Crohn's disease

GENERAL INFORMATION

Description:

Defects in CARD15 are the cause of Crohn disease and Blau syndrome. Inflammatory bowel disease is characterized by a chronic relapsing intestinal inflammation. IBD is subdivided into Crohn disease and ulcerative colitis. Crohn disease may involve any part of the gastrointestinal tract, terminal ileum and colon. Bowel inflammation is transmural and discontinuous; it may contain granulomas or be associated with intestinal or perianal fistulas. In ulcerative colitis, the inflammation is continuous and limited to rectal and colonic mucosal layers; fistulas and granulomas are not observed. In approximately 10% of cases confined to the rectum and colon, definitive classification of Crohn disease or ulcerative colitis cannot be made and are designated 'indeterminate colitis.' Both diseases include extraintestinal inflammation of the skin, eyes, or joints.

Alternative names:

- IBD1, CD
- Inflammatory bowel disease 1
- Regional enteritis
- Granulomatous colo-ileitis

Classification:

- Periodic fever syndromes

Inheritance:

Autosomal dominant

OMIM:

- #266600 Inflammatory bowel disease 1
- *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 Granulomatous sinovitis with uveitis and cranial neuropathies

Incidence:

Incidence it varies according to the geographic area. 140/100,000 in Scandinavia, Great Britain, USA, Canada to 50/100,000 in Southern Europe.

CLINICAL INFORMATION

Description:

Patients have diarrhea, abdominal pain, rectal bleeding, anorexia, weight loss. Extra digestive manifestations may occur in parallel with the digestive symptoms during attacks such as fever, arthralgia, arthritis, buccal aphtosis, erythema nodosa, pyoderma gangrenosum, iritis, episcleritis.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Crohn disease, eMedicine

Genetic:

- IDdiagnostics

Therapeutic options:

- The main drugs are salicylates, steroids, immunomodulators. Surgery is often required for treatment of bowel stenosis, abscess, internal fistula, as well as ano-perineal manifestations of the disease. Patients may require either enteral or parenteral nutrition.
- Crohn disease, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

GENE INFORMATION

Names:

HUGO name: CARD15
Alias(es): ACUG, BLAU, PSORAS1, caspase recruitment domain family, member, nucleotide-binding oligomerization domain protein, Caspase recruitment domain protein 15

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.

Tissue specificity:

Monocytes-specific.

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 resourses:

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:

- International Patient Organization for Primary Immunodeficiencies (IPOPI)

Disease specific:

- American Autoimmune Related Diseases Association
- Chron and Colitis Foundation of America

Other information sources:

- Immunodeficiencies