Familial cold urticaria and Muckle-Wells syndrome

GENERAL INFORMATION

Description:

Defects in CIAS1 are a cause of familial cold autoinflammatory syndrome. FCAS is rare autosomal dominant systemic inflammatory disease characterized by episodes of rash, arthralgia, fever and conjunctivitis after generalized exposure to cold.

Alternative names:

- FCAS
- Familial cold autoinflammatory syndrome, Cold hypersensitivity, Familial cold-induced autoinflammatory syndrome, Familial polymorphous cold eruption

Classification:

- Periodic fever syndromes
  - Cold autoinflammatory syndrome

Inheritance:

Autosomal dominant

OMIM:

- #120100 Familial cold autoinflammatory syndrome
- #191900 Muckle-Wells syndrome
- *606416 CIAS1 gene; CIAS1

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Chronic infantile neurological cutaneous and articular syndrome
- IDR factfile for Granulomatous sinovitis with uveitis and cranial neuropathies
- IDR factfile for Crohn's disease

Incidence:

Incidence unknown.

CLINICAL INFORMATION

Description:

Most patients present at birth or during the first few months of life with rash. The attacks usually occur 1-2 hours following exposures to cold and last less than 24 hours. Other symptoms include fever, joint pain, myalgia, conjunctivitis, sweating, drowsiness, headache, extreme thirst, and nausea. Renal diseases is rare.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Familial cold urticaria, ORPHANET

Genetic:

- IDdiagnostics

Therapeutic options:

- Supportive treatments with warming and non-steroidal anti-inflammatory drugs. High dose of corticosteroids can be effective. Other treatments include anabolic steroids and gold.
- Mediterranean fever, familial, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

GENE INFORMATION

Names:

HUGO name: CIAS1
Alias(es): AGTAVPRL, AII/AVP, C1orf7, FCAS , FCU, MWS, NALP3, PYPAF1 , chromosome 1 open reading frame 7, Cryopyrin, PYRIN-containing APAF1-like protein 1, Angiotensin/ vasopressin receptor AII/AVP-like

Localization:

Reference sequences:

DNA: AY051117 (EMBL) , **cDNA:** AF410477 (EMBL) , **Protein:** Q96P20 (SWISSPROT) Other Sequences

Chromosomal Location:

1q44

Maps:

CIAS1 (Map View)

Other gene-based resources:

Ensembl: ENSG00000162711, GENATLAS: CIAS1, GeneCard: CIAS1, UniGene: 159483, Entrez Gene: 114548, euGenes: 114548, GDB: 9957338, HomoloGene: 3600

PROTEIN INFORMATION

Description:

Protein function:

May function as a potential inducer of apoptosis. Interacts selectively with apoptosis-associated specklike protein containing a card domain (asc). This complex may function as an upstream activator of NF-kappa-b signaling.

Tissue specificity:

Expressed in blood leukocytes. Strongly expressed in polymorphonulclear cells, undetectable or expressed at a lower magnitude in B and T lymphoblasts, respectively. High level of expression detected in chondrocytes. Low or no expression in the other tissues tested.

Similarity:

Contains 1 DAPIN domain.

Domains:

DAPIN domain: 1-91

NACHT domain: 218-534

Other features:

Other related resourses:

InterPro: IPR001611; LRR, InterPro: IPR007091; LRR_RNinh, InterPro: IPR003590; LRR_RNinh_sub, InterPro: IPR007111; NACHT_NTPase, InterPro: IPR004020; PAAD_DAPIN, Pfam: PF00560; LRR_1, Pfam: PF05729; NACHT, Pfam: PF02758; PAAD_DAPIN, PRINTS: PR00019; LEURICHRPT, SMART: SM00368; LRR_RI, PROSITE: PS50824; DAPIN, PROSITE: PS50837; NACHT

Expression pattern for human:

Tissue	Exp. (%)	Clones
neuroblastoma	52.03	1:1205
thymus	27.88	1:2249
pooled	10.86	1:5774
pancreatic islet	4.54	1:13811
bone_marrow	2.07	1:30226
testis	1.10	2:113913
mixed	0.69	3:270774
other	0.57	4:442392
lung	0.26	1:244782

Animal models:

Mouse:

MGD: ; 4

OTHER RESOURCES

Societies:

General:

- IPOPI, International Patient Organization for Primary Immunodeficiencies
- The Jeffrey Modell Foundation
- Immune Deficiency Foundation
- European Society for Immunodeficiencies

Other information sources:

- Immunodeficiencies