Chronic infantile neurological cutaneous and articular syndrome

GENERAL INFORMATION

Description:

CINCA syndrome is a rare congenital inflammatory disorder characterized by a triad of neonatal onset of cutaneous symptoms, chronic meningitis, and joint manifestations with recurrent fever and inflammation.

Alternative names:

- FCAS, CINCA, NOMID
- Multisystem inflammatory disease, neonatal-onset
- Neonatal onset multisystem inflammatory disease
- Chronic infantile neurologic cutaneous articular syndrome

Classification:

- Periodic fever syndromes
  - Cold autoinflammatory syndrome

Inheritance:

Autosomal dominant

OMIM:

- #607115 CINCA syndrome
- *606416 Cias1 gene; CIAS1

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Familial cold urticaria and Muckle-Wells syndrome
- IDR factfile for Granulomatous sinovitis with uveitis and cranial neuropathies
- IDR factfile for Crohn's disease

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have maculopapular urticarial skin rash, often present at birth but whose presence varies with time, articular signs of variable of expression including transient swelling or anomalies of growth cartilage with no inflammatory cells, symptoms of central nervous system like headache. Infants are born preterm or dysmature.

Diagnosis:

Diagnostic laboratories:

Clinical:

- CINCA syndrome, ORPHANET

Genetic:

- IDdiagnostics

Therapeutic options:

- Treatment is symptomatic, oral steroids and anti-inflammatory drug therapy.

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:

- International Patient Organization for Primary Immunodeficiencies (IPOPI)

Other information sources:

- Immunodeficiencies