Statutory Approvals Committee – minutes

Centre 0044 (Centre for Reproductive and Genetic Health)

PGD for Congenital Adrenal Insufficiency OMIM #613743

Thursday, 29 August 2019

HFEA, Spey Meeting Room, Level 2, 10 Spring Gardens, London, SW1A 2BU

Committee members	Margaret Gilmore (Chair) Bobbie Farsides (Deputy Chair) Anne Lampe	
Members of the Executive	Moya Berry Catherine Burwood	Committee Secretary Licensing Manager (Observer)
Specialist Adviser	Dr. Alan Fryer	
Legal Adviser	Dawn Brathwaite	Mills & Reeve LLP
Observer:	Darryn Hale	DAC Beachcroft LLP

Declarations of interest

- Members of the committee declared that they had no conflicts of interest in relation to this item.
- Apologies were received from Rachel Cutting and Tony Rutherford.

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 UK statement
- Statutory Approvals Committee Minutes – 24 November 2016, PGD for X-Linked Adrenal Hypoplasia Congenita (XL-AHC), OMIM #300200
- Statutory Approvals Committee Minutes – 28 January 2016, PGD for 46XY Sex Reversal 6, OMIM #613762
- Statutory Approvals Committee Minutes – 29 October 2015, PGD for Partial Androgen Insensitivity Syndrome, OMIM #312300
- Licence Committee Minutes – 24 June 2010, PGD for Alpha Reductase Deficiency, OMIM #264600
- Licence Committee Minutes – 19 December 2005. PGD for Congenital Adrenal Hypoplasia, OMIM #201910

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 PGD application for Congenital Adrenal Insufficiency, OMIM #613743 was 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 Congenital Adrenal Insufficiency, OMIM #613743 is inherited in an autosomal recessive pattern, 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 the penetrance of the condition is 100%.

1.9. Congenital Adrenal Insufficiency is characterised by the insufficient production of cortisol, aldosterone and sex hormones. Cases can result in miscarriage, premature birth and early onset adrenal failure which is life threatening. Salt loss due to aldosterone insufficiency causes low blood sodium and high blood potassium, which can result in heart beat defects and seizures. Lack of androgen can lead affected individuals with an XY chromosome complement having female external genitalia.

1.10. There is no cure for the condition, and treatment focuses on managing the symptoms with life-long medication. However, life-threatening adrenal crises may not be completely prevented.

1.11. The committee noted the executive's request to consider Congenital Adrenal Insufficiency, OMIM #613743 for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

1.12. The committee also noted the executive's request for the change of name of the condition Congenital Adrenal Insufficiency, OMIM #613743. The condition is currently listed on the OMIM website as Adrenal Insufficiency, Congenital, with 46, XY sex reversal, partial or complete, OMIM #613743. The committee agreed to change the condition's name to ensure there is consistency with the OMIM website and the HFEA PGD list, if the application is approved.

2. Decision

2.1. The committee considered that, in the worst-case scenario, Congenital Adrenal Insufficiency, OMIM #613743 is a serious condition due to the on-going risk of adrenal crises which untreated can lead to death. In addition, the committee noted the requirement for life long

compliance with steroid treatment and the fact that affected individuals may experience delayed puberty and infertility.

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 abnormalities 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 conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act. The committee agreed to authorise testing for:

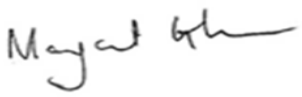
- Congenital Adrenal Insufficiency, OMIM #613743

2.4. The committee also confirmed that Congenital Adrenal Insufficiency, OMIM #613743 should be recorded on the HFEA PGD list as Adrenal Insufficiency Congenital, with 46, XY sex reversal, partial or complete, OMIM #613743.

3. Chairs signature

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

Signature



Name
Margaret Gilmore

Date
16 September 2019