Editorial

Medical Genetics in India – What needs to be done?

Genetics in India has come a long way, but it still has even a longer way to go. All that we have been able to achieve so far is some demonstration of its utility, creation of some demand and generation of interest among medical students and trainee doctors. Yet a large mass of medical profession, public at large and government remains largely indifferent to the potential of medical genetics for public good. A major limiting factor could be the high cost of genetic tests and lack of their accessibility. Equally important is the lack of exposure to medical genetics in medical education and trained manpower in the specialty.

Genetics in public health

It needs to be appreciated that strength of genetics lies in prevention of disease and not its cure. It therefore needs to be taken up as public health measure for prevention of physical and mental handicap, cancer and other debilitating disorders rather than just a clinical superspeciality. More than 50 per cent of clinical services of genetic centers abroad are engaged primarily in newborn screening for inborn errors of metabolism, screening of pregnancies for birth defects and chromosomal disorders and screening of adult population for various genetic markers including those for cancer, coronary artery disease, and more recently, for drug/xenobiotic metabolizing enzymes. One national program that can transform the scenario of medical genetics in India could be the control of thalassemias and haemoglobinopathies. Such a decision is to be taken at a political level than at the scientific or professional level. Another national programme could be periconceptional folic acid supplementation for the prevention of neural tube defects which have an occurrence of 5 in 1000 births. Other applications of genetics are in fields of foetal medicine and cancer. These disciplines could well take medical genetics to newer heights.

Reliable and affordable genetic tests

The Achilles heel in medical genetics continues to be the laboratory infrastructure for genetic tests. This needs to cover 3 distinct areas viz., cytogenetics, biochemical genetics and molecular genetics, each requiring specially trained manpower, highly specialized and sophisticated instrumentation and a blend of research and clinical service culture to provide prompt and reliable results of ever increasing number of tests. Sequencing and microarray are now becoming a part of routine clinical genetics laboratory rather than just research tools. There is also a need for strong quality control of genetic tests, requiring accreditation and monitoring of test results. In addition, there should be a statutory requirement of pre-test and post-test counseling to discuss the need and utility of testing, interpretation of test results and action/s to be taken in response to them. Both the clinical and laboratory components need to be developed as a composite unit for efficient functioning. It is a new paradigm for the medicine of tomorrow. The testing needs to be covered by health insurance to make access equitable, and by confidentiality/privacy laws to protect individuals and families from discrimination. It needs to be noted that currently medical insurance companies even in the west do not cover all genetic tests and all countries are grappling with the issue of discrimination on basis of inherent genetic make up of the individual.

Networking of clinics and laboratories

The uniqueness of genetic disorders is variety and rarity. In medicine proficiency comes by numbers, whereas most individual genetic disorders are rare having the prevalence of 1:5000-15000 births, or even more. At the same time there are several thousand genetic disorders, affecting all systems of the body. So, how do we achieve proficiency in genetics? If we wish...
to achieve competency there must be pooling of clinical and laboratory resources – in a collaborative network. Fortunately DNA is a hardy material and can be easily transported. Regarding clinical material, we should make use of the internet. We should evolve standardized clinical databases and be benefited by the clinical expertise wherever it exists. The interest and expertise in genetics must percolate to all specialties and super-specialties. Since the variety is so immense, we must restrict specialized laboratory investigations, area wise, to different laboratories according to their interest and expertise so that greater expertise is developed for wider coverage. There is a need to develop medical genetics in an organized manner as a medical school/hospital activity rather than as research tool.

Education in medical genetics

At a larger level, there is a strong need towards improving the standards of medical education across the country with specific focus on New biology, which is largely an offshoot of advances in genetics, both in undergraduate and postgraduate medical curriculum. The role of genes in influencing the risk of disease, its course and response to therapy, now pervades virtually all branches of medicine. These subjects constitute a major part of articles published in core general medical journals like NEJM, BMJ and Lancet, etc. Although the Medical Council of India has incorporated quite a bit of genetics in medical curriculum, but most medical colleges are ill prepared for it21-24. The Council should seriously consider prescribing establishment of Medical Genetics unit/department as an integral component of medical colleges. To begin with, it can be a part of any of the major clinical departments, viz., Medicine, Paediatrics or Obstetrics and Gynaecology. The MCI should also ensure that evaluation of knowledge of Medical Genetics and its application is incorporated in the final certifying examination. It would not be trite to say that without this knowledge the practitioner of medicine of tomorrow would be practically illiterate in its scientific aspects.

S.S. Agarwal
Former Head of the Department of Medical Genetics
Sanjay Gandhi Postgraduate Institute of Medical Sciences
Lucknow 226 014, India
agarwal_ss2000@yahoo.com

References


