Saudi Medical Journal
Volume ٢٩, Issue ١٠, ٢٠٠٨, Pages ١٤٩٤-١٤٩٦

# Rare gross deletion in T-cell immune regulator-١ gene in Iranian family with infantile malignant osteopetrosis

Abbaszadegan, M.R.[a] , Modarresi, A.[b], Khadivi-Zand, F.[c], Velayati, A.[a]
[a] Division of Human Genetics, Immunology Research Center, **Mashhad University** of **Medical Sciences**, Bu-Ali Square, **Mashhad**, Iran
[b] Pediatric Internal Division, Dr. Sheikh Hospital, **Mashhad Medical** Genetic Counseling Center, **Mashhad**, Iran
[c] Dr. Sheikh Hospital, **Mashad Medical** Genetic Counseling Center, **Mashbad**, Iran

View references (٧)

## Abstract

Infantile malignant osteopetrosis (arOP) is an autosomal recessive disorder. Mutations in the T-cell immune regulator ١ (TCIRG١) gene were found as the cause of arOP. We found the first Iranian patient with a rare gross deletion in this gene. The patient was a ٥-year-old girl with macrocephaly, facial dysmorphism, blindness, mental retardation, hepatosplenomegaly, pancytopenia, and osteosclerotic changes in the skull and limb. Molecular analysis was performed using reverse transcriptase-polymerase chain reaction for exons ١٠-١٩ of the TCIRG١ gene followed by whole gene sequencing. She showed a ٢٧٥bp unexpected amplified segment. Sequencing revealed a gross deletion in exons ١٠-١٥ transcript region of TCIRG١ that affected codon ٣٨٩ to ٥١٨. Various types of mutations in the TCIRG١ gene in arOP have been reported, however, gross deletions are reported rarely. This gross deletion is the first mutation reported among Iranian patients in this gene. This deletion is also the largest deletion of TCIRG١ gene reported to date.

## Reaxys Database Information

|

## Indexed Keywords

**EMTREE drug terms:** chromosome protein; T cell immune regulator ١ protein; unclassified drug

**EMTREE medical terms:** amino acid sequence; article; autosomal recessive disorder; blindness; case report; chromosome ١١q; codon; exon; exophthalmos; face dysmorphia; female; gene deletion; gene sequence; hepatosplenomegaly; human; infantile malignant osteopetrosis; Iran; macrocephaly; mental deficiency; nucleotide sequence; osteosclerosis; pancytopenia; pedigree analysis; preschool child; reverse transcription polymerase chain reaction; sequence analysis

**Molecular Sequence Numbers:** GENBANK,CG٠٤٤٥٩(referenced), U٤٥٢٨٥(referenced); SWISSPROT,Q١٣٤٨٨(referenced)

**ISSN:** ٠٣٧٩٥٢٨٤ **CODEN:** SAMJD**Source Type:** Journal **Original language:** English
**Document Type:** Article