The availability of non-invasive prenatal diagnostic procedures such as sonography appears to have led to a higher rate of acceptance of such procedures, especially in younger patients. The expectations of pregnant women and their partners concerning prenatal diagnosis focus on reducing uncertainty in regards to the normality of the pregnancy. However, any prenatal diagnosis procedure can cause emotional stress in the pregnant woman. There are two different types of stress in this situation. There may be anxiety about the invasive nature of the procedure and the attendant risk of the loss of a wanted pregnancy. Separately, there may be anxiety over the outcome of the investigation. Any unexpected finding, especially an abnormal one, often has an adverse impact on the pregnant woman and her partner. With the advent of this now commonly shared experience, the male partner also gives up his role as a passive onlooker and becomes more actively involved in the pregnancy. Couples require multidisciplinary care if an abnormal foetus is diagnosis.

Keywords: Pregnancy, Foetus, Prenatal medicine, Anxiety.

Main Document

Prenatal diagnosis is supposed to reduce perinatal mortality and morbidity by recognizing or excluding the morphological, structural, functional, chromosomal or molecular fetal abnormalities. A range of investigations is available to the pregnant woman, all of which are focused on the detection of potential abnormality in the fetus. The advances in “risk free” sonography have changed the relationship between pregnancy and prenatal care. A “genetic scan” includes a detailed examination looking for fetal morphological abnormalities (“markers”) which are associated with chromosomal aberrations [1]. The classic morphological marker for chromosomal abnormality is the nuchal translucency, usually performed in the late first trimester (NT). Nuchal translucency is the maximum thickness of the subcutaneous translucency between the skin and the soft tissue overlying the cervical spine. The risk for a chromosomal abnormality rises with increasing thickness of NT and the risk is reduced below the background when a thin NT is found. Chromosomal abnormalities that are related to changes in nuchal translucency include the trisomies, 21, 13 and 18, Turner's Syndrome and the triploidies. In the course of a second trimester sonographic examination between the 16th and 20th week of gestation, additional morphological markers may aid in detecting chromosomal abnormalities. Some “soft markers” for Down's syndrome include increased space between 1st and 2nd toes, short femur and humerus, flat facial profile and short nasal bones. Although soft markers are often associated with a significant increase in foetal risk for chromosomal abnormality, they are seldom diagnostic by themselves. The combination of several markers increases the detection rate but also the false positive rate. If no abnormalities are detected following malformation screening by sonography, the theoretical risk of chromosomal abnormalities in that foetus is reduced by approximately 50%. Apart from sonography, there are varieties of other screening and diagnostic tests in use. However, in terms of psychological issues, they are similar and they will not be considered separately. A new test which used cell-free foetal DNA (cfDNA) circulating in the maternal blood stream, which is also called non-invasive prenatal testing (NIPT), will result in a change of paradigm. Respective studies indicate that in screening for trisomy 21, the detection and false-positive rates are 99% and 0.1%, respectively. For trisomies 18 and 13, there is less evidence but recent studies report detection rates of 98% and 86% [2]. The impact of non-invasive prenatal testing (NIPT) on anxiety and prenatal diagnosis is yet unexplored.

Care of the woman undergoing any prenatal diagnosis procedure, including sonography should not focus exclusively on detection of abnormality in the foetus, to the exclusion of all else. The clinician should be sensitive to psychological issues the couple may have in relation to these procedures. The procedures may strain the relationship between the pregnant woman, her partner and the unborn child.

The expectations of the pregnant woman and her partner attending for prenatal diagnosis are focused on reducing their uncertainty concerning their pregnancy. Most pregnant women...
who seek prenatal diagnosis want to have their expectation of a normal child confirmed and any uncertainty about the unborn child being unhealthy removed [3]. The commonest prenatal diagnosis procedure is sonography. By seeking the demonstrating of normal foetal morphology, the pregnant woman seeks to be reassured about her baby. It is common for parents-to-be to access all available resources to gain certainty about the normality of the foetus. For example, some women obtain repeated scans, from different providers, in this quest.

However, seeking the confirmation of normality of the foetus often produces anxiety [4]. Coping with this stress can involve external and internal mechanisms [5]. The expectations, which are placed on prenatal medicine clinicians, signify an activation of internal coping strategies. Psychoanalytically, the hope that the prenatal examination will confirm the healthy state of the unborn baby is interpreted as a defence mechanism against the anxiety and guilt felt by the mother for putting the pregnancy under scrutiny in the first place. Such a defence mechanism protects the pregnant woman and enables her to have a relationship with the baby. The overwhelming majority of women who are confirmed to have a normal baby feel internal justification for their action and will in fact benefit from this means of defence.

The function of the prenatal diagnosis is to recognize morphological, structural, functional, chromosomal and molecular defects before birth. The presence of an abnormal finding is called “positive” and the absence of abnormal finding is described as “negative” by clinicians. Therefore, there is often a diametrically opposed construct of what a “positive” result is between the patient and her attending doctor. To the pregnant woman and her partner, the adjective “positive” is related to well being and normality. The adjective “positive” means “affirmative, favourable, promising, advantageous, certain”. However, to the clinician, the word positive, in the context of prenatal diagnosis, often denotes the detection of an abnormal foetus.

The care of a woman who has been diagnosed to be carrying an abnormal fetus should be multidisciplinary [6,7]. The care is often complex, not only physically but also psychologically. Management options should be discussed with the woman. Issues related to whether a termination is warranted, the likely prognosis of the baby if termination is not chosen and a whole host of related issues require multidisciplinary expertise and input. Clinicians have to be sensitive to the fact that very often the woman may seek certainty to enable her and her partner to make difficult decisions. On the other hand, certainty, for example, in prognosis may not always be possible. The dilemma for the advice-seeking couple is thus; whilst they have to consider whether to terminate the pregnancy or not on the one hand and on the other, not knowing how to consider the specific life situation of the child if born alive.

A consideration of the psychological issues involved in prenatal diagnosis can help improve care and the experience of pregnancy in women. Pregnant women have already adapted to the increased capability of modern technology and expectations have changed substantially. The desire of parents-to-be to obtain a confirmation of the good health of the foetus during pregnancy is central to this new expectation. In psychological terms, prenatal diagnosis is an anxiety inducing procedure, independent of whether the procedure is invasive or not. However, the finding of normality can reduce this anxiety [8]. The image of the unborn baby at sonography endows the foetus with a real identity and imparts it an existence independent from her in the eyes of the woman. Seen this way, sonography can be viewed as an introduction to parenthood, albeit a technology mediated one. The identification of an abnormal foetus is usually unexpected and its impact all the greater because of it. It imposes an enormous psychological burden and may arouse inner conflicts and fears. This is compounded by the necessity to make difficult decisions within a limited time span. The couple in this predicament require multidisciplinary care and support during this difficult time [6].

Conclusion

Prenatal diagnosis have resulted in great technical advances in the medical attendance of pregnant women in the past thirty years. From a psychological perspective is Prenatal Diagnosis is like a double bind. Prenatal Diagnosis induces anxiety and reduces anxiety.

References