LAD3 deficiency

GENERAL INFORMATION

Description:

LAD-III deficiency involves a general defect in integrin activation. Defects in the activation of β1, β2 and β3 integrin subunits have been observed and it seems that this rare syndrome may be due to several defects in molecules involved in integrin activation. In LAD3 deficiency, the definitive diagnosis is established by the presence of defects in the integrin activation process while the CD18 molecule is structurally intact.

Alternative names:

- LAD III
- Leukocyte adhesion deficiency type 3

Classification:

- Defects of phagocyte function
  - Leukocyte adhesion defects

Inheritance:

Autosomal recessive

OMIM:

- #116920 Leukocyte adhesion deficiency
- *605577 Ras guanyl nucleotide-releasing protein 2; RASGRP2

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Leukocyte adhesion deficiency I
- IDR factfile for Leukocyte adhesion deficiency II
- IDR factfile for LAD caused by Rac2 deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have recurent bacterial infections, localised to skin and mucosal surfaces with no pus formation, omphalitis with delayed separation of the umbilical cord. Severe bleeding tendency starting at delivery or later and defects in platelet activation. Impaired healing of traumatic or surgical wounds.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Leukocyte adhesion deficiency (LAD), ORPHANET

Genetic:

- RASGRPP2, IDdiagnostics

Therapeutic options:

- Prompt antibiotic therapy initiated early in case of acute infection. Granulocyte transfusion is restricted to life-threatening situations when all other measures have failed. Blood transfusion in case of bleeding episodes.
- Leukocyte adhesion deficiency, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

GENE INFORMATION

Names:

HUGO name: RASGRP2

Localization:

Reference sequences:

DNA: D0127 (IDRefSeq) , **cDNA:** AF081194 (EMBL) , **Protein:** Q7LDG7 (SWISSPROT) Other Sequences

Chromosomal Location:

11q13

Maps:

RASGRP2 (Map View)

Variations / Mutations:

- RASGRP2base; Mutation registry for LAD-III

Other gene-based resources:

Ensembl: ENSG00000068831, GENATLAS: RASGRP2, GeneCard: RASGRP2, UniGene: 99491, Entrez Gene: 99491, euGenes: 10235, GDB: 9955786, HomoloGene: 4250

PROTEIN INFORMATION

Description:

Other features:

Other related resourses:

InterPro: IPR002219; DAG_PE_bd, InterPro: IPR011992; EF-Hand_type, InterPro: IPR002048; EF_hand_Ca_bd, InterPro: IPR008937; Ras_GEF, InterPro: IPR000651; RasGef_N, InterPro: IPR001895; RasGRF_CDC25, Pfam: PF00130; C1_1, Pfam: PF00036; efhand, Pfam: PF00617; RasGEF, Pfam: PF00618; RasGEF_N, PRINTS: PR00008; DAGPEDOMAIN, SMART: SM00109; C1, SMART: SM00054; EFh, SMART: SM00147; RasGEF, SMART: SM00229; RasGEFN, PROSITE: PS00018; EF_HAND_1, PROSITE: PS50222; EF_HAND_2, PROSITE: PS50009; RASGEF_CAT, PROSITE: PS50212; RASGEF_NTER, PROSITE: PS00479; ZF_DAG_PE_1, PROSITE: PS50081; ZF_DAG_PE_2

Expression pattern for human:

Tissue	Exp. (%)	Clones
cranial_nerve	14.75	3:14885
spleen	13.42	9:49068
uncharacterized_tissue	12.63	37:214464
bone_marrow	10.85	6:40470
lymph_node	7.31	8:80139
thymus	6.73	7:76122
blood	5.50	6:79803
whole_brain	4.75	10:154222
prostate	3.10	5:117863
trachea	2.87	2:51052

Animal models:

Mouse:

MGD: ; Rasgrp2

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- March of Dimes Birth Defects Foundation
- NIH/National Institute of Allergy and Infectious Diseases
- European Society for Immunodeficiencies

Other information sources:

- Leukocyte Adhesion Deficiency (LAD) Syndromes
- Immunodeficiencies+RASGRP2