is serious, given the significant risk of babies born with little or no structure to the inner and outer ear leading to deafness and an inability to balance resulting in poor motor skills and increasing the likelihood of accidents. The committee considered that cosmetic surgery may be required to build the outer structure of the ear but this is not treatment to assist hearing. The committee considered that the physical and psychological impact on an affected individual could also impact on the individual's quality of life.

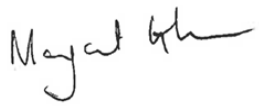
2.2. The committee had regard to its explanatory note and noted that on the basis of the information presented, given the condition's worst symptoms, it was satisfied that there is both a particular risk and a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition of Congenital Deafness with inner ear agenesis, microtia and microdontia, OMIM #610706 does meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

2.3. The committee agreed to authorise the testing for Congenital Deafness with inner ear agenesis, microtia and microdontia, OMIM #610706.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

12 October 2017