copy.

Joanna Pamballi
Student ID: 090149368
Word count: 1172

Elective Report: Fetal Medicine at the Harris Birthright Centre, King's College Hospital London

For the second part of my elective I observed the activities taking place at the Harris Birthright Centre (King's College Hospital, London). This is a leading specialist unit in Fetal Medicine and research, attracting patients from the whole of the UK and doctors in training from the whole world, under the supervision of Professor Nicolaides.

Objectives: 1. What is the prevalence of Down's Syndrome (DS) and what is the importance of screening in the UK?

Down's syndrome is the most common chromosomal abnormality, associated with a prevalence of 1 in 700 live births. In the 20th century the development of DS screening occurred, now integrated in the National Screening programme, offered routinely to all pregnant women in the UK. The importance of screening is colossal. It allows women with high-risk to carry out diagnostic testing and know as early as the first trimester whether the pregnancy is affected. This provides a window of opportunity for decisions to be made concerning the fate of the pregnancy and setting of the appropriate medical, financial and psychological framework for the arrival of the affected child. Additionally it enables women with low-risk to rest assured for the remaining part of the pregnancy that the child is unlikely to be affected. However, importance is not solely associated with the risk assessment. DS screening invites women to be assessed in hospital and provides opportunistic viability and fetal anatomy assessment identifying major abnormalities.

2. Compare the provision of routine screening to cell free DNA (cfDNA) testing for calculating DS risk?

The current national screening programme for DS includes combined screening at $11\text{-}13^{+6}$ weeks of gestation. This is a combination of blood biochemistry (βhCG and PAPP-A), ultrasonographic measurement of the nuchal translucency and maternal age. I was lucky to experience assessment of additional markers such as absent nasal bone and tricuspid regurgitation, which also included into the algorithm at this specialist centre. All the above, provide the pregnant woman with a risk of the pregnancy being affected with DS, expressed as a ratio of 1:x. Low risk is considered nationally as <1:150 and consequently high-risk >1:150. Women in the high-risk group are offered diagnostic invasive testing.

The discovery of cell-free DNA in maternal blood revolutionised screening for DS. It is now feasible with a single blood test to classify a pregnancy as very high or very low risk for DS as early as 10 weeks gestation. The advantages over routine screening include: higher DR, lower FPR limiting the number of patients requiring invasive testing, implementation from 10 weeks onwards, simplicity, independent of operator skills, no risk to the fetus and the ability to detect the gender something particularly important for some medical conditions or generally for some parents. However, it is significantly more costly, in 5% of cases there is failure to generate a result and it introduces a window of 2 weeks between testing and results, while not assessing for abnormalities in fetal anatomy.

3. Discuss the acceptability and impact of a new policy for DS screening.

In King's College Hospital there is an undergoing trial of offering women with intermediate risk, from the combined test, a cf-DNA test. Women with a positive

result on the cf-DNA test (very high risk) are subsequently offered an invasive test for diagnosis. It was particularly interesting to observe the consultations. Every single woman offered the cf-DNA test accepted it, while some patients expressed disappointment for not qualifying for it. The simplicity of the test, being a blood test, increases the acceptability of screening for DS and targets the population that would not consider invasive testing due to the risk it poses to the fetus. Implementation of cf-DNA classifies women as very high or very low risk eliminating the intermediate risk group which is discomforting for the patients and the clinicians. Should this prove cost-effective there is the potential of national screening changes. One suggested model could involve cf-DNA testing at 10 weeks with an ultrasound scan and discussion of results +/- invasive testing at 12 weeks. This however reverses the success of a one-stop clinic and some women might be lost between appointments.

Introducing cf-DNA under regulated qualifications can cause a major impact to the policy for screening, as it may provide the numbers required to increase DR to 100%. Cost will also be reduced by the limitation in the numbers requiring CVS and potentially replacing invasive testing in the future. However, as it stands at the moment it will not replace the $11\text{-}13^{+6}$ scan as it cannot yet provide all the answers. I do believe though that offering it on the NHS screening programme will eventually increase the number of women opting for screening. An obstacle that will need to be overcome however will be the regulations of the new policy as to the characteristics qualifying a patient for cf-DNA.

4. Further my understanding and knowledge of Fetal Medicine and the challenges posed in the practice of this field.

This experience has not only enriched and deepened my knowledge of Fetal Medicine, but has also fed my enthusiasm to pursue a career in this field.

Every moment is treasured and valued in its own way. It is difficult to describe the warmth and satisfaction it gives you looking at the parents when they hear their unborn child's heartbeat for the first time, or watch it move on the screen. I found it fascinating how my understanding improved by the day, becoming increasingly more able to identify anatomical features on a fetus measuring only about 6cm in length. Being at a leading clinical centre provided me with a daily encounter of pathologies I had previously only identified in books and journals, some of which include: hypoplastic left heart syndrome, twin-to-twin transfusion syndrome (TTTS), congenital diaphragmatic hernia etc. I was present when Professor performed procedures like tracheal ballooning of diaphragmatic hernia and intrauterine laser ablation of placental vessels for the treatment of TTTS.

However it was not always pleasant, as I observed breaking bad news, explaining that the fetus has died or that severe abnormalities had been detected some compatible and others non-compatible with life. It was particularly hard to deal with the pain and despair of some parents, but it was a challenge for me to overcome this and improve. I found myself observing professional manners I once hope to develop. Ultimately, the most challenging aspect is the ethical issues arising in cases of fetal anomalies, and being able to distinguish which is the best route to follow. I guess it is important to expel all prejudgement and treat each case individually having the benefit of the fetus and the mother as a guide.

These 3 weeks have been a very valuable experience for me. It has given me insight into the present and future of Fetal Medicine, has improved my understanding of fetal ultrasonography, built onto previous knowledge, improved my communication skills and has armed me with confidence and determination to work towards a career in this field.