Statutory Approvals Committee – minutes

Centre 0102 (Guy's Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for

Paroxysmal extreme pain disorder (PEPD), OMIM #167400

Thursday, 26 October 2017

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

Committee members	Margaret Gilmore (Chair) Anne Lampe Anthony Rutherford Bobbie Farsides	
Members of the Executive	Bernice Ash Dee Knoyle Susanna Nyarko-Parkin	Committee Secretary Committee Secretary (Observing) Governance Officer (Observing)
External adviser	Mary Porteous	
Legal Adviser	Jane Williams	Mills & Reeve LLP
Observers	Gerard Hanratty	Browne Jacobson 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:

- 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
- One additional paper submitted with the redacted peer review – Tang et al.
- Email communication from the centre stating that they would like other similar conditions to be considered

1. Consideration of application

1.1. The committee welcomed the advice of its Specialist Adviser, Dr Mary Porteous, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the application for Paroxysmal extreme pain disorder (PEPD), OMIM #167400 is consistent with the peer review.

1.3. The committee noted that at the time this application was submitted an opinion from Genetic Alliance was not available.

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 dominant manner which means there is 50% chance of an embryo being affected with the condition if either parent has a relevant mutation.

1.8. The committee noted that Paroxysmal extreme pain disorder (PEPD) is characterised by periods of excruciating pain. Onset is generally noted at birth. However, in some cases symptoms may not be recognised until later life.

1.9. Affected individuals in the new-born period suffer from a slow heart rate and the heart can stop briefly. Later, sudden, severe attacks of pain are reported in various parts of the body, typically in the lower part, especially around the rectum (described as "worse than childbirth") but can also affect the head and face, especially the eyes and jaw as well as causing associated skin flushing and warmth, watering of eyes or nose, hypersalivation, and weakness related to the site of pain that can last from hours to days. The duration of the painful episodes can be from seconds to hours. The penetrance is seen to be very high.

1.10. The committee noted there is no cure for this condition, but patients with PEPD have experienced some benefit with carbamazepine. Nevertheless, some patients report no benefit or have had to stop treatment because of side-effects. Other neurological drugs have been tried or are being trialled but all can have significant side-effects. Non-traditional methods can be tried such as massage, acupuncture, cold compresses and heating pads, depending on the site of the pain. The impact on mental health can be very significant and quality of life can be significantly disturbed by the pain, with it described as the “worst pain ever experienced”.

1.11. The committee noted the inspectorate’s recommendation that the additional types of Paroxysmal extreme pain disorder, Inherited Erythromelalgia and Small-fibre neuropathy, both OMIM #133020, also be considered for inclusion on the PGD list.

1.12. The committee noted that the centre has confirmed that it would like to include the additional types of pain disorder caused by mutations in the SCN9A gene; Inherited Erythromelalgia and Small-fibre neuropathy, both OMIM #133020, in its application.

1.13. The committee noted the inspectorate's request to consider whether Paroxysmal extreme pain disorder (PEPD), OMIM #167400 should be approved for inclusion on the PGD List. The inspectorate also recommended that the committee considers whether to approve Inherited Erythromelalgia (IEM) and Small-fibre neuropathy (SFN), both OMIM #133020. The committee agreed to consider the application on this basis.

2. Decision

2.1. The committee considered that Paroxysmal extreme pain disorder (PEPD), OMIM #167400 is serious, given the significant risk of sudden, unprovoked and severe attacks of pain, impacting on an individual's everyday functions and the profound impact on the quality of life. This is a distressing and painful condition for which no reliable, therapeutic treatment is available.

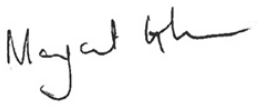
2.2. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk, given the condition's worst symptoms, that a 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 Paroxysmal extreme pain disorder (PEPD), OMIM #167400 meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

2.3. The committee agreed to authorise testing for Paroxysmal extreme pain disorder (PEPD), OMIM #167400. The committee also agreed to authorise testing for Inherited Erythromelalgia (IEM) and Small-fibre neuropathy(SFN), both OMIM #133020.

3. Chair's signature

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

Signature



Name

Margaret Gilmore

Date

16 November 2017