GATA 4 sequence variation in Egyptian children with congenital heart disease

Mashaly M.H, Omran S., Sharaf E., Shaker O.

Cairo university, Cairo, Egypt

Introduction:
GATA 4 is a transcription factor that is expressed in the heart and is essential for cardiac development. Recent studies show relation between GATA 4 sequence variation and congenital heart disease.

The aim of this work is to investigate relation between GATA 4 sequence variation and congenital heart disease (CHD) among Egyptian children.

Methods:
A total of 48 cases 38 cases of CHD of different types and 10 control cases were included in our study. Exon 1 of GATA 4 was investigated for sequence variants using denaturing high-performance liquid chromatography or conformation-sensitive gel electrophoresis. Samples showing peak or band shifts were reamplified from genomic DNA and sequenced.

Results:
Novel mutation in Exon 1 in GATA 4 was found in 5 cases of CHD 13% (3 cases of Isolated Ventricular septal defect and 2 cases of Ventricular septal defect with atrial septal defect). Mutation was found in position 193 in exon 1 with base substitution (A) instead of (C) resulting in Amino acid change Histidine instead of Proline.

Conclusion:
There is a significant correlation between Ventricular septal defect and GATA 4 sequence in Egyptian Children. We need to further investigate this relation by having larger scale of cases and focusing on Ventricular septal defect.