CD3γ deficiency

GENERAL INFORMATION

Description:

CD3γ deficiency is an autosomal recessive immunodeficiency caused by mutations in the gene coding for T-cell surface glycoprotein CD3γ (gamma) chain precursors. Patients with CD3γ deficiency have a severe defect in the expression of the T-cell receptor CD3-complex. Affected patients have decreased T-cell numbers and function; B cells are variably affected.

Alternative names:

- CD3-gamma deficiency
- CD3 complex components

Classification:

- Combined B and T cell immunodeficiencies
  - CD3 deficiency

Inheritance:

Autosomal recessive

OMIM:

- *186740 CD3 antigen, gamma subunit; CD3G

Cross references:

Incidence:

Incidence unknown.

CLINICAL INFORMATION

Description:

Patients with CD3# deficiency and CD3# deficiency suffer recurrent bacterial and viral infections. Age of onset, when symptoms are present, is early in life. In advanced age, patients experience less infection. They had no history of chronic pyogenic or fungal infections, eczema, or chronic moniliasis. Patients have mild immunodeficiencies.

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: CD3G
Alias(es): T3G, CD3G antigen, gamma polypeptide (TiT3 complex), T-cell surface glycoprotein CD3 gamma chain precursor, T-cell receptor T3 gamma chain

Localization:

Reference sequences:

DNA: M23317 (EMBL) , **cDNA:** X04145 (EMBL) , **Protein:** P09693 (SWISSPROT) Other Sequences

Chromosomal Location:

11q23

Maps:

CD3G (Map View)

Variations / Mutations:

- CD3Gbase; Mutation registry for autosomal recessive CD3gamma deficiency

Other gene-based resources:

Ensembl: ENSG00000160654, GENATLAS: CD3G, GeneCard: CD3G, UniGene: 2259, Entrez Gene: 917, euGenes: 917, GDB: 119765

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: 23-111

Cytoplasmic domain: 139-182

Other features:

Signal peptide: 1-22

T-cell surface glycoprotein CD3 gamma chain: 23-182

Other related resourses:

PIR: A25468, InterPro: IPR003110; ITAM, InterPro: IPR003598; Ig_c2, Pfam: PF02189; ITAM, SMART: SM00408; IGc2, SMART: SM00077; ITAM

Expression pattern for human:

Tissue	Exp. (%)	Clones
blood, white cells	56.95	1:910
mammary gland	35.97	1:1441
pool, liver+spleen	4.23	5:61327
pool, lung+testis+B-cell	1.86	2:55714
lymph	0.80	1:64395
brain	0.19	1:274929

Animal models:

Mouse:

MGD: ; Cd3g

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