

Genetic Counseling in Developing Countries

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ABSTRACT Genetic counseling is a vital component for the diagnosis and the management of genetic disorders. A number of pre-requisites are essential for appropriate genetic counseling. The current definition of genetic counseling is based on the state of clinical genetics in the developed countries. Presented here are some of the challenges in the genetic counseling in the developing countries and some suggestions for solutions.

INTRODUCTION

Genetic counseling is defined as the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to the disease. This process integrates:

1. Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence,
2. Education about inheritance, testing, management, prevention, resources and research and
3. Counseling to promote informed choices and adaptation to the risk or the condition.

This definition, put forward by the National Society of Genetic Counselors (National Society of Genetic Counselors' Definition Task Force 2006), was based on a much evolved field of Genetic Counseling in the developed countries, like the United States of America. In recent years, developing countries have recognized the role of genetics in human disease and the need for genetic counseling services in these countries.

It is important to examine if the pre-requisites for genetic counseling are available in the developing countries. Such an exercise would help in identifying elements that will need development or enhancement to successfully foster the field of genetic counseling. A robust genetic counseling service would reflect on the diagnosis and management of genetic disorders.

OBSERVATIONS AND DISCUSSION

Recognizing or suspecting a genetic basis for a disorder is essential for identifying conditions in patients that may have a genetic basis. Physicians in almost all the clinical specialties

have the opportunity to encounter patients with genetic disorders. With access to information on the internet, patients themselves and family members are empowered with the knowledge to suspect a genetic disorder. In such instances the skills of a trained clinician are required for confirming a diagnosis before genetic counseling can be provided. Trained clinical geneticists and access to these professionals is very important for appropriate genetic counseling. To fully appreciate the number of trained Clinical Geneticists required for a given population size, one could look at the numbers in the United States. According to the American College of Medical Genetics and Genomics in 2007, there were approximately 1100 Board Certified MD geneticists in the U.S. for a population of 301 million. This ratio of 1: 274,000 was considered inadequate. It is essential that, in the developing countries, clinicians in all the disciplines should be provided with the required training (Agarwal 2009) to identify or suspect a genetic disorder, so that if required the patient is referred for evaluation by a clinical geneticist, and genetic counseling by a trained genetic counselor.

Just as there is a need for trained Clinical Geneticists in sufficient numbers, there is also a need for sufficient numbers of trained Genetic Counselors (Elackatt 2013). Counseling for genetic disorders should include helping the patient and/or the family adapt to psychological and familial implications. An in-depth understanding of the family dynamics and the cultural norms is vitally important for genetic counseling. The scientific and technical component of genetic counseling is universal; however, techniques involved in genetic counseling have to incorporate the social and the cultural norms and the sensitivities of the society. Training of

genetic counselors should include training to evaluate the family for receiving the information as well as training on social and cultural norms. Language is also a factor to be considered in genetic counseling in the developing countries. Majority of the population in the developing countries use a language other than English. Hence creating a vocabulary in the local languages to adequately explain the scientific information and recurrence risk is an important requirement for informative genetic counseling in the developing countries is required. Training genetic counselors from within communities/populations to receive genetic counseling services would likely be most ideal and beneficial for addressing the issue of social and cultural norms and language.

The diagnosis of genetic disorders requires the use of a variety of laboratory techniques. The choice of technique depends on the condition under consideration. While some diagnoses require readily available techniques (like chromosome analysis) others may require less readily available techniques such as biochemical assays, mutation studies (including sequencing of genes). The access and the cost of some techniques may impede the ability to make an accurate diagnosis and subsequently genetic counseling. A major challenge for genetic counseling in the developing countries involves dealing with this limitation¹. Experts in the field of genetics and genetic counseling should discuss how to deal with this limitation and put forth guidelines. The community of geneticists should simultaneously work towards developing affordable diagnostic assays. Initially, the focus could be on disorders with a relatively high incidence (for example, thalassemia, sickle cell anemia and Duchenne muscular dystrophy). With concerted effort the development of affordable diagnostic testing of genetic disorders is feasible and should be pursued.

Estimating risk is an important component of genetic counseling. For recessive disorders, if the laboratory techniques are not available to identify a carrier, carrier frequency in the population needs to be used for estimating the risk. The availability and the access to a reliable population frequency of the abnormal allele is vital in providing accurate risk figures. Collecting the required data to establish population frequency of the abnormal allele for disorders that would benefit from this information is important. Compiling data on the distribution of disorders (and mutations) in specific communities would be

valuable towards this goal. An example of this is the use of the frequency of Tay-Sachs, in the Ashkenazi Jewish community, in genetic counseling. In the early days of clinical genetics, when mutation analysis was not feasible, the empirical population carrier frequency of Tay-Sachs was used in estimating the risk and was used in genetic counseling.

Genetic counseling involves educating families on resources available for management. Often a family at risk for a genetic disorder is identified through an affected proband. The availability of affordable options for the management is essential, especially for these families with an affected proband. Strategies for the management of genetic disorders vary. Many disorders, like DiGeorge syndrome, require surgical intervention. Inborn errors of the metabolism, like Phenylketonuria, require dietary restriction or special diets. For cases of thalassemia, the treatment options are long-term transfusion therapy and hematopoietic stem cell transplantation. The availability of resources for optimal management may either not be readily available or may be expensive. Options for management should be developed as per the disorder and guidelines for the management should be readily available to the genetics community, including the genetic counselors.

Patient support groups are an invaluable source of help for the patients with genetic abnormalities and family members. Such groups are also a valuable source for genetic counselors for referring patients and families. Cataloguing available resources as well as helping to organize and expand such groups would go a long way in helping patients and families.

Despite these limitations, there are numerous opportunities for valuable genetic counseling in the developing countries. Genetic counseling includes "helping patients and families understand the medical implications" following the diagnosis of a genetic disorder. Educating patients and the family on the implications of a cytogenetic abnormality or a well characterized single gene disorder (medical needs, recurrence risk for the immediate family and extended family members, diagnostic and reproductive options etc.) is one example.

CONCLUSION

Genetic counseling services can be effectively delivered for some genetic disorders in the developing countries with facilities that are cur-

rently available. Here are however, a number of additional steps that need to be taken to expand the role of genetic counseling to serve a larger group of patients and families with genetic disorders. These include (1) incorporation of genetics in the physician training curriculum, (2) training sufficient numbers of clinical geneticists, (3) training genetic counselors (with the emphasis on local training programs to address the issues of social and cultural norms and language), (4) developing affordable diagnostic capabilities, (5) developing databases for deriving allele frequencies (6) developing manage-

ment options for genetic disorders and (7) organizing support groups.

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