T-cell immunodeficiency, congenital alopecia, and nail dystrophy

GENERAL INFORMATION

Description:

Mutation in the FOXN1(WHN) gene cause an autosomal recessive syndrome characterised by decrease of mature T and a low number of helper T cells, whereas the number of suppressor/cytotoxic T cells was relatively normal. Two patients have been described until now.

Alternative names:

- Human Nude/SCID

Classification:

- Combined B and T cell immunodeficiencies
  - T⁻B⁺ SCID

Inheritance:

Autosomal recessive

OMIM:

- #601705 T-cell immunodeficiency, congenital alopecia, and nail dystrophy
- *600838 Winged helix nude; WHN

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Omenn syndrome

Incidence:

Incidence is not known yet.

CLINICAL INFORMATION

Description:

The clinical symptoms are similar with those for SCID: failure to thrive, intractable diarrhea, and recurrent lung infection. Beside these symptoms patients can have congenital alopecia, severe T-cell immunodeficiency, and ridging and pitting of all nails.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Severe Combined Immunodeficiency, eMedicine
- T-cell disorder, eMedicine

Genetic:

- Alopecia and T-cell deficiency, Gene Test

Therapeutic options:

- Bone marrow transplantation is the only treatment of SCID. Other recommendations include intravenous gamma-globulin infusion, irradiation of all blood products, antibiotherapy.
- BMT, supportive treatment, prophylaxis of infection and tailored conditioning regimen, National Marrow Donor Program
- Omenn syndrome, eMedicine
- T-cell disorder, eMedicine
- Severe Combined Immunodeficiency, eMedicine

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: FOXN1
Alias(es): FKHL20, WHN, Transcription factor winged-helix nude, Forkhead box N1, Winged helix nude, Winged-helix nude, Forkhead box protein N1

Localization:

Reference sequences:

DNA: Y11741 (EMBL) Y11739 (EMBL) , **cDNA:** X58957 (EMBL) , **Protein:** O15353 (SWISSPROT) Other Sequences

Chromosomal Location:

17q11-q12

Maps:

WHN (Map View)

Markers:

SHGC-57757

Variations / Mutations:

- FOXN1base; Mutation registry for T-cell immunodeficiency, congenital alopecia, and nail dystrophy

Other gene-based resources:

Ensembl: ENSG00000109101, GENATLAS: FOXN1, GeneCard: FOXN1, UniGene: 198313, EntrezGene: 8456, euGenes: 8456, GDB: WHN, HomoloGene: 8456

PROTEIN INFORMATION

Description:

Protein function:

Transcriptional regulator involved in development

Subcellular location:

Nuclear

Tissue specificity:

Expressed in thymus

Similarity:

Contains 1 fork-head domain

Other features:

DNA-binding region Fork-head: 271-367

Other related resourses:

InterPro: IPR001766; TF_Fork_head, Pfam: PF00250; Fork_head, SMART: SM00339; FH, PROSITE: PS00657; FORK_HEAD_1, PROSITE: PS00658; FORK_HEAD_2, PROSITE: PS50039; FORK_HEAD_3

Expression pattern for human:

Tissue	Exp. (%)	Clones
eNSG00000109101		
eNSG00000109101		

Animal models:

Mouse:

MGD: ; Foxn1

OTHER RESOURCES

Societies:

General:

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

Disease specific:

- National Alopecia Areata Foundation, USA