

## **TNF-receptor associated Periodic Fever Syndrome and Response to Interleukin 1 Antagonist Therapy**

Reem Ali Husain AlDoseri, MB, BCH, BAO\* Maryam Busehail, MBBCh\*\* Barrak Mahmood Al Moosawi, MB, BCh BAO\*\*\* Zakiya Saleh Adnan Al Mosawi, MBBCH, ABMS-Pediatric\*\*\*\*

**Tumor necrosis factor receptor-associated periodic syndrome (TRAPS) is an autosomal dominant disease caused by mutations in the gene encoding the Tumor Necrosis Factor (TNF) Receptor Super Family1A (TNFRSF1A) on chromosome 12p13. TRAPS is characterized by recurrent attacks of fever, abdominal pain, migratory rash, and myalgia. Despite several hypotheses explaining the pathogenesis of TRAPS, the exact etiology remains obscure.**

**We report a female child who presented with clinical manifestations suggestive of macrophages activating syndrome (MAS) in addition to features of TRAPS syndrome. Next-generation sequencing was used to assess the genomic DNA of the patient and heterozygous c.362G>A p. (Arg121Gln) a variant in the TNFRSF1A gene (chr.12); an autosomal dominant inheritance was identified. After being treated for MAS manifestations, the patient was initiated on anti-inflammatory drugs including TNF-receptor antagonist with partial clinical response. However, the patient had a significant clinical response to interleukin 1 $\beta$  (IL-1 $\beta$ ) therapy and normalization of the inflammatory markers. Our findings suggest that IL-1 $\beta$  antagonist is a more effective alternative treatment for TRAPS complicated with MAS.**