# TMEM142 deficiency

## GENERAL INFORMATION

### Description:

Defects in TMEM142a are a cause of severe combined immunodeficiency (SCID) with defects in store-operated Ca(2+) entry and CRAC channel function [mim:610277]. The SCID T-cells show a severe impairment in NFAT-dependent gene activation.

### Alternative names:

- Orai1
- Autosomal recessive severe combined immunodeficiency, with CRAC channel dysfunction

### Classification:

- Combined B and T cell immunodeficiencies
  - Other

### Inheritance:

Autosomal recessive

### OMIM:

- *610277 Transmembrane protein 142A

### Cross references:

**Phenotype related immunodeficiencies:**

- ZAP-70 deficiency

### Incidence:

Incidence is not known yet.

## CLINICAL INFORMATION

### Description:

Patients have marked propensity for fungal and viral infections. Myopathy, retardation, ectodermal dysplasia in one patient.

### Diagnosis:

### Diagnostic laboratories:

**Clinical:**

- Severe Combined Immunodeficiency, eMedicine

**Genetic:**

- IDdiagnostics

### Therapeutic options:

- Bone marrow transplantation is the only treatment of SCID. Other recommendations include intravenous gamma-globulin infusion, irradiation of all blood products, antibiotherapy.

### Research programs, clinical trials:

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## GENE INFORMATION

### Names:

**HUGO name:** ORAI1
**Alias(es):** TMEM142, CRACM1, FLJ14466

## Localization:

**Reference sequences:**

**DNA:** IDRefSeq: D0114 (IDRefSeq) , **cDNA:** BC013386 (EMBL) , **Protein:** Q96D31 (SWISSPROT)  Other Sequences

**Chromosomal Location:**

12q24.31

**Maps:**

TMEM142 (Map View)

## Variations / Mutations:

- ORAI1base; Mutation registry for TMEM142 deficiency

## Other gene-based resources:

Ensembl: ENSG00000182500, GENATLAS: TMEM142, GeneCard: TMEM142, UniGene: 55148, Entrez Gene: 84876, euGenes: Hugn84876, GDB: , HomoloGene: 13117

# PROTEIN INFORMATION

## Description:

**Protein function:**

Ca(2+) release-activated Ca(2+) (crac) channel subunit which mediates Ca(2+) influx following depletion of intracellular Ca(2+) stores and channel activation by the Ca(2+) sensor, stim1. Crac channels are the main pathway for Ca(2+) influx in T-cells and promote the immune response to pathogens by activating the transcription factor NFAT.

**Subcellular location:**

Cell membrane; multi-pass membrane protein

**Miscellaneous:**

In greek mythology, the 'orai' are the keepers of the gates of heaven: eunomia (order or harmony), dike (justice) and eirene (peace)

**Similarity:**

Belongs to the Orai family

## Other features:

**Calcium release-activated calcium channel protein 1: 1-301**

**N-linked (glcnac...) glycosylation sites:** 223

**Other related resourses:**

InterPro: IPR012446; DUF1650, Pfam: PF07856; DUF1650

## Expression pattern for human:

| Tissue | Exp. (%) | Clones |
|---|---|---|

# OTHER RESOURCES

## Societies:

**General:**

- European Society for Immunodeficiencies
- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- March of Dimes Birth Defects Foundation
- NIH/National Institute of Allergy and Infectious Diseases

## Other information sources:

- Severe Combined Immunodeficiency, Patient and Family Handbook
- Severe Combined Immunodeficiency, KidsHealth
- ORAI1 deficiency