Common Genetic Disorders: Every Clinician's Responsibility

Editorial

Though we do not have prevalence data for most genetic disorders in India, sufficient prevalence data is available for at least some of the common genetic disorders such as beta thalassemia, Down syndrome and neural tube defects. The facilities for primary and secondary prevention of these common disorders are available in India and are rapidly increasing. At the same time, awareness about these disorders amongst clinicians and laypersons is also increasing. However, the level of facilities, knowledge of clinicians and awareness are markedly variable. There are no government-supported screening programs and neither are there any specific guidelines given by scientific or medical bodies. These facilities are not available at present in most of the medical colleges and hence, the approach to these common genetic problems is not being included in the practical training of medical students at the undergraduate or postgraduate levels.

In addition to prevention of birth of a first child with any of these genetic disorders, genetic counseling of the families with one child affected with a genetic disorder is very important. To identify a family at risk of a genetic disorder, a three-generation pedigree and screening of members of the extended family are important steps. These have been well illustrated in the two cases reported in this issue. Both the families had two genetic disorders in the family and due to timely genetic counseling and investigations, prenatal diagnosis could be offered to the at-risk pregnancies. In clinical genetics departments such coincidental occurrences of two genetic disorders in a patient or in a family are seen, though not commonly. Hence, it is important to offer screening for common genetic disorders to all families irrespective of the genetic disorder in the proband. As most of the patients with Down syndrome, thalassemia and neural tube defects are seen by pediatricians, they need to be confident about providing genetic counseling to the families and interpretation of the genetic tests. Similarly, obstetricians also need to play a more proactive role in the identification of families at-risk of genetic disorders by taking a detailed family history and offering screening tests. It is high time that every clinician be well conversant with pedigree drawing, basic genetics and principles of genetic counseling. Common genetic disorders can be easily tackled by most of the clinicians, especially those who are interested and have even had short term training in medical genetics.

This can partly happen by spreading basic knowledge about genetics applicable in clinical practice through short term training programs for clinicians at various stages of their careers. However, for future generations we need to impart an understanding of the clinical applications of medical genetics to undergraduate and postgraduate students. Training of committed medical college teachers from various specialties like pediatrics, obstetrics, pathology, anatomy, biochemistry, internal medicine, oncology, etc. and helping them to collaboratively set up genetic units in each medical college is the only way to take clinical genetics to all clinics. Training of teachers will have rapid, long term and multiplying effects. We need to take the fruits of research in genetics to the masses through clinicians from all specialties.

In this regard, I am glad to inform that the Indian Council of Medical Research has already initiated plans for creating a Network of Genetic Centers in medical colleges and I hope this initiative proves to be successful in achieving its goal of disseminating the knowledge of genetics to all medical practitioners.

Dr. Shubha R Phadke
1st October, 2014