Hepatic veno-oclussive disease with immunodeficiency syndrome

GENERAL INFORMATION

Description:

This syndrome is characterized by the association of severe hypogammaglobulinemia, combined T and B cell immunodeficiency, absent lymph node germinal centers, absent tissue plasma cells and hepatic veno-occlusive disease.

Alternative names:

- VODI
- Hepatic veno-occlusive disease-immunodeficiency

Classification:

- Other well-defined immunodeficiency syndromes

Inheritance:

Autosomal recessive

OMIM:

- *604457 Nuclear body protein sp110; SP110
- #235550 Hepatic venooclussive disease with immunodeficiency; VODI

Cross references:

Incidence:

1:2,500 live births in the Lebanese population of Sydney, Australia.

CLINICAL INFORMATION

Description:

Patients have Pneumocystis jerovici infection, enteroviral infection or mucocutaneous candidiasis. Hepatic vascular oclusion disease was verified by biopsy. B cell immunodeficiency include hypogamaglobulinemia, absent memory B cells and tonsilar lymph nodes. Absence of lymph node germinal centers and tissue plasma cells. T cell immunodeficiency include reduced number of memory T cells, and low or reduced intracellular T cell cytokine expression, IL2, IL4, and IL10.

Diagnosis:

Diagnostic laboratories:

Clinical:

- ORPHANET

Genetic:

- IDdiagnostics

Therapeutic options:

- Intravenous immunoglobulin and Pneumocystis jerovici prophylaxis.
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Research programs, clinical trials:

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GENE INFORMATION

Names:

HUGO name: SP110
Alias(es): FLJ22835, IFI41, IFI75, VODI

Localization:

Reference sequences:

DNA: D0116 (IDRefSeq) , **cDNA:** L22342 (EMBL) , **Protein:** NP_536349.1 (GenBank) Other Sequences

Chromosomal Location:

2q37.1

Maps:

SP110 (Map View)

Variations / Mutations:

- SP110base; Mutation registry for Hepatic veno-oclussive disease with immunodeficiency syndrome (VODI)

Other gene-based resources:

Ensembl: ENSG00000135899, GENATLAS: SP110, GeneCard: SP110, UniGene: 145150, Entrez Gene: 3431, euGenes: 3431, GDB: 266530, HomoloGene: 82192

PROTEIN INFORMATION

Description:

Protein function:

Transcription factor. May be a nuclear hormone receptor coactivator. Enhances transcription of genes with retinoic acid response elements (rare).

Subunit:

Isoform 3 interacts with HCV core protein.

Subcellular location:

Nucleus. Found in the nuclear body.

Post-translational modification:

Phosphorylated (isoform 2).

Protein function:

Event=alternative splicing; named isoforms=5; name=1; isoid=q9hb58-1; sequence=displayed; name=2; synonyms=ifi75, 75; isoid=q9hb58-2; sequence=vsp_005992, vsp_006001, vsp_006002; name=3; synonyms=sp110b; isoid=q9hb58-3; sequence=vsp_005998, vsp_005999; name=4; synonyms=ifi41, 41; isoid=q9hb58-4; sequence=vsp_005991, vsp_005994, vsp_005995, vsp_005997, vsp_006000; name=5; isoid=q9hb58-5; sequence=vsp_005993, vsp_005996, vsp_005997, vsp_006000;

Induction:

By interferon gamma and by all-trans retinoic acid.

Tissue specificity:

Highly expressed in peripheral blood leukocytes and spleen. Detected at intermediate levels in thymus, prostate, testis, ovary, small intestine and colon, and at low levels in heart, brain, placenta, lung, liver, skeletal muscle, kidney and pancreas.

Similarity:

Contains 1 bromo domain.

Domains:

HSR domain: 6-109

SAND domain: 454-535

Bromo domain: 581-676

Other features:

Sp110 nuclear body protein: 1-689

Zinc finger region Phd-type: 534-580

Other related resourses:

InterPro: IPR001487; Bromodomain, InterPro: IPR000770; SAND, InterPro: IPR010919; SAND_like, InterPro: IPR004865; Sp100, InterPro: IPR011011; Znf_FYVE_PHD, InterPro: IPR001965; Znf_PHD, Pfam: PF00628; PHD, Pfam: PF01342; SAND, Pfam: PF03172; Sp100, SMART: SM00297; BROMO, SMART: SM00249; PHD, SMART: SM00258; SAND, PROSITE: PS00633; BROMODOMAIN_1, PROSITE: PS50014; BROMODOMAIN_2, PROSITE: PS50864; SAND, PROSITE: PS01359; ZF_PHD_1, PROSITE: PS50016; ZF_PHD_2

Expression pattern for human:

Tissue	Exp. (%)	Clones
bone_marrow	13.36	15:41353
tonsil	10.22	5:18016
spleen	7.78	10:47318
lymph	7.59	9:43667
lymph_node	6.65	15:83039
blood	5.40	9:61437
ovary	4.53	10:81370
mixed	4.21	33:288723
thymus	3.21	6:68776
bone	3.14	6:70388

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

Other information sources:

- Hepatic veno-occlusive disease with immunodeficiency syndrome (VODI)