

# Genetic Counselors: Plan for the Best Utilization of the New Armamentarium

## Editorial

Genetic counseling is an integral and very important part of the management of genetic disorders, as lack of curative treatment and possibility of recurrence in the family are the realities for many genetic disorders even in the twenty-first century. Genetic counseling is a demanding and challenging scientific and communication skill and successful genetic counseling helps the family or the consultand in understanding the disease, the possibilities of recurrence, and ways to take reproductive decisions in addition to understanding the available treatment and possible outcomes. The process of getting educated about the disease makes the concerned person and family get control over the situation and helps them to adjust and cope up with the situation in a better way. Genetic counseling services thus are an integral component of medical genetics units and patient care can be enhanced by specialist genetic counselors. Training programs for genetic counselors are well established in the western world and the demand for such programs is rapidly increasing. India has also been seeing developments in this direction and recently, an update on Genetic Counseling was held in Manipal. Genetic counseling program directors from USA shared their experience and provided insights into their programs which are running for more than two decades.

Genetic counselors are trained in genetic aspects of the disorders and communication skills. With this background and better availability of time, they can reinforce the genetic information for better understanding of the patients and their families and provide the required continued support to them while they are adjusting to the genetic problem and in decision making. They can also help in organization and implementation of investigations and ancillary management services offered / advised by the clinical geneticist or physician. The main strength of genetic counselors is the knowledge of genetic aspects of diseases on the background of communication skills and availabil-

ity of adequate time which may be constraints for the clinician. The Medical Genetics specialty has taken roots in India and awareness about the specialty amongst clinicians and lay persons is rapidly increasing and the same is true regarding the availability of genetic investigations. Hence, the need for genetic counselors is being felt by the medical community and a few training programs in genetic counseling have started in the country. It is now time to think in what ways the genetic counselors should work in the environment of medical genetics and medicine, so that benefits of the latest exciting developments in medical genetics reach the patients in India.

The role of genetic counselors can be seen in two settings. The first is as a resource person for the antenatal screening programs for trisomy 21, thalassemia, neural tube defects and newborn screening programs for treatable disorders. These are routine services that every obstetric centre should provide and appropriate pre-test and post-test counseling is essential for the success of these screening programs. These tests if offered without pre-test counseling will not be done at the appropriate time and quite often result in undue anxiety and misinterpretation by the family. The concept of population-based background risks, of screening tests, of the probability associated with the results, and the need for conducting the test in the appropriate time frame, together with the knowledge of invasive confirmatory tests are the pre-requisites for the success of any population-based program. As these tests are to be offered to all, there is a real and great need for genetic counselors in each obstetric set up to run population-based screening programs. Though the doctors would want to, they may not be able to give enough time to convey all the information. The genetic counselors can be the resource persons for such population-based screening programs and manage the program in addition to providing pre-test and post-test counseling in individual or group sessions.

The other role of the genetic counselors is in Medical Genetics departments where patients with common and rare genetic disorders are seen. After the clinical geneticist or a specialist clinician like a neurologist or a haematologist makes a diagnosis of a genetic disorder, the process of genetic counseling starts. In this situation, the genetic counselors sit in the session for counseling with the medical geneticists or the clinician and then continue with the family by providing reinforcement of the information, answering queries and walk the consultant through the process of acceptance, reproductive decisions and organizing prenatal diagnostic tests. Here also, the ability of the genetic counselor to give time and communicate is the strength of the process. As many clinical specialists like neurologists, ophthalmologists, dermatologists, oncologists, cardiologists and practically all medical specialists see loads of patients with genetic disorders, the hospitals catering to these specialists will need genetic counselors who closely work with the clinician and provide genetic counseling. Genetic counselors may get specialized in a subspecialty under the supervision of the clinical specialist. Such specialised genetic counselors can also be resource persons for patient support groups and take up the responsibility of advocacy-related activities. In Medical Genetics departments or specialised hospitals, genetic counselors are also involved in counseling for research participation and informed consent taking, counseling about the utility and challenges of techniques like next generation sequencing (NGS). In the present era when NGS-based testing is becoming the first tier test, genetic counselors have the additional responsibility of interpreting and communicating the results of NGS in collaboration with

the NGS laboratory and clinician who usually are not comfortable with the uncertainties of results which are common with NGS.

Increasing availability and access to genetic investigations and prenatal diagnostic facilities makes the need of genetic counselors felt strongly and the training programs need to come on a uniform platform and their role as a complementary and integral part of medical genetics services in hospital set ups needs to be clearly defined. It needs to be emphasized that medicine is a very complex and wide faculty and genetic disorders are extremely heterogeneous and numerous. The knowledge of genetic disorders is very rapidly increasing. This poses great challenges to the practice of the rapidly evolving field of medical genetics. The diagnostic challenges of rare genetic disorders are enormous. Intricacies of the underlying bases of genetic disorders and complexities of the rapidly advancing vast genetics knowledge are beyond the scope of short term training programs of genetic counselors from non-medical background, but their expertise in the genetic aspects of genetic disorders can be judiciously used in clinical / hospital settings for the benefit of the patients and families with common genetic disorders. At the same time, it is essential to incorporate more genetics training in medical curricula and train practicing clinicians in clinically applicable modern genetics. Genetic counselling is a patient-centric activity and needs to function under the supervision of clinicians who need to get ready for the era of genomic medicine!

  
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