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Angiotensin converting enzyme (I/D) analysis and congenital heart diseases in Pakistani population

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Angiotensin converting enzyme (ACE) is a functional enzyme in renin-angiotensin system (RAS) during heart regulation. It is a 21-kb long gene which encodes an enzyme, on long arm of chromosome 17. ACE gene comprises of 26 exons with 25 introns. Genomic studies found that on ACE intron 16, an insertion or a deletion of a 287-bp noncoding Alu repeat sequence effects enzyme activity. This insertion/deletion polymorphism have a significant role in cardiovascular diseases, diabetes chronic obstructive pulmonary disease and skin disorders, furthermore, it was reported that D allele has shown an association with elevated levels of plasma ACE. Congenital heart diseases (CHDs) are the structural, functional and multifactorial defects, present at birth. Chromosomal aberration, alteration of gene (InDel, SNP) and environmental factors are some of the known causative agents of CHDs. However, to date there is limited information on the association of InDel polymorphism of ACE gene in Pakistani population. Total 207 patients and 150 healthy controls were recruited in the study to test the hypothesis. The aim of the current research is to find the association of D allele of ACE insertion/deletion polymorphism with congenital heart disease. The allelic frequencies of patients are D allele 56.7% and I allele 43.2% in patients and D allele 76.6% and I allele 23.3% in controls, reveals that presence of D allele can be a factor for the disturbed levels of enzyme in patients. It is needs to be further exploring its consequences.

Biography

Afsheen Arif has an expertise in rare genetic disorders. She has done her PhD on congenital heart disorders and their genetics in Pakistan for the first time. The unique genetic makeup of the patients from our population should have personalized methods of treatment. She has fifteen publication and five students currently working in her lab. Her future aims are to develop the genetic diagnostic setup for these patients.

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