TAP2 deficiency

GENERAL INFORMATION

Description:

HLA class I deficiency, also named type I Bare lymphocyte syndrome (BLS), is revealed when HLA class I molecules on lymphocytes cannot be typed by serological techniques due to a low expression of HLA class I molecules on the plasma membrane. This syndrome is caused by any of several defects that result in the lack of cell surface expression of MHC class I or class I and II. It affects both maturation of T cells in the thymus and peripheral immune reactions. The number of well-documented cases of HLA class I molecule deficiencies with normal expression of class II molecules is low.

Alternative names:

- HLA class I deficiency
- Bare lymphocyte syndrome (BLS) type I

Classification:

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

Inheritance:

Autosomal recessive

OMIM:

- #604571 Bare lymphocyte syndrome, type I
- *170261 Transporter, ATP-binding cassette, major histocompatibility complex, 2; TAP2

Cross references:

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Contrary to type II and type III Bare lymphocyte syndrome, which are characterized by the early onset of Severe Combined Immunodeficiency, class I antigen deficiencies are not accompanied by particular pathologic manifestations during the first years of life, although chronic lung disease develops in late childhood. Also in contrast to type II or type III BLS, pathology of the gut (diarrhea) is not observed. Systemic infections have not been described in HLA class I-deficient patients. Chronic bacterial infections, often beginning in the first decade of life, are restricted to the respiratory tract and extend from the upper to the lower airway. Bronchiectasis, emphysema, panbronchiolitis, and bronchial obstruction have been described.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Defective expression of HLA class 1, ORPHANET

Therapeutic options:

- Bone marrow transplantation is the only treatment of SCID. Other recommendations include intravenous gamma-globulin infusion, irradiation of all blood products, antibiotherapy.
- Bone marrow transplant, UCSF Medical Center
- Stem Cell Transplant, National Marrow Donor Program (NMDP)

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: TAP2
Alias(es): ABCB3, D6S217E, PSF2, RING11, Y1, ATP-binding cassette, sub-family B (MDR/TAP), member 3, transporter 2, ABC, ATP binding cassette, transporter 2, ATP-binding cassette, sub-family B (MDR/TAP), Antigen peptide transporter 2, APT2, Peptide transporter TAP2, Peptide transporter PSF2, Peptide supply factor 2, PSF-2, Peptide transporter involved in antigen processing 2

Localization:

Reference sequences:

DNA: X66401 (EMBL) , **cDNA:** M84748 (EMBL) , **Protein:** Q03519 (SWISSPROT)

Chromosomal Location:

6p21.3

Maps:

TAP2 (Map View)

Variations / Mutations:

- TAP2base; Mutation registry for TAP2 deficiency

Other gene-based resources:

Ensembl: ENSG00000080469, GENATLAS: TAP2, GeneCard: TAP2, UniGene: 502, EntrezGene: 6891, euGenes: 6891, GDB: 132669

PROTEIN INFORMATION

Description:

Protein function:

Involved in the transport of antigens from the cytoplasm to a membrane-bound compartment for association with MHC class I molecules.

Subunit:

Heterodimer of TAP1 and TAP2.

Subcellular location:

Integral membrane protein.

Induction:

By interferon gamma.

Polymorphism:

The following alleles of TAP2 are known: TAP2*0101, TAP2*0102 (TAP2e), TAP2*0103 and TAP2*0201 (psf2b).

Other features:

ATP nucleotide phosphate-binding region: 503-510

Other related resourses:

PIR: B41538, InterPro: IPR003593; AAA_ATPase, InterPro: IPR003439; ABC_transportr, InterPro: IPR001140; ABCtranprtrTM, InterPro: IPR005293; Ag_transporter2, Pfam: PF00005; ABC_tran, Pfam: PF00664; ABC_membrane, SMART: SM00382; AAA, PROSITE: PS00211; ABC_TRANSPORTER

Expression pattern for human:

Tissue	Exp. (%)	Clones
leukocyte	13.25	3:8982
aorta, endothelium	13.04	1:3042
ovary (pool of 3)	9.06	1:4380
leukopheresis	8.71	1:4557
rpe and choroid	7.51	2:10565
blood	6.27	2:12646
B-cells	4.80	2:16533
blood, lymphocyte	3.50	1:11328
mixed	3.29	5:60341
stomach	2.88	8:110283

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