HFEA Licence Committee Meeting

29 September 2011

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

Minutes – Item 1

Centre 0102 (Guy's Hospital) – PGD for Stickler Syndrome type 1 (OMIM#108300/ #609508), 2 (#604841), 3 (#184840) and autosomal recessive (#120210)

Members of the Committee:	Committee Secretary:
David Archard (lay) – Chair Debbie Barber (professional) Anna Carragher (lay) Sally Cheshire (lay) (videoconference) Mair Crouch (lay) (videoconference) Rebekah Dundas (lay) (videoconference) Sue Price (professional)	Terence Dourado Legal Adviser: Graham Miles, Morgan Cole

Declarations of Interest: members of the Committee declared that they had no conflicts of interest in relation to this item.

The following papers were considered by the Committee:

- Executive Summary
- PGD application form
- Redacted peer review
- Redacted supporting email from patient (dated August 2011)

The Committee also had before it:

- HFEA Protocol for the Conduct of Licence Committee Meetings and Hearings
- 8th edition of the HFEA Code of Practice
- Human Fertilisation and Embryology Act 1990 (as amended)
- Decision trees for granting and renewing licences and considering requests to vary a licence (including the PGD decision tree); and
- Guidance for members of Authority and Committees on the handling of conflicts of interest approved by the Authority on 21 January 2009.
- Guidance on periods for which new or renewed licences should be granted
- Standing Orders and Instrument of Delegation
- Indicative Sanctions Guidance

- HFEA Directions 0000 – 0012
- Guide to Licensing
- Compliance and Enforcement Policy
- Policy on Publication of Authority and Committee Papers
- HFEA Pre-Implantation Diagnostic Testing (“PGD”) Explanatory Note For Licence Committee

Tabled document

- Genetic Alliance Opinion

1. The Committee had regard to its Decision Tree. The Committee was satisfied that the Centre has considerable 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.

2. The Committee noted that the Centre’s proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of schedule 2 of the Act, ie. ‘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’.

3. The Committee noted that Stickler Syndrome is inherited in two patterns; type 1 (OMIM#108300/#609508), 2 (#604841), 3 (#184840) are inherited in an autosomal dominant manner. Only one copy of the affected gene is required to cause the disorder, i.e. there is a 1 in 2 chance of the embryo being affected in a family where one parent is affected and the other is unaffected; Stickler Syndrome autosomal recessive (OMIM#120210) is inherited in an autosomal recessive manner, if an embryo inherits a copy of the faulty gene from both parents it will develop the disease, ie there is a 1 in 4 chance of the embryo having the abnormality.

4. The Committee considered that there is a significant risk that a person with the abnormality will develop a serious medical condition because it is almost fully penetrant by adulthood.

5. The Committee considered that the condition, which is a multisystem disorder, is serious because it affects the eyes, facial bone structure, hearing, skeleton and joints. The symptoms associated with Stickler Syndrome may be apparent at birth, childhood or adulthood depending upon the severity of the disorder. The symptoms vary in severity between affected individuals and even within the same family some affected people

will be more significantly affected than others. Eye problems are related to very severe myopia (short-sightedness) leading to detachment of the retina; around 4% will have complete visual loss. Cataracts (clouding of lenses) and glaucoma (high pressure in the eye) are also found. Facial bones are affected due to the under development of the jaw bone and nasal bridge. This can make eating difficult due to poor contact between the upper and lower jaw. Cleft palate is common. 90% of those affected with Stickler have joint laxity (over flexible joints) and resulting in joint pain and degeneration of the joints leading to osteoarthritis. Hearing loss is common although the severity is variable with some children being significantly affected and some adults experiencing minimal loss. Treatment will include surgical repair of cleft palate, cryotherapy (freezing) or laser treatment for retinal detachment, hearing aids and mobility aids to help with movement which may be affected by the join laxity and joint degeneration. Surgical joint replacements are also used when necessary. Learning difficulties can be present as a result of hearing loss and visual problems. Some individuals experience a curvature of the spine and heart valve problems. Sticker syndrome type 3 has similar symptoms to types 1 and 2, but without related eye problems.

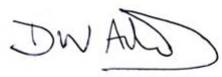
6. The Committee recognised that there are four distinct types or syndromes listed and that there is a distinct set of symptoms associated with each type of syndrome.

7. Paragraph 10.5 of the Code of Practice (8th edition)/ HFEA guidance for Centres states: ‘The use of PGD should be considered only where there is a significant risk of a serious genetic condition being present in the embryo. When deciding if it is appropriate to provide PGD in particular cases, the seriousness of the condition in that case should be discussed between the people seek treatment and the clinical team. The perception of the level of risk for those seeking treatment will also be an important factor for the centre to consider’. The Committee noted that clinics should satisfy themselves of the above, particularly in respect of the non-ocular types of the condition.

8. The Committee had regard to its explanatory note and considered that on the basis of the information presented, the Committee 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. Accordingly, it was appropriate to grant the application under paragraph 1ZA(1)(b) of Schedule 2 to the Act.

9. The Committee agreed to authorise the testing of embryos for Stickler Syndrome type 1 (OMIM#108300/ #609508), 2 (#604841), 3 (#184840) and autosomal recessive (OMIM#120210). The Committee confirmed that this condition will be added to the published list of conditions for which PGD may be carried out.

Signed: Date: 11/10/2011



David Archard (Chair)