

Should Genetics be Limited to Advanced Centers or Department?

Kuldeep Singh

We have now come to a crossroad where many specialties are meeting together mainly due to advance in technology, development of expertise and a need for collaboration. The new information has created gaps in our knowledge and understanding of human being and the diseases. At the turn of this millennium we have witnessed the impact of Human Genome Map which culminated in massive progress in Genetics due to concerted work of biologists, physicians and basic scientists. Genetics in India has developed very fast in last 4 decades thanks to painstaking and consistent efforts by the founder geneticist supported by many biomedical scientists and researcher. Health services demands have increased. These pioneering stalwarts in Genetic Medicine have time and again emphasized the need for Community genetics through various platforms and fora. Largely they were successful in igniting interest and generating demands by establishing Genetics in India [1, 2]. However, they raised their voice for not letting genetics restricted to few rare disorders at specialized centers using expensive technology but to make it more accessible to population at large for common conditions. They also called for involvement of policy makers and a political will to do so [1].

It has now been recognized by young generation of Medical geneticist that there is lot to be done in India. Handshake is required between clinical scientists, basic scientists and public health researcher to have some impact on community. Advance level genetic technologies are being set up and used at centers at Lucknow, New Delhi, Manipal, Bangalore, Mumbai, Pune, Hyderabad and Ahmedabad [3]. It is really gratifying to know that this young generation is highly focused in their

approach. However, their expertise is accessible to a very small segment of our population. Time has come that we have to take the field to community convincing policy makers and educating public at large. There is also a need for interdepartmental collaboration. Genetics unite not divide. If we just review the development of Genetics in India, there are many unsung heroes who have worked relentlessly to the cause of genetics. In our medical course don't we remember that brush with the field of genetics was carried out by Anatomy department? Later it was joined by biochemistry and physiology. Pediatricians come across the diseased child in their first contact. Their curiosity, perseverance, patience and efforts mostly lead to diagnosis [4]. However, they have to depend heavily on expertise of their biochemist, pathologist or a geneticist counterpart for confirming their suspicion. Not to forget advances in radio-imaging and nuclear scan in the further workup. A time comes when the diagnosis has to be discussed with the parents and their family. A hurriedly told diagnosis have grave consequences not only on child and their parents but has deep penetrating effect on family and their community which may even lead to discrimination, guilt and social outcast. Here comes the role of social and psychological assessment for preparedness for the condition.

Time has also come that we tread outside of labs, department and hospital to community. Hitherto considered to be mainly environmental conditions are now having major role of Genomics [5] and goes beyond newborn screening. The perceived role of genetics in public health is changing, as is the definition of what is a genetic disease. The role of genetics in public health is broadened if we consider all the diseases for which genetics might play a role,

Author's Affiliation: Additional Professor & Head, Department of Pediatrics, All India Institute of Medical Sciences, Jodhpur-342005.

Corresponding Author: Kuldeep Singh, Additional Professor & Head, Department of Pediatrics, All India Institute of Medical Sciences, Jodhpur-342005.
Email: singhk@aiimsjodhpur.edu.in

either by the presence of a genetic susceptibility for the development of this disease or for response to treatment, or by the presence of protective genetic factors, such as in resistance to infection.

This issue of IJGMR will be having articles dealing with reproductive failures among males wherein DNA fragmentation can impact fertility. Urticaria is a very common condition but hereditary angioedema (HAE) can at times be life threatening. A review will highlight the role of genetics in HAE. Role of Micro RNAs in diagnosis and therapy is being discussed for Osteo-arthritis. The issue also has a review on prospects of DNA fingerprinting in diagnosis and lineage study.

Fetal autopsy at times play a very crucial role in unearthing underlying developmental defect not apparent clinically on physical examination. How a pediatrician got relieved from guilt of failed intubation due to congenital anomaly is being highlighted through a case report. The case emphasizes role of fetal autopsy which is fast being replaced by newer imaging technologies. Diagnosis of rare condition help relieve parent from fear of unknown and offer a scope for management is being highlighted in a case on Fibrodysplasia Ossificans Progressiva.

References

1. Agarwal SS. Medical genetics in India- What needs to be done?. *Indian J Med Res* 2009; 130: 354-356.
2. Verma IC, Bijarnia S. The burden of genetic disorders in India and a framework for community control. *Community Genet* 2002; 5: 192-196.
3. Agarwal S, Phadke SR. Medical genetics and genomic medicine in India: current status and opportunities ahead. *Mol Genet Genomic Med* 2015; 160-171.
4. Phadke S, Gowda M. Genetic Testing in Children, *Indian Pediatr* 2013; 50: 823-827.
5. Bowen MS, Kolor K, Dotson WD, Ned RM, Khoury MJ. Public Health Action in Genomics is now needed beyond newborn screening. *Public Health Genomics* 2012; 15: 327-334.

