CD3ε deficiency

GENERAL INFORMATION

Description:

CD3ε deficiency is an autosomal recessive immunodeficiency caused by mutations in the gene coding for T-cell surface glycoprotein CD3ε (epsilon) chain precursors. Patients with CD3ε deficiency have a severe defect in the expression of the T-cell receptor CD3-complex.

Alternative names:

- CD3E deficiency
- CD3 complex component

Classification:

- Combined B and T cell immunodeficiencies
  - CD3 deficiency

Inheritance:

Autosomal recessive

OMIM:

- *186830 CD3 antigen, epsilon subunit; CD3E

Cross references:

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have recurrent Hemophilus influenzae pneumonia and otitis media over 6-month period at 2 years of age. CD3 deficiency present as mild combined immunodeficiency, with later onset than SCID. No autoimmune phenomena were detected.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Severe combined immunodeficiency, eMedicine

Genetic:

- CD3G, IDdiagnostics

Therapeutic options:

- Patients respond well to antibiotic treatment and it is no need for bone marrow transplant.
- T-cell disorders, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies 2001-2004

GENE INFORMATION

Names:

HUGO name: CD3E
Alias(es): T3E, TCRE, CD3E antigen, epsilon polypeptide (TiT3 complex), T-cell surface glycoprotein CD3 epsilon chain precursor, T-cell surface antigen T3/LEU-4 epsilon chain

Localization:

Reference sequences:

DNA: M23317 (EMBL) , **cDNA:** X03884 (EMBL) , **Protein:** P07766 (SWISSPROT) Other Sequences

Chromosomal Location:

11q23

Maps:

CD3E (Map View)

Variations / Mutations:

- CD3Ebase; Mutation registry for autosomal recessive CD3epsilon deficiency

Other gene-based resources:

Ensembl: ENSG00000198851, GENATLAS: CD3E, GeneCard: CD3E, UniGene: 3003, Entrez Gene: 916, euGenes: 916, GDB: 119764

PROTEIN INFORMATION

Description:

Protein function:

The CD3 complex mediates signal transduction.

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.

Subcellular location:

Type I membrane protein.

Domains:

Extracellular domain: 23-126

Cytoplasmic domain: 153-207

Other features:

Signal peptide: 1-22

T-cell surface glycoprotein CD3 epsilon chain: 23-207

Other related resourses:

InterPro: IPR003110; ITAM, InterPro: IPR003598; Ig_c2, Pfam: PF02189; ITAM, SMART: SM00408; IGc2, SMART: SM00077; ITAM

Expression pattern for human:

Tissue	Exp. (%)	Clones
T cells from T cell leukemia	23.42	3:2397
blood, white cells	20.56	1:910
lymph, T-cell	13.20	6:8503
leukocyte	12.50	6:8982
thymus, pooled	11.81	2:3169
spleen	5.18	2:7229
blood	2.96	2:12646
germ cell	2.80	3:20077
mixed	2.17	7:60341
rpe and choroid	1.77	1:10565

Animal models:

Mouse:

MGD: ; Cd3e

Fly:

euGenes: ; emp

C. elegans:

euGenes: ; Y49E10.20

OTHER RESOURCES

Societies:

General:

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