p14 deficiency

GENERAL INFORMATION

Description:

p14 deficiency resembles known lysosomal diseases associating partial albinism and immunodeficiency, such as Chediak-Higashi syndrome (LYST deficiency), Griscelli syndrome type 2 (RABD27A deficiency) or Hermansky-Pudlak syndrome type 2 (HPS2, AP3B1 deficiency). The point mutation of p14 leads to decreased RNA stability, decreased protein levels and aberrant lysosomal function.

Alternative names:

- p14D
- MAPBPIP deficiency

Classification:

- Defects of phagocyte function
  - Disorders of pigmentation and immunodeficiency

Inheritance:

OMIM:

- #610798 Immunodeficiency due to defect in MAPBPIP-interacting protein
- *610389 MAPBPIP-interacting protein (MAPBPIP)

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Chediak-Higashi syndrome
- IDR factfile for Griscelli syndrome, type 2

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients had short stature, hypopigmented skin, coarse facial features, and recurrent bronchopulmonary infections due to Streptococcus pneumoniae. Affected individuals had consistently low peripheral neutrophil counts, with absolute neutrophil counts less than 500 per microliter, although neutrophil maturation in the bone marrow was intact. Compared to healthy sibs, CD8(+) cytotoxic T cells from affected individuals had decreased cytotoxic activity. Structural and functional analysis of patients' immune cells and melanocytes suggested abnormal maturation and function of specialized lysosomes in cytotoxic T cells, melanocytes, and neutrophil granulocytes.

Diagnosis:

Diagnostic laboratories:

Clinical:

- ORPHANET

Genetic:

- IDdiagnostics

Therapeutic options:

- eMedicine

Research programs, clinical trials:

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GENE INFORMATION

Names:

HUGO name: MAPBPIP

Localization:

Reference sequences:

DNA: D0123 (IDRefSeq) , **cDNA:** C0123 (EMBL) , **Protein:** Q9Y2Q5 (SWISSPROT)

Chromosomal Location:

Maps:

MAPBPIP (Map View)

Variations / Mutations:

- MAPBPIPbase; Mutation registry for p14 deficiency

Other gene-based resources:

Ensembl: ENSG00000116586, GENATLAS: MAPBPIP, GeneCard: MAPBPIP, UniGene: 632483, Entrez Gene: 289562, euGenes: 51639, GDB: 632483, HomoloGene: 8518

PROTEIN INFORMATION

Description:

Other features:

Other related resourses:

InterPro: IPR004942; Robl_LC7, Pfam: PF03259; Robl_LC7

Expression pattern for human:

Tissue	Exp. (%)	Clones
other	17.40	1:3857
umbilical_cord	7.11	1:9438
bone_marrow	6.63	4:40470
bone	4.81	5:69792
prostate	4.56	8:117863
ascites	4.28	3:47004
mammary_gland	4.27	5:78510
mixed	3.97	17:287479
uncharacterized_tissue	3.44	11:214464
lymph_node	3.35	4:80139

Animal models:

Mouse:

MGD: ; 4

OTHER RESOURCES

Societies:

General:

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

Other information sources:

- Immunodeficiencies