Tapasin deficiency

GENERAL INFORMATION

Description:

Bare lymphocyte syndrome type I or class I antigen deficiencies can be caused by mutations in the TAP2, TAP1 or TAPBP. The symptoms develop in late childhood and include chronic lung disease, bronchiectasis, emphysema, panbronchiolitis, and bronchial obstruction.

Alternative names:

- TPSN deficiency
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Classification:

- Combined B and T cell immunodeficiencies
  - Major histocompatibility complex class I deficiency

Inheritance:

Autosomal recessive

OMIM:

- #604571 Bare lymphocyte syndrome, type I
- *601962 Tap-binding protein; TAPBP

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for TAP2 deficiency
- IDR factfile for TAP1 deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

TAP-deficient patients have respiratory inflammations and skin ulcers. The number of peripheral blood CD8 T cells and NK cells is reduced.

Diagnosis:

Diagnostic laboratories:

Clinical:

- ORPHANET

Genetic:

- IDdiagnostics

Therapeutic options:

- Intravenous gamma-globulin infusion, irradiation of all blood products, antibiotherapy.
- eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

GENE INFORMATION

Names:

HUGO name: TAPBP
Alias(es): NGS17, TAPA, TAPASIN, TPN, TPSN, TAP binding protein (tapasin), Tapasin precursor, TAP-associated protein

Localization:

Reference sequences:

DNA: Z97183 (EMBL) , **cDNA:** AF029750 (EMBL) , **Protein:** O15533 (SWISSPROT) Other Sequences

Chromosomal Location:

6p21.3

Maps:

TAPBP (Map View)

Variations / Mutations:

- TAPBPbase; Mutation registry for Tapasin deficiency

Other gene-based resources:

Ensembl: ENSG00000112493, GENATLAS: TAPBP, GeneCard: TAPBP, UniGene: 370937, Entrez Gene: 6892, euGenes: 6892, GDB: 9784212, HomoloGene: 2401

PROTEIN INFORMATION

Description:

Protein function:

Involved in the association of MHC class I with transporter associated with antigen processing (TAP) and in the assembly of MHC class I with peptide (peptide loading).

Subunit:

Interacts with TAP1 and is thus a subunit of the TAP complex. Interaction with TAP1 is TAP2 independent and is required for efficient peptide-TAP interaction. Obligatory mediator for the interaction between newly assembled MHC class I molecules, clareticulin, erp57 and TAP. Up to 4 MHC class I/tapasin complexes bind to 1 TAP.

Subcellular location:

Type I membrane protein. Endoplasmic reticulum.

Polymorphism:

The 2 alleles of tapbp; tapbp*01 (tapasin*01) and tapbp*02 (tapasin*02); are in linkage disequilibria with the HLA-drb1 locus in a japanese population.

Tissue specificity:

Neutrophils, mostly in fully differentiated cells.

Similarity:

Contains 1 Ig-like C1-type (immunoglobulin-like) domain.

Domains:

Ig-like C1-type domain: 292-399

Other features:

Signal peptide: 1-20

Tapasin: 21-448

Disulfide bonds: 315-382
N-linked (glcnac...) glycosylation sites: 253
Other related resourses:

InterPro: IPR007110; Ig-like, InterPro: IPR003597; Ig_c1, InterPro: IPR003006; Ig_MHC, InterPro: IPR008056; Tapasin, Pfam: PF07654; C1-set, PRINTS: PR01669; TAPASIN, PROSITE: PS50835; IG_LIKE, PROSITE: PS00290; IG_MHC

Expression pattern for human:

Tissue	Exp. (%)	Clones
colon mucosa	13.55	1:1345
thymus	8.10	1:2249
tongue	6.76	1:2696
cervix	5.49	10:33205
pericardium	4.59	1:3970
mammary_gland	4.31	14:59134
small_intestine	4.09	3:13365
thyroid	3.96	2:9195
peripheral_nervous_system	3.32	4:21972
thymus	3.29	1:5542

Animal models:

Mouse:

MGD: ; 4, NCBI Gene: ; 21356 (75.17 % aminoacid similarity to human)

Rat:

NCBI Gene: ; 25217 (75.84 % aminoacid similarity to human)

OTHER RESOURCES

Societies:

General:

- IPOPI, International Patient Organization for Primary Immunodeficiencies
- The Jeffrey Modell Foundation
- National Center for Biotechnology Information
- Immune Deficiency Foundation
- European Society for Immunodeficiencies

Other information sources:

- Immunodeficiencies