Griscelli syndrome, type 3

GENERAL INFORMATION

Description:

Defects in MLPH are a cause of Griscelli syndrome type 3. GS3 is a rare autosomal recessive disorder characterized by pigmentary dilution of the skin and hair, the presence of large clumps of pigment in hair shafts, and an accumulation of melanosomes in melanocytes, without other clinical manifestations.

Alternative names:

- GS3
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Classification:

- Defects of phagocyte function
  - Griscelli syndrome

Inheritance:

Autosomal recesseive

OMIM:

- *606526 Melanophilin; MLPH
- #609227 Griscelli syndrome type 3

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Chediak-Higashi syndrome
- IDR factfile for Griscelli syndrome, type 1
- IDR factfile for Griscelli syndrome, type 2

Incidence:

Incidence is not known yet.

CLINICAL INFORMATION

Description:

In Griscelli syndrome type 3, patients have hypomelanosis with no immunologic or neurologic manifestations.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Griscelli disease, ORPHANET
- Griscelli Syndrome, eMedicine

Genetic:

- IDdiagnostics

Therapeutic options:

- Bone marrow transplantation.
- Griscelli Syndrome, eMedicine

Research programs, clinical trials:

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GENE INFORMATION

Names:

HUGO name: MLPH

Localization:

Reference sequences:

DNA: D0059 (IDRefSeq) , **cDNA:** AK022207 (EMBL) , **Protein:** Q9BV36 (SWISSPROT) Other Sequences

Chromosomal Location:

2q37

Maps:

MLPH (Map View)

Variations / Mutations:

- MLPHbase; Mutation registry for Griscelli syndrome, type 3

Other gene-based resources:

Ensembl: ENSG00000115648, GENATLAS: MLPH, GeneCard: MLPH, UniGene: 102406, Entrez Gene: 79083, euGenes: 79083, GDB: 11523848, HomoloGene: 11465

PROTEIN INFORMATION

Description:

Protein function:

RAB effector protein involved in melanosome transport. Serves as link between melanosome-bound RAB27a and the motor protein MYO5a.

Subunit:

Binds RAB27a that has been activated by GTP-binding via its N-terminus. Binds MYO5a via its C-terminal coiled coil domain.

Subcellular location:

Cytoplasm

Protein function:

Event=alternative splicing; named isoforms=2; name=1; isoid=q9bv36-1; sequence=displayed; name=2; isoid=q9bv36-2; sequence=vsp_007554;

Similarity:

Contains 1 fyve-type zinc finger.

Domains:

RabBD domain: 4-124

Other features:

Melanophilin: 1-600

Zinc finger region Fyve-type: 64-107

Other related resourses:

InterPro: IPR010911; Rab_bd, InterPro: IPR000306; Znf_FYVE, InterPro: IPR011011; Znf_FYVE_PHD, PROSITE: PS50916; RABBD, PROSITE: PS50178; ZF_FYVE

Expression pattern for human:

Tissue	Exp. (%)	Clones
mammary_gland	17.84	76:79431
rectum	9.71	3:5759
prostate	9.05	58:119494
trachea	7.64	19:46384
abdominal_cavity	6.33	16:47125
skin	6.01	56:173577
thyroid	3.65	2:10201
uterus	2.96	30:189232
nerve_tissue	2.93	4:25421
salivary_gland	2.87	3:19467

Animal models:

Mouse:

MGD: ; Mlph

OTHER RESOURCES

Societies:

General:

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

Disease specific:

- European Hair Research Society

Other information sources:

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