Schimke immuno-osseous dysplasia

GENERAL INFORMATION

Description:

Defects in SMARCAL1 are a cause of schimke immuno-osseous dysplasia [mim:242900].

Alternative names:

- SIOD
- chondroitin-6-sulfate mucopolysacharidosis, Immuno osseous dysplasia schimke type, Schimke syndrome, Spondyloepiphyseal dysplasia nephrotic syndrome

Classification:

- Combined B and T cell immunodeficiencies
  - Other

Inheritance:

Autosomal recessive

OMIM:

- *606622 SWI/SNF-related, matrix-associated, actin-dependent regulator of chromatin, subfamily A-like protein 1; SMARCAL1
- #242900 Immunoosseous dysplasia, Schimke type

Cross references:

Incidence:

Incidence is not known yet.

CLINICAL INFORMATION

Description:

SIOD causes spondyloepiphyseal dysplasia, causing disproportionate stature, renal dysfunction, lymphocytopenia with T-cell immunodeficiency, facial dysmorfism. Approximately half of all patients also exhibit hyperthyroidism, while around half also exhibit episodal cerebral ischemia. Other features include hyperpigmented macules on the trunk, corneal opacities, other hematological abnormalities.

Diagnosis:

Diagnostic laboratories:

Clinical:

- ORPHANET

Genetic:

- ORPHANET
- GeneTest
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Therapeutic options:

- Management of SIOD patients is symptomatic. Renal disease progresses from proteinuria to end-stage renal disease. ACE-inhibitor enalapril plus ATII receptor antagonist losartan can reduce proteinuria. Cyclosporin A, tacrolimus, or corticosteroids have had a transient reduction in the rate of renal disease progression. Renal transplantation effectively treats the nephropathy. Prophylaxis against Pneumocystis carinii pneumonia is usually recommended. Bone marrow transplantation (BMT) treats neutropenia resulting in immunodeficiency and recurrent infections.

Research programs, clinical trials:

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GENE INFORMATION

Names:

HUGO name: SMARCAL1

Localization:

Reference sequences:

DNA: IDRefSeq: D0079 (IDRefSeq) , **cDNA:** AF082179 (EMBL) , **Protein:** Q9NZC9 (SWISSPROT) Other Sequences

Chromosomal Location:

2q34-q36

Maps:

SMARCAL1 (Map View)

Variations / Mutations:

- SMARCAL1base; Mutation registry for Schimke immuno-osseous dysplasia

Other gene-based resources:

Ensembl: ENSG00000138375, GENATLAS: SMARCAL1, GeneCard: SMARCAL1, UniGene: 516674, Entrez Gene: 50485, euGenes: HUgn0050485, GDB: 10796078, HomoloGene: 8558

PROTEIN INFORMATION

Description:

Protein function:

Possesses intrinsic ATP-dependent nucleosome remodelling activity (by similarity)

Subcellular location:

Nucleus (by similarity)

Tissue specificity:

Ubiquitously expressed, with high levels in testis

Similarity:

Belongs to the SNF2/RAD54 helicase family

Domains:

HARP 1 domain: 248-302

HARP 2 domain: 343-397

Helicase ATP-binding domain: 445-600

Helicase C-terminal domain: 716-869

Other features:

Swi/snf-related matrix-associated actin-dependent regulator of chromatin subfamily a-like protein 1: 1-954

ATP nucleotide phosphate-binding region: 458-465

Other related resourses:

PIR: T34557, InterPro: IPR014001; DEAD-like_N, InterPro: IPR010003; HARP, InterPro: IPR014021; Helic_SF1/SF2_ATP_bd, InterPro: IPR001650; Helicase_C, InterPro: IPR000330; SNF2_N, Pfam: PF07443; HARP, Pfam: PF00271; Helicase_C, Pfam: PF00176; SNF2_N, SMART: SM00487; DEXDc, SMART: SM00490; HELICc, PROSITE: PS51192; HELICASE_ATP_BIND_1, PROSITE: PS51194; HELICASE_CTER

Expression pattern for human:

Tissue	Exp. (%)	Clones
tongue	8.25	5:37267
bone_marrow	7.43	5:41353
umbilical_cord	6.59	1:9321
testis	6.30	29:282747
thymus	5.36	6:68776
cervix	4.65	3:39652
connective_tissue	4.48	7:96057
blood	4.00	4:61437
other	2.83	9:195669
lymph	2.81	2:43667

Animal models:

Mouse:

MGD: ; 4

OTHER RESOURCES

Societies:

General:

- European Society for Immunodeficiencies
- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- March of Dimes Birth Defects Foundation
- NIH/National Institute of Allergy and Infectious Diseases

Disease specific:

- Human Growth Foundation
- Little People of America (LPA)
- The MAGIC Foundation
- International Skeletal Dysplasia Registry

Other information sources:

- Schimke immuno-osseous dysplasia