STAT5b deficiency

GENERAL INFORMATION

Description:

Laron syndrome is an autosomal recessive disorder characterized by marked short stature, clinical hyposomatotropism, failure to generate somatomedin, or insulin-like growth factor-1 in response to growth hormone, and normal or increased levels of growth hormone. Patients have occasionally blue sclerae and hip degeneration. Laron syndrome is caused by dysfunction of the growth hormone receptor. There are two types: type I is associated with defects in growth hormone receptor while the second, type II, is apparently due to postreceptor defects in signal transduction to produce insulin-like growth factor-1 or possibly defects in its receptor.

Alternative names:

- LTD2
- Growth hormone insensitivity with immunodeficiency
- Laron syndrome due to postreceptor defect

Classification:

- Defects of innate immune system, receptors and signaling components

Inheritance:

Autosomal recessive

OMIM:

- #245590 Laron syndrome, type II
- *604260 Signal transducer and activator of transcription 5b; STAT5B

Cross references:

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have clinical and biochemical characteristics of GH insensitivity dwarfism and combined phenotype of GH insensitivity, immunodeficiency consistent with the presence of a defect in the JAK/STAT system, dysmorphic features, eczema, lymphocytic interstitial pneumonitis and modestly decreased circulating T cells.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Nanism due to growth hormone resistance, ORPHANET, France
- Short stature, eMedicine, USA
- Laron syndrome, eMedicine, USA

Therapeutic options:

- Laron syndrome, eMedicine, USA
- Short stature, eMedicine, USA

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

GENE INFORMATION

Names:

HUGO name: STAT5B
Alias(es): STAT5, Signal transducer and activator of transcription 5B, Transcription factor STAT5B

Localization:

Reference sequences:

DNA: U48730 (EMBL) U47686 (EMBL) AJ412888 (EMBL) , **cDNA:** X58957 (EMBL) , **Protein:** P51692 (SWISSPROT) Other Sequences

Chromosomal Location:

17q11.2

Maps:

STAT5B (Map View)

Markers:

WI-11886, RH45469, STS-U48730, STS-T90203, SHGC-64296, D17S1673, RH36528, D17S1802, RH91495, D17S1143

Variations / Mutations:

- STAT5Bbase; Mutation registry for Growth hormone insensitivity with immunodeficiency

Other gene-based resources:

Ensembl: ENSG00000173757, GENATLAS: STAT5B, GeneCard: STAT5B, UniGene: 132864, Entrez Gene: 6777, euGenes: 6777, GDB: 5772854

PROTEIN INFORMATION

Description:

Protein function:

Carries out a dual function: signal transduction and activation of transcription. Binds to the GAS element and activates PRI-induced transcription.

Subunit:

Forms a homodimer or a heterodimer with a related family member. Binds NR3C1. Interacts with NMI.

Subcellular location:

Nuclear; translocated into the nucleus in response to phosphorylation.

Post-translational modification:

Tyrosine phosphorylated.

Similarity:

Belongs to the transcription factor STAT family.

Domains:

SH2 domain: 589-686

REQUIRED FOR INTERACTION WITH NMI domain: 232-321

Other features:

Other related resourses:

InterPro: IPR008967; P53-like, InterPro: IPR000980; SH2, InterPro: IPR001217; STAT, Pfam: PF00017; SH2, Pfam: PF01017; STAT, Pfam: PF02864; STAT_bind, Pfam: PF02865; STAT_prot, SMART: SM00252; SH2, PROSITE: PS50001; SH2

Expression pattern for human:

Tissue	Exp. (%)	Clones
Bulk germ cell seminoma	19.01	1:290
Normal endometrium, late proliferative phase, cycle day 13	5.35	1:1030
Lymph	5.07	18:19564
Thymus	5.04	2:2187
Lung Focal Fibrosis	4.23	3:3912
Testis	4.16	4:5295
Peripheral nervous system	3.55	1:1553
Germ cell tumor	2.58	1:2133
Schizophrenic brain S-11 frontal lobe	2.47	1:2229
Trabecular meshwork	2.06	1:2671
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Animal models:

Mouse:

MGD: ; Stat5b

OTHER RESOURCES

Societies:

General:

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