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Research Highlight

Exome Sequencing (ES): An Efficient Method to Sequence the Genomic Coding Regions

Fakher Rahim
Hearing Research Center,
Health Research Institute,
Ahvaz Jundishapur University of Medical Sciences,
Ahvaz, Iran Tel: +986113367562

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Exome sequencing: ES is an efficient and cost-effective technique to sequence the genomic coding regions.

During the last few years, the utilization of ES has considerably improved our knowledge regarding several complex diseases[1]. ES is a novel tool in medical research as well as clinical practice with a reduced cost that enables personalized medicine to come to maturity[2].

Inborn errors of metabolism: IEM are a genetically heterogeneous group of diseases that can cause production or buildup of toxic metabolites in the body. The IEMs are found in all ethnic groups and across every age.

ES is an economical and efficacious method which plays a key role in the detection of above 30 different IEMs in one single blood spot specimen with commendable analytical precision. Also, it guarantees systematic healthcare service delivery across the pre-analytical, analytical as well as post-analytical phases.

Accordingly, scientists conducted a review to examine all the accessible literature and to establish if the existing data supports any polymorphism to be conclusively linked with IEM[3].

For this purpose, the research team searched PubMed by employing MeSH terms such as, "Metabolism, Inborn Errors" as well as "Exome" and all other possible combinations. The search returned fifty-four unique articles of which nineteen articles met the established inclusion criteria and were included in the examination[3].

This review revealed that ES can assist to recognize novel mutations that may contribute to IEM pathogenesis. Furthermore, ES studies using multistage design might be more useful to study the role of genetic components in complex ailments such as IEM. However, knowledge of complicated mechanisms of genetic pathways involved in IEM etiology and pathogenesis would be supportive to identify subjects at high risk in a better way. Moreover, based on complex etiology, it is extremely uncertain that any single SNP contributes considerably to the development of IEM.

Conclusively, exome sequencing is a promising method to identify new

mutations that contribute in a significant way to IEM pathogenesis. However, scientists suggested performing detailed investigations that focus on other low penetrance polymorphisms by employing more comprehensive techniques including ES to identify potential genetic variations.

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