CD45 deficiency

GENERAL INFORMATION

Description:

CD45 deficiency is an autosomal recessive disease with T and B lymphocyte dysfunction, due to a large deletion at one allelle and a point mutation at the other. The point mutation resulted in the alteration of intervening sequence 13 donor splice site.The population of T lymphocytes is diminished and unresponsive to mitogen stimulation. The level of B lymphocyte numbers, serum immunoglobulin decreased with age.

Alternative names:

- CD45 Ly5, Human homolog of B220 Severe combined immunodeficiency due to PTPRC Deficiency
- CD45

Classification:

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

Inheritance:

Autosomal recessive

OMIM:

- #202500 Severe Combined Immunodeficiency 1; SCID1
- +151460 Protein-tyrosine phosphatase, receptor-type, c; PTPRC

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for X-linked SCID(gamma-chain deficiency)
- IDR factfile for JAK3 deficiency

Incidence:

Incidence it is not known yet.

CLINICAL INFORMATION

Description:

Patients with SCID usually develop failure to thrive and persistent diarrhea, respiratory symptoms and/or thrush in the first 2 to 7 months of life. Pneumocystis pneumonia, significant bacterial infections and disseminated BCG infection are common presenting illnesses. Occasional patients do not have failure to thrive and are not recognized to have immunodeficiency until late in the first year of life. SCID is fatal in the first 2 years of life unless the patient is treated with extremely restrictive isolation, hematopoietic stem cell transplant or therapy that replaces the abnormal gene or gene product.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Severe Combined Immunodeficiency, eMedicine

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.
- National Marrow Donor Program
- 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

GENE INFORMATION

Names:

HUGO name: PTPRC
Alias(es): CD45, GP180, LCA, T200, protein tyrosine phosphatase, receptor type, C, protein tyrosine phosphatase, receptor type, c polypeptide, Leukocyte common antigen precursor, CD45 antigen

Localization:

Reference sequences:

DNA: PTPRC_DNA (IDRefSeq) , **cDNA:** Y00638 (EMBL) , **Protein:** P08575 (SWISSPROT)

Chromosomal Location:

1q31-q32

Maps:

PTPRC (Map View), RH142448, D1Mit514, STS-T93530, RH70956

Variations / Mutations:

- PTPRCbase; Mutation registry for CD45 deficiency

Other gene-based resources:

Ensembl: ENSG00000081237, GENATLAS: PTPRC, GeneCard: PTPRC, UniGene: 192039, Entrez Gene: 5788, euGenes: 5788, GDB: 119768

PROTEIN INFORMATION

Description:

Protein function:

Required for T-cells activation through the antigen receptor. The first PTPase domain has enzymatic activity, while the second one seems to affect the substrate specificity of the first one.

Catalytic activity:

Protein tyrosine phosphate + H(2)o = protein tyrosine + phosphate

Subcellular location:

Type I membrane protein

Post-translational modification:

Heavily N- and O-glycosylated

Protein function:

At least 8 isoforms are produced by alternative splicing

Domains:

Extracellular domain: 24-575

Cytoplasmic domain: 598-1304

Fibronectin type-III 1 domain: 387-479

Fibronectin type-III 2 domain: 480-571

Protein-tyrosine phosphatase 1 domain: 670-919

Protein-tyrosine phosphatase 2 domain: 961-1235

Other features:

Signal peptide: 1-23

Leukocyte common antigen: 24-1304

Other related resourses:

InterPro: IPR003961; FN_III, InterPro: IPR000387; TYR_phosphatase, InterPro: IPR000242; Tyr_PP, Pfam: PF00041; fn3, Pfam: PF00102; Y_phosphatase, PROSITE: PS00383; TYR_PHOSPHATASE_1, PROSITE: PS50056; TYR_PHOSPHATASE_2, PROSITE: PS50055; TYR_PHOSPHATASE_PTP

Expression pattern for human:

Tissue	Exp. (%)	Clones
nasopharynx	12.00	1:646
adipose	9.13	1:849
blood, white cells	8.52	1:910
nose, olfactory epithelium	6.94	1:1116
lymph, T-cell	6.38	7:8503
subchondral bone	5.82	1:1332
lymph	5.17	43:64395
pheochromocytoma	4.97	1:1560
thymus, pooled	4.89	2:3169
chondrosarcoma	3.81	6:12201

Animal models:

Mouse:

MGD: ; PTPRC

Fly:

euGenes: ; Ptp52F

C. elegans:

euGenes: ; C02B10.6

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

Disease specific:

- The SCID Homepage

Other information sources:

- Severe Combined Immunodeficiency, Patient and Family Handbook
- Severe Combined Immuno-Deficiency (SCID), JMF
- Severe Combined Immunodeficiency, KidsHealth