

Genetic Counseling and Healthcare

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Letter to the Editor

Especially in this century it is provided to the patient clear information that is effective in both the diagnostic approach and the management of the disease with increase in the data infrastructure of diseases in the world. Latest developments in genetic testing play a huge role in preparing this ground. The importance of genetic counseling has understood better than past with the rapid developments in genetics science. In recent years the next generation sequences have provided many benefits in genetic diagnosis. It is a great progress for more clear diagnosis. We have more information about diagnosis of the disease, frequency of the disease, and distribution of the disease due to the international genetic databases offered to clinicians for using.

First of all, it is important to take a detailed anamnesis from the individuals before the genetic test and made a pedigree analysis. This allows to evaluate both identify the disease, seeing its distribution in the family (if any) and the options of necessary applying testing or unnecessary do not applying testing. The existence of many new variants can be detected with the next generation sequences. The results obtained by the next generation sequencing are classified as benign, likely benign, VUS, likely pathogenic and pathogenic variants.

In management of disease areas where a multidisciplinary approach is required that genetic counseling is indispensable. Genetic counseling is a vital aspect of healthcare. The different management is provided in carriers that have pathogenic/ likely pathogenic variants about the possible disease situation. In this situation, other than pathogenic or non-pathogenic variants, the changes of unknown clinical significance, are named as VUS (Variants of Uncertain Significance), are very important for the genetic counseling. This change in VUS carriers should be checked by using databases in every 6-12 months. It is extremely important whether there is a change in the variant classification for potential disease management and this must be emphasized.

The important point is avoid from unnecessary invasive proceeding in this group of patients and not to give up control of the patients, the consideration to keep in mind that this change may pass to a likely pathogenic or a pathogenic form.

It is the planning of the health management of the patient and family members, which is carried out under the responsibility of the genetic counselor. The process is very important that not only for the surviving individuals, but also for the birth of healthy generations in the field of reproductive medicine.

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