

# Granulomatous sinovitis with uveitis and cranial neuropathies

## GENERAL INFORMATION

### Description:

Defects in CARD15 are the cause of Blau syndrome, an autosomal dominant disorder characterized by early-onset granulomatous arthritis, uveitis and skin rash.

### Alternative names:

- BS, ACUG
- Blau syndrome
- Granulomatosis, familial juvenile systemic
- Arthrocutaneouveal granulomatosis
- Jabs syndrome
- Granulomatous inflammatory arthritis, dermatitis, and uveitis, familial
- Granulomatosis, familial, Blau type
- Jabs houk bias syndrome
- Synovitis granulomatous uveitis cranial neuropathies

### Classification:

- Periodic fever syndromes

### Inheritance:

Autosomal dominant

### OMIM:

- #186580 Synovitis, granulomatous, with uveitis and cranial neuropathies
- \*605956 Caspase recruitment domain-containing protein 15; CARD15

### Cross references:

#### Phenotype related immunodeficiencies:

- IDR factfile for Familial cold urticaria and Muckle-Wells syndrome
- IDR factfile for Chronic infantile neurological cutaneous and articular syndrome
- IDR factfile for Crohn's disease

### Incidence:

Incidence is not known.

## CLINICAL INFORMATION

### Description:

Patients have granulomatous arthritis, skin eruption, and uveitis occurring in the absence of lung or other visceral involvement. Sometimes they can present camptodactyly (flexion contractures of the elbows and of the third, fourth, and fifth proximal interphalangeal joints of the hands bilaterally).

### Diagnosis:

### Diagnostic laboratories:

#### Clinical:

- Synovitis granulomatous uveitis cranial neuropathies, ORPHANET

#### Genetic:

- IDdiagnostics

### Therapeutic options:

- Oral steroids treatment.

## Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

## GENE INFORMATION

### Names:

**HUGO name:** CARD15

**Alias(es):** ACUG, BLAU, CD, IBD1, NOD2 , PSORAS1, Nucleotide-binding oligomerization domain protein 2, Caspase recruitment domain protein 15, Nod2 protein, Inflammatory bowel disease protein 1

### Localization:

#### Reference sequences:

**DNA:** AF385089 (EMBL) , **cDNA:** AF178930 (EMBL) , **Protein:** Q9HC29 (SWISSPROT) Other Sequences

#### Chromosomal Location:

16q12

#### Maps:

CARD15 (Map View)

### Other gene-based resources:

Ensembl: ENSG00000167207, GENATLAS: CARD15, GeneCard: CARD15, UniGene: 135201, Entrez Gene: 64127, euGenes: 64127, GDB: 11026232, HomoloGene: 11156

## PROTEIN INFORMATION

### Description:

#### Protein function:

Induces NF-kappa-b via rick (cardiak, rip2) and IKK-gamma. Confers responsiveness to intracellular bacterial lipopolysaccharides (lps).

#### Subunit:

Binds to rick by CARD-CARD interaction.

#### Subcellular location:

Cytoplasmic.

#### Similarity:

Contains 2 card domains.

### Domains:

**CARD 1 domain: 26-122**

**CARD 2 domain: 126-218**

**NACHT domain: 293-618**

### Other features:

**Caspase recruitment domain protein 15, isoform 1: 1-1040**

**Caspase recruitment domain protein 15, isoform 2: 28-1040**

**ATP nucleotide phosphate-binding region: 299-306**

#### Other related resources:

InterPro: IPR001315; CARD, InterPro: IPR011029; DEATH\_like, InterPro: IPR001611; LRR, InterPro: IPR007091; LRR\_RNinh, InterPro: IPR007111; NACHT\_NTPase, Pfam: PF00619; CARD, Pfam: PF00560; LRR\_1, Pfam: PF05729; NACHT, SMART: SM00114; CARD, PROSITE: PS50209; CARD, PROSITE: PS50837; NACHT

## Expression pattern for human:

Tissue	Exp. (%)	Clones
pooled	63.09	1:5774
heart	8.01	1:45449
blood	7.61	1:47876
muscle	7.45	2:97778
lung	5.95	4:244782
kidney	2.75	1:132304
uterus	2.66	1:136836
placenta	2.47	1:147272

## Animal models:

### Mouse:

MGD: ; 4, NCBI Gene: ; 257632 (79.21 % aminoacid similarity to human)

### Rat:

NCBI Gene: ; 291912 (80.04 % aminoacid similarity to human)

## 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