IL-2 receptor α-chain deficiency (CD25 deficiency)

GENERAL INFORMATION

Description:

It is a severe combined immunodeficiency, with absence of CD1 and of IL2RA expression on cortical thymocyte. It is characterized by decreased numbers of peripheral T cells but normal B-cell development. Extensive lymphocytic infiltration of tissues, including lung, liver, gut, and bone accompanied by tissue atrophy and inflammation. The absence of CD25 affected the differentiation of thymocytes.

Alternative names:

- CD25 deficiency, IL2RA deficiency, IL-2 receptor alfa chain (CD25)

Classification:

- Other Combined B and T cell immunodeficiencies
  - Other

Inheritance:

Autosomal recessive

OMIM:

- #606367 Interleukin 2 receptor, alpha, deficiency of
- *147730 Interleukin 2 receptor, alpha; IL2RA

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

CD25 deficiency begins early in life with recurrent infections and dramatic lymphocytes infiltrates in multiple tissues. The patient had a history of increased susceptibility to viral, bacterial and fungal infections. At the age of 6 months he presented cytomegalovirus pneumonitis, persistant oral thrush, and Candida esophagitis. He had also adenovirus gastroenteritis, developed chronic diarrhea, and failed to thrive. From the age of 8 months he presented lymphadenopathy and hepatosplenomegaly. He had repeted episodes of lung disease and by the age of 3 years developed gingivitis, iron deficiency anemia.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Severe Combined Immunodeficiency, eMedicine

Therapeutic options:

- Intravenous gamma-globulin (IVIg) should be started early. Treatment of infections with antibacterials, antivirals and antifungals.
- Severe Combined Immunodeficiency, eMedicine
- National Marrow Donor Program

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies 2001-2004, coord.Edvard Smith.

GENE INFORMATION

Names:

HUGO name: IL2RA
Alias(es): IL2R, interleukin 2 receptor, alpha, Interleukin-2 receptor alpha chain precursor, IL-2 receptor alpha subunit, P55, TAC antigen, CD25 antigen

Localization:

Reference sequences:

DNA: IL2RA_DNA (EMBL) , **cDNA:** X01057 (EMBL) , **Protein:** P01589 (SWISSPROT) Other Sequences

Chromosomal Location:

10p15-p14

Maps:

IL2RA (Map View)

Variations / Mutations:

- IL2RAbase; Mutation registry for IL2RA deficiency

Other gene-based resources:

Ensembl: ENSG00000134460, GENATLAS: IL2RA, GeneCard: IL2RA, UniGene: 231367, Entrez Gene: 3559, euGenes: 3559, GDB: 119345

PROTEIN INFORMATION

Description:

Protein function:

Receptor for interleukin-2.

Subunit:

Non covalent dimer of an alpha and a beta chains. Il-2R exist in 3 different forms: a high affinity dimer, an intermediate affinity monomer (beta chain), and a low affinity monomer (alpha chain). The high and intermediate affinity forms also associate with a gamma chain.

Subcellular location:

Type I membrane protein.

Structures (PDB):

1ILM IL2RA
1ILN IL2RA

Domains:

Extracellular domain: 22-240

Cytoplasmic domain: 260-272

Sushi 1 domain: 23-81

Sushi 2 domain: 124-185

Other features:

Signal peptide: 1-21

Interleukin-2 receptor alpha chain: 22-272

Disulfide bonds: 24-67, 51-80, 125-168, 152-184

Other related resourses:

PIR: UHHU2, InterPro: IPR000436; Sushi_SCR_CCP, Pfam: PF00084; sushi, SMART: SM00032; CCP

Expression pattern for human:

Tissue	Exp. (%)	Clones
smooth muscle	46.59	1:647
blood, white cells	33.12	1:910
grade-2-chondrosarcoma	11.42	1:2639
adrenal gland	3.47	2:17391
leukocyte	3.36	1:8982
unclassified	1.16	2:51898
lymph	0.47	1:64395
brain	0.22	2:274929
lung	0.19	1:155782

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