Artemis deficiency

GENERAL INFORMATION

Description:

Artemis deficiency is included in the most severe phenotype, T-B- SCID and it is inherited as an autosomal recessive trait. The disease is characterized by a profound deficiency of both T cell and B cell immunity. It is caused by a mutation of Artemis gene which codes for a novel V(D)J recombination/DNA repair factor that belongs to the metallo-lactamase superfamily.

Alternative names:

- RS-SCID
- Native American SCID
- Severe combined immunodeficiency with sensitivity to ionizing radiation
- Severe Combined Immunodeficiency, Athabaskan type
- SCIDA
- Artemis
- Athabaskan severe combined immunodeficiency
- SCID with radiosensitivity

Classification:

- Combined B and T cell immunodeficiencies
  - T⁻B⁻ Severe combined immunodeficiency (SCID)

Inheritance:

Autosomal recessive

OMIM:

- #602450 Severe combined immunodeficiency with sensitivity to ionizing radiation
- *605988 DNA cross-link repair protein 1C; DCLRE1C

Incidence:

1 in 2,000 live births among Navajo Indians

CLINICAL INFORMATION

Description:

Clinically, affected children present with oral/ genital ulcers and life-threatening infections within the first 3 months of life. Without a bone marrow transplant, most die by 6 months of age.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Severe combined immunodeficiency, T-B-, ORPHANET
- Severe combined immunodeficiency, eMedicine

Genetic:

- DCLRE1C, IDdiagnostics

Therapeutic options:

- Bone marrow transplantation is the only treatment of SCID. Other recommendations include intravenous gamma-globulin infusion, irradiation of all blood products, antibiotherapy.
- Severe combined immunodeficiency, T-B-, ORPHANET
- Severe combined immunodeficiency, eMedicine
- BMT for Severe Combined Immunodeficiencies

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: DCLRE1C
Alias(es): A-SCID, ARTEMIS PROTEIN, FLJ11360, HGNC:10578, SCIDA, SNM1C, DNA cross-link repair 1C (PSO2 homolog, S. cerevisiae), severe combined immunodeficiency, type a (Athabascan)

Localization:

Reference sequences:

DNA: AJ296101 (EMBL) , **cDNA:** AJ296101 (EMBL) , **Protein:** Q96SD1 (SWISSPROT)

Chromosomal Location:

10p13

Maps:

A007G45, STS-R68462

Variations / Mutations:

- DCLRE1base; Mutation registry for Artemis deficiency

Other gene-based resources:

Ensembl: ENSG00000152457, GENATLAS: DCLRE1C, GeneCard: DCLRE1C, UniGene: 524156, Entrez Gene: 64421, euGenes: 64421, GDB: DCLRE1C

PROTEIN INFORMATION

Description:

Other features:

Expression pattern for human:

Tissue	Exp. (%)	Clones
stem cells	32.43	1:394
frontal lobe	11.50	4:4446
five pooled sarcomas, including myxoid liposarcoma, solitary fibrous tumor, malignant fibrous histiocytoma, gastrointest	9.74	1:1312
brain	8.71	2:2933
whole embryo, mainly head	3.83	1:3340
alveolar macrophage	2.73	1:4674
kidney tumor	2.58	1:4953
B-cell, chronic lymphotic leukemia	2.22	2:11493
bone marrow	2.19	1:5839
germinal center B cell	2.05	6:37460

Animal models:

FlyBase::

euGenes: ; FlyBase

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:

- The SCID Homepage

Other information sources:

- Severe Combined Immunodeficiency, Patient and Family Handbook
- Severe Combined Immunodeficiency, KidsHealth