



EDITORIAL

Clinical Genetics and Genetic Counselling in Pakistan

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ABSTRACT

Monogenic hereditary diseases are diseases caused by defects in genes and currently it has been estimated that there are more than 10,000 monogenic diseases with a global prevalence of 10/1000 at birth. According to estimation more than 350 million people are affected by monogenic genetic diseases globally. Thalassaemia, sickle cell anaemia, colour blindness, haemophilia, deafness, etc., are some examples of monogenic hereditary diseases. Pakistan is among the countries which are on high alert for rapid increase or such genetic disorders. The reasons for high prevalence of hereditary disorder are many: one is consanguinity due to cultural preference of cousin marriages and mate selection from near or distant blood relatives. Thus 80 percent consanguinity in Pakistan, is perhaps the highest in the world. The incidence of genetic diseases at birth can be reduced through introduction of premarital genetic counselling as a part of healthcare system. Genetic counselling and premarital genetic testing should be practiced in Pakistan to stop this rapidly increasing burden of hereditary diseases.

Keywords: Clinical genetics, genetic disorders, monogenic disorders, genetic counselling, consanguinity, Pakistan

Clinical genetics is termed as the clinical practice dealing with hereditary or genetic disorders, which include genetic counselling to modern therapeutic approaches for treating genetic disorders. It has been described that almost all monogenic disorders can be eradicated / eliminated by genetic counselling or awareness among people. Genetic clinics can also deal with a number of clinical problems such as birth defects, developmental delay, autism, epilepsy, short stature, etc. Some common genetic

disorders are thalassaemia, cystic fibrosis, Duchene muscular dystrophy, hereditary breast cancer (BRCA1, BRCA 2), Huntington disease, deafness, etc. Disorders due to chromosomal aberrations are many and worldwide: some very common are: Down syndrome, DiGeorge syndrome (22q11.2 deletion syndrome), fragile X syndrome, Turner syndrome, etc. Completion of human genome project and introduction of modern whole genome sequencing programmes, like CLINSEQ® Study, have made us capable to look into the genes and to predict future health status of society [1, 2]. Last decade has been witnessed for a

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number of researches explaining phenotype-genotype relationships explaining genome and environmental interactions for genetic disorders such as thalassemia [3].

There is a vast scope of clinical genetics in Pakistan because of consanguinity, which is as high as 80 percent [4]. The common practice of preference to marry a cousin and / or near or distant blood relative has led to a culture of marriage bondage within intra caste or isonym groups all over Pakistan [5]. Our rural population is almost totally ignorant of deleterious effects of consanguinity. In urban areas less than 20 percent people know about harmful effects of the so called "cousin marriage". This awareness is caused by the appearance of some mental, physical or reproductive disability in a family, which has been diagnosed by treating physicians as due to cousin marriage. Recent studies have explained severity of such disorders in Pakistan such as polydactyly [5] and inherited retinal disease, [6]. Elite class of Pakistani community, including doctors, lawyers, medical students and members of parliament are very positive for genetic diagnosis at an early stage. It was also found that the families of patients suffering genetic disorders also showed very positive attitudes for genetic diagnosis as they are well aware about their sufferings [7]. It has also been surveyed that 77% of our elite class favours premarital screening of carriers of recessive disease [8]. As genetic counselling is still not a part of Pakistani healthcare system, it has been described that more than 90% of our medical doctors are ready to refer their routine patients to the genetic counsellors if they found some possible genetic defects. Medical practitioners are worried about the rapid increase of genetic disorders and paucity of genetic counsellors in the country [9]. As the genetic diseases are on the increase, partly due to population explosion in Pakistan, a rapid development in genetic counselling services is required throughout the country. Human resource development of genetic

counsellors will help to rise in the market of genetic developments and applications of novel therapeutic approaches in cells-based and gene-based approaches as a step to prevent the spread of genetic disorders [10]. It has been proposed that monogenic genetic diseases are preventable. For primary prevention of genetic disabilities, whether intellectual or physical or pertaining to reproduction, early detection and genetic counselling are inevitable tools for primary prevention of disabilities [11].

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