GeNeDit

Emotional Burden of Prenatal Screening

Editorial

Prevention of disorders with significant morbidity and mortality, by prenatal diagnosis, is an acceptable option for most families. It is a boon for families who already have or had a patient with such a disorder and are anxious about the possibility of recurrence. Primary prevention by population-based screening for carrier parents and pregnant women, for common disorders, has also gained acceptance. Large experience is available about screening for trisomy 21 and some common monogenic disorders like thalassemia, cystic fibrosis and some population-specific disorders and is getting accepted by the population. Increased awareness about the disorders makes population-based screening possible and the families without experience of a disorder in concern have enough knowledge about the disorder to make informed decisions.

Ease and increased detection rates by technical advancements are obvious over the last few years in screening for trisomy 21. Now, non-invasive screening for aneuploidies of all chromosomes, known microdeletion syndromes and copy number variations (CNV) has become available to the population. The tests based on latest techniques immediately become available in the market and are offered to the patients. The positive predictive values of the screening tests for rare aneuploidies are low. Hence many more patients have to undergo invasive testing. More important is the anxiety created by the screen-positive results. The magnitude of anxiety in screen-positive families has not been documented but genetic counsellors and obstetricians have enough experiences of intense emotional disturbances faced by screen-positive families. In spite of pre-test counselling, tremendous anxiety is experienced by the pregnant woman, who is in an emotionally vulnerable situation and a similar situation is faced by her family members. Many times, screening tests are offered one after the other and the whole process of screening and waiting for results spans over many weeks. Added are the dilemmas in cases of screen positivity or true positivity for sex chromosomal aneuploidies, many of which namely trisomy X or XYY do not justify termination of pregnancy. In this era of assisted reproductive techniques, XXY and monosomy X also have become manageable disorders. Also, one wonders if disorders without significant physical or mental handicap justify prevention by termination of pregnancy. Inclusion of sex chromosomal abnormalities in non-invasive prenatal screening though possible, needs reconsideration for inclusion in screening.

The GenExpress in this issue discusses many advances in non-invasive prenatal screening (NIPS). The options seem to be increasing. Families at risk of serious genetic disorders are greatly helped by these developments. But before these tests are offered to the low-risk general population, one needs to understand the utility of screening including the burden of uncertainties, dilemmas and anxiety of the screened population and acceptance and quantification of the emotional cost involved. The GenExpress talks about non-invasive screening for monogenic disorders with great specificity. However, for many disorders there may be phenotypic variability. The phenotype also may not be easy to understand. For example, the phenotype of Marfan syndrome or Ehlers-Danlos syndrome may be beyond the understanding of a lay person to make informed decisions. Non-directive counselling is the backbone of genetic counselling. But for this, the understanding about the disorder in concern should be adequate. For numerous genetic disorders, educating the families adequately so that they can take a decision about accepting or not accepting the screening test is a great challenge. Even after theoretical explanations, one wonders if they will have adequate understanding about the disorder. Even a clinician or a genetic counsellor who has seen patients with these disorders will face difficulty in deciding about screening for monosomy X or Marfan syndrome. Just telling that the counselling is non-directive and passing the responsibility of making decisions in situations with dilemmas onto the patient and the family is not correct. Decisions regarding inclusion of the disorder in screening programs and to develop enough capabilities for pre and post-test counselling are prerequisites and may be limiting factors in screening programs. Without adequate counselling support, screening tests for a large number of rare disorders may do more harm than good. It is time to stop and think: how many and which disorders to screen in the general population? Just screening for

aneuploidies, leads to a few months of anxiety during the nine months period of gestation for many families, for preventing one trisomy 21 out of around 800 pregnancies. More screening will increase the anxiety, duration of anxiety and number of pregnant women facing anxiety. It may not be good for the pregnant woman who is traditionally pampered and is supposed to remain happy throughout her pregnancy.

Everyone wants a healthy baby but balance between the efforts for prevention of genetic disorders and the emotional burden of screening tests should be achieved, especially when prevention is by termination of pregnancy!

Dr. Shubha Phadke

1st July 2022