

CD3Zeta deficiency

GENERAL INFORMATION

Description:

CD3zeta is necessary for the development and function of T cells.

Alternative names:

- CD3Z
- Cd3 antigen, zeta subunit
- Cd3-zeta
- T-cell antigen receptor complex, zeta subunit of cd3; TCRZ
- CD3 complex components

Classification:

- Combined B and T Cell Immunodeficiencies
 - CD3 deficiency

Inheritance:

Autosomal recessive

OMIM:

- *186780 Cd247 antigen; CD247

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for CD3D deficiency
- IDR factfile for CD3E deficiency
- IDR factfile for CD3G deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Erythroderma, protracted diarrhea, and pulmonary abscesses caused by *Pseudomonas aeruginosa*, recurrent episodes of herpes simplex virus infection of the mouth and skin, two episodes of oral and skin infections with *Candida albicans*, and two pulmonary infections, one of which was caused by *Streptococcus pneumoniae*.

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: CD247

Alias(es): CD3Z, T3Z, TCRZ

Localization:

Reference sequences:

DNA: D0115 (IDRefSeq) , **cDNA:** J04132 (EMBL) , **Protein:** P20963 (SWISSPROT)
Other Sequences

Chromosomal Location:

1q22-q23

Maps:

CD247 (Map View)

Variations / Mutations:

- CD247base; Mutation registry for CD3Zeta deficiency

Other gene-based resources:

Ensembl: ENSG00000198821, GENATLAS: CD247, GeneCard: CD247, UniGene: 156445, Entrez Gene: 919, euGenes: 0000919, GDB: CD3Z, HomoloGene: 12818

PROTEIN INFORMATION

Description:

Protein function:

Probable role in assembly and expression of the TCR complex as well as signal transduction upon antigen triggering.

Subunit:

The TCR/CD3 complex of T-lymphocytes consists of either a TCR alpha/beta or TCR gamma/delta heterodimer coexpressed at the cell surface with the invariant subunits of CD3 labeled gamma, delta, epsilon, zeta, and eta. Cd3-zeta forms either homodimers or heterodimers with CD3-eta. Interacts with Sla and Sla2. Interacts with Dock2 and Trat1. Interacts with HIV-1 Nef protein. Interacts with SHB.

Subcellular location:

Membrane; single-pass type i membrane protein.

Post-translational modification:

Phosphorylated on Tyr residues after T-cell receptor triggering (by similarity).

Protein function:

Event=alternative splicing; named isoforms=2; name=cd-3-zeta; isoid=p20963-1; sequence=displayed; name=cd-3-eta; isoid=p20963-2; sequence=not described;

Similarity:

Belongs to the CD3Z/fcer1g family.

Structures (PDB):

1TCE .

Domains:

ITAM 1 domain: 61-89

ITAM 2 domain: 100-128

ITAM 3 domain: 131-159

Other features:

Signal peptide: 1-21

T-cell surface glycoprotein cd3 zeta chain:
22-164

Disulfide bond interchain: 32

Other related resources:

Uniprot: CD3Z_HUMAN, InterPro: IPR003110;
Phos_rcpt_ITAM, Pfam: PF02189; ITAM,
SMART: SM00077; ITAM, PROSITE:
PS51055; ITAM_1

Expression pattern for human:

Tissue	Exp. (%)	Clones
thymus	56.12	43:68776
spleen	11.38	6:47318
blood	4.38	3:61437
uncharacterized_tissue	4.16	10:215582
mixed	3.11	10:288723
heart	2.61	2:68859
muscle	2.50	3:107777
prostate	2.25	3:119494
lymph_node	2.16	2:83039
embryonic_tissue	1.97	1:45612

Animal models:**Mouse:**

MGD: ; Cd247

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:

- CD3Zeta deficiency