CD3delta deficiency

GENERAL INFORMATION

Description:

CD3delta deficiency is characterized by the absence of T cells but normal numbers of B cells. CD3D is essential for T cell development.

Alternative names:

- SCID with CD3D chain-deficiency, CD3 complex components

Classification:

- Combined B and T cell immunodeficiencies
  - T⁻B⁺ SCID

Inheritance:

Autosomal recessive

OMIM:

- #600802 Severe combined Immunodeficiency, autosomal recessive, T-negative/B-positive type
- *186790 CD3 antigen, delta subunit; CD3D

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for CD3ε deficiency
- IDR factfile for CD3γ deficiency

Incidence:

Incidence it is not known until now.

CLINICAL INFORMATION

Description:

The clinical symptoms are similar with those for SCID: failure to thrive, intractable diarrhea, and recurrent lung infection. The patients had an early arrest in T cell development, with a nearly complete absence of circulating mature T cells and a complete absence of gamma/delta T cells.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Severe combined immunodeficiency, eMedicine

Genetic:

- IDdiagnostics

Therapeutic options:

- Treatment of infections with antibacterials, antifungals, and antivirals. Bone marrow transplantation is the only treatment of SCID. Other recommendations include: intravenous gamma-globulin infusion, irradiation of all blood products.
- Severe combined immunodeficiency, eMedicine

Research programs, clinical trials:

- Pilot Study of Allogeneic Bone Marrow Transplantation Plus Cyclosporine and Mycophenolate Mofetil to Induce Mixed Hematopoietic Chimerism in Patients With Primary T-Cell Immunodeficiency Disorders, ClinicalTrial.gov
- European Initiative for Primary Immunodeficiencies 2001-2004, coord.Edvard Smith.

GENE INFORMATION

Names:

HUGO name: CD3D
Alias(es): CD3-DELTA, T3D, CD3D antigen, delta polypeptide (TiT3 complex), T-cell surface glycoprotein CD3 delta chain precursor

Localization:

Reference sequences:

DNA: X03934 (EMBL) M12727 (EMBL) X01451 (EMBL) , **cDNA:** X58957 (EMBL) , **Protein:** P04234 (SWISSPROT) Other Sequences

Chromosomal Location:

11q23

Maps:

CD3D (Map View)

Markers:

CD3D, RH93888

Variations / Mutations:

- CD3Dbase; Mutation registry for CD3delta deficiency

Other gene-based resources:

Ensembl: ENSG00000167286, GENATLAS: CD3D, GeneCard: CD3D, UniGene: 504048, LocusLink: 915, euGenes: 915, GDB: 120578, HomoloGene: 585

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

Similarity:

Contains 1 ITAM domain

Domains:

Extracellular domain: 22-100

Cytoplasmic domain: 128-171

ITAM domain: 146-166

Other features:

Signal peptide: 1-21

T-cell surface glycoprotein cd3 delta chain: 22-171

N-linked (glcnac...) glycosylation sites: 38,74

Other related resourses:

PIR: RWHUD1, InterPro: IPR003110; ITAM, Pfam: PF02189; ITAM, SMART: SM00077; ITAM

Expression pattern for human:

Tissue	Exp. (%)	Clones
thymus	37.03	2:2187
pooled	28.41	4:5702
normal prostate	10.08	2:8035
leukocyte	9.16	2:8838
2 pooled tumors (clear cell type)	6.32	3:19218
fetal eyes, lens, eye anterior segment, optic nerve, retina, retina foveal and macular, RPE and choroid	2.35	1:17221
lymph	2.07	1:19564
carcinoid	1.66	2:48801
pooled human melanocyte, fetal heart, and pregnant uterus	1.25	1:32508
unclassified	1.20	26:879462
thymus	37.03	2:2187
pooled	28.41	4:5702
normal prostate	10.08	2:8035
leukocyte	9.16	2:8838
2 pooled tumors (clear cell type)	6.32	3:19218
fetal eyes, lens, eye anterior segment, optic nerve, retina, retina foveal and macular, RPE and choroid	2.35	1:17221
lymph	2.07	1:19564
carcinoid	1.66	2:48801
pooled human melanocyte, fetal heart, and pregnant uterus	1.25	1:32508
unclassified	1.20	26:879462

Animal models:

Mouse:

MGD: ; Cd3d, NCBI Gene: ; 12500 (64.33 % aminoacid similarity to human)

Rat:

NCBI Gene: ; 25710 (69.01 % aminoacid similarity to human)

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies (IPOPI)
- Immune Deficiency Foundation
- European Society for Immunodeficiencies