Statutory Approvals Committee - minutes

Centre 0035 (Oxford Fertility Unit) – PGD application for Lacrimo-auriculo-dento-digita (LADD) syndrome OMIM #149730

Thursday, 29 October 2015

HFEA, Finsbury Tower, 103-105 Bunhill Row, London, EC1Y 8HF

Committee members	David Archard (Chair) Rebekah Dundas (Deputy Chair) Sue Price Margaret Gilmore Anthony Rutherford	
Members of the Executive	Trent Fisher Sam Hartley	Secretary Head of Governance and Licensing
External adviser	Dr Alan Fryer	
Legal Adviser	Graham Miles	Blake Morgan
Observers	Anna Quinn	Scientific Policy Officer

Declarations of interest:

- Members of the panel declared that they had no conflicts of interest in relation to this item.

The panel 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

1. Consideration of application

1.1. 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.

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

1.3. 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'.

1.4. The committee noted that the condition is inherited in an autosomal dominant and there is a 1 in 2 of an embryo being affected with the condition where one parent is affected.

1.5. The committee noted that the LADD syndrome is a very rare condition. Symptoms can include missing or abnormal tear ducts which can cause excessive tearing, absence of tears, dry irritable eyes and eye infections. Individuals may also have missing or abnormal of the major salivary glands causing dry mouth and a predisposition to early onset dental erosion.

1.6. The committee noted that other symptoms associated with this condition include abnormalities of the ears which can cause sensorineural and/or conductive hearing loss. Individuals may present with thumb defects ranging from small and undeveloped thumbs to missing thumbs. There also have been reported cases of individuals with LADD syndrome to present with kidney disease and genital abnormalities.

1.7. The committee noted that the condition is extremely variable and is likely to demonstrate a high penetrance. Symptoms will be present from birth in affected individuals

1.8. The committee noted that there is no curative treatment for the condition. Treatment will depend of the severity of the condition which can include surgery to correct problems with the tear ducts and limb defects. Artificial tear substitutes may be used to treat dry eyes. The impact of LADD syndrome on individual quality of life will vary dependent upon the severity of symptoms but repeated medical procedures can have a significant effect on individuals and treatment may not resolve all symptoms.

1.9. The committee noted that the application is consistent with the Peer Review.

1.10. The committee welcomed the advice of its specialist advisor, Dr Alan Fryer, who confirmed that the condition is as described in the papers adding that the condition is extremely variable even within the same family.

2. Decision

2.1. The committee considered that the condition is serious due to the unpredictable cumulative impact of the symptoms associated with the condition and repetitive invasive surgery that an individual will need.

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 a significant risk that a person with the abnormality will have or develop a serious physical

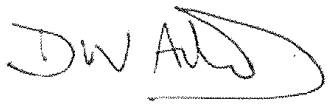
or mental disability, a serious illness or any other serious medical condition. The committee was therefore satisfied that the condition meets the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 to the Act.

2.3. The committee agreed to authorise the testing of embryos for Lacrimo-auriculo-dento-digital syndrome, OMIM #149730

3. Chair's signature

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

Signature



Name

David Archard

Date

11 November 2015