

Antenatal Diagnosis and Management of Fetal Abnormalities

The incidence of congenital abnormalities is approximately 2% - 3%. Of this number 50% are minor and 50% are deemed to be major. This latter category would break down into abnormalities that result in major disability, death or requiring major surgery. Thus most parents can look forward to a healthy normal baby and are seeking to be reassured in their pregnancy that their expected baby is normal.

There are three main methods of providing this much wanted reassurance but unfortunately this is not forthcoming in all cases. In Ireland ultrasound is probably the main method of diagnosing babies in utero who have anatomical abnormalities. These can be diagnosed at any time in pregnancy from about 10 weeks onwards and throughout pregnancy. Most hospitals, though not all, provide what is called an anatomy scan between about 19 and 22 weeks. This is in line with best international practice and this is often the time in pregnancy when an abnormality is diagnosed. In addition to ultrasound there are invasive procedures called amniocentesis and chorionic villus sampling, which involve placing needles into the pregnant uterus and obtaining either a sample of amniotic fluid or a sample of the placenta. This provides some fetal cells which can then be examined in the laboratory, most often to look at the chromosomal make-up of a baby and occasionally tell about inheritable genetic problems. More recently, in the past three years, there has been a major development in prenatal diagnosis with the advent of non-invasive prenatal testing. It is now possible to obtain a blood sample from the mother and in the laboratory be able to extract some fetal DNA from this sample. This can be analysed using quite sophisticated molecular techniques and give 99% accuracy for the diagnosis of common chromosome problems which would include Downs, Edwards and Patau Syndrome. It is also possible to tell the gender. This test can be done as early as 10 weeks and can be done at any stage in the pregnancy. Initially this test was very expensive but with a greater uptake from patients the price has come down considerably and is likely to come down further, making it more accessible to patients. At the current time the State does not pay for this form of testing and thus the expectant mother needs to pay for this test.

When a diagnosis of a fetal abnormality is made using any of the above techniques it is extremely important that patients be given this information in a clear, easy to understand, sensitive manner. The initial breaking of the news is often done by the midwife ultrasonographer who is the person who most frequently carries out the ultrasound at 19 - 22 weeks. If there is a suspicion or diagnosis of a fetal abnormality these patients will usually be referred to a Fetal Medicine Specialist.

There are now, in recent years Fetal Medicine Specialists in all of the major hospitals though not in the smaller peripheral hospitals, though there may be a Consultant Obstetrician that has a particular interest in prenatal diagnosis. The Fetal Medicine Specialist will give a full

explanation of the diagnosis though the exact diagnosis may not always be known. This will include, in as much as possible, a discussion on the prognosis of the particular abnormality. Where there is an abnormality with a significant risk of death or serious physical and / or mental handicap all management options are discussed. This would happen in a supportive and non-directive manner. In the situation where a fetal abnormality is diagnosed the vast majority of women attending our service continue the pregnancy. However, an increasing minority of women who have babies with major abnormalities are choosing not to continue the pregnancy.

Where patients choose to continue the pregnancy they are given full consultant care away from the busy antenatal clinic. In addition they will have specialised midwifery care. Arrangements will be made for them to meet with the Neonatologist (baby doctor) or relevant specialist. e.g. if a baby has spina bifida they will meet the Neurosurgeon or if a baby has a heart problem they will meet with the Paediatric Cardiologist in order to discuss the diagnosis and prognosis in more detail, and to be informed of what is likely to happen after birth in the form of surgery or medical care. Patients are given a direct telephone number to have instant contact with a specialised midwife. Social work, chaplaincy and bereavement support is organised as appropriate to the situation. A multi-disciplinary meeting takes place every week where these difficult cases are discussed and a plan of management is agreed on and entered into the notes.

There are an increasing number of mothers / parents who are deciding not to continue the pregnancy which involves travelling abroad. We as health professionals are not allowed to refer, promote or advocate termination of pregnancy but we are allowed to provide all of the information to the relevant unit in the UK. The most commonly used referral hospital is in Liverpool where a sensitive caring service is provided with excellent feedback from patients. The cost of flights, accommodation and the medical procedure is borne by the patient. One of the difficulties with travelling abroad is the frequent lack of a post-mortem. Thus it is not possible in a number of cases to pinpoint the diagnosis and thus give appropriate counselling for a future pregnancy. It is important for the patient to be aware of what a termination of pregnancy entails and thus this would be explained to them in advance of their travel abroad.

There are three main methods. The first is what would be called medical and this involves giving tablets that promote uterine contractions and delivery of the baby, secondly a surgical procedure where under anaesthetic the neck of the womb is dilated and the contents of the pregnant womb are evacuated, and thirdly induction of labour. In 2015 3,451 Irish women, who gave their address as being in Ireland, underwent a termination of pregnancy. Of these women 135, or 4%, had a termination of pregnancy under Section E of the 1967 Abortion Act. This is the section that states that termination is carried out because of a risk of a serious physical or mental handicap or death. Of these 135 women 69 were

carried out because of a chromosomal problem (40 with Downs Syndrome, 13 with Edwards Syndrome and 7 with Patau Syndrome and 9 other chromosome problems). The remaining terminations were carried out because of anatomical abnormalities involving various different systems in the body. The most common were brain / spine abnormalities of which there were 23. There were 10 heart abnormalities, 4 kidney / bladder abnormalities and 13 musculoskeletal system abnormalities.

It is likely that in the future access to ultrasound in Ireland will improve as will the operators and the technology. This will probably result in an increasing number of pregnancies being diagnosed with fetal abnormalities. In addition to this there is definitely going to be an increase in non-invasive prenatal testing as this becomes cheaper and more accessible. This will lead to an increased number of pregnancies being diagnosed with chromosomal abnormalities. It is also likely that with further advances in molecular genetics that there will be an increased number of conditions diagnosable with this non-invasive blood test.