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Abstract Topic: - Molecular effects of genetic variation

Abstract Title: - Single Nucleotide Polymorphisms (SNPs) and Copy Number Variations (CNVs) associated with Fc Gamma Receptors (FcγRs) in patients with Kawasaki Disease

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Aims: - The current study was designed to understand functionally relevant genetic variants in the FCGR2/3 locus in Indian patients with Kawasaki Disease.

Methods: - Children with KD and healthy controls were enrolled. Evaluation of SNPs and CNVs was done using Multiplex Ligation-dependent Probe Amplification (MLPA) assay.

Results: - 45 patients with KD were enrolled. Deletion in Copy Number Regions (CNRs) : (CNR1) (FCGR2C, HSPA7, FCGR3B); CNR2 (FCGR2A, HSPA6, FCGR3A, FCGR2C) and CNR4 (FCGR2C, HSPA7, FCGR3B, FCGR2B) were found in 10 patients. Functional SNPs [rs201218628; rs1801274; rs1050501] were also found in these patients.

A patient with acute KD with CNVs in CNR1 and CNR2 region also had reduced expression of these receptors on flow cytometry.

Conclusions: - Patients with KD have functionally relevant CNVs/SNPs in FcγRs. These changes may have therapeutic implications. Additionally, it help stratify an early response to IVIg thus classifying patient as IVIG responder or IVIg resistant KD and preventing cardiac complications in these patients.

Keywords: - 45 patients with KD were enrolled. Deletion in Copy Number Regions (CNRs) : (CNR1) (FCGR2C, HSPA7, FCGR3B); CNR2 (FCGR2A, HSPA6, FCGR3A, FCGR2C) and CNR4 (FCGR2C, HSPA7, FCGR3B, FCGR2B) were found in 10 patients. Functional SNPs [rs201218628; rs1801274; rs1050501] were also found in these patients.

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