CD19 deficiency

GENERAL INFORMATION

Description:

Mutation of the CD19 gene causes a type of hypogammaglobulinemia with a defective response of mature B cells to antigenic stimulation. CD19 protein forms a complex with CD21, CD81 and CD225 in the membrane of mature B cells.

Alternative names:

- CD19D
- Antibody deficiency due to defect in CD19

Classification:

- Deficiencies predominantly affecting antibody production
  - Other antibody deficiencies

Inheritance:

Autosomal recessive

OMIM:

- +107265 Cd19 antigen; CD19

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Common variable immunodeficiency of unknown origin
- IDR factfile for ICOS deficiency

Incidence:

Incidence is not known yet.

CLINICAL INFORMATION

Description:

Patients have susceptibility to recurrent bacterial infections (sinusitis, otitis, bronchitis), hypogammaglobulinemia and a lack of antibody production in response to vaccination.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Common Variable Immune Deficiency-workup, eMedicine

Genetic:

- CVID, GeneTest

Therapeutic options:

- (Intravenous) immunoglobulins and antibiotic therapy together with physiotherapy and postural drainage in case of lung damage. Oral poliovaccine should not be given because there is risk of paralytic disease.
- Common Variable Immunodeficiency (CVID), eMedicine
- Hypogammaglobulinemia, eMedicine

Research programs, clinical trials:

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GENE INFORMATION

Names:

HUGO name: CD19
Alias(es): μ, Immunoglobin heavy constant μ, immunoglobulin μ, Ig μ chain C region

Localization:

Reference sequences:

DNA: CD19D (IDRefSeq) , **cDNA:** M21097 (EMBL) , **Protein:** P15391 (SWISSPROT) Other Sequences

Chromosomal Location:

16p11.2

Maps:

CD19 (Map View)

Variations / Mutations:

- CD19base; Mutation registry for CD19 deficiency

Other gene-based resources:

Ensembl: ENSG00000177455, GENATLAS: CD19, GeneCard: CD19, UniGene: 632193, Entrez Gene: 930, euGenes: 930, GDB: 127605, HomoloGene: 1341

PROTEIN INFORMATION

Description:

Protein function:

May be involved in growth regulation of B-cells.

Subunit:

Interacts with Vav. Interacts with Grb2 and Sos when phosphorylated on Tyr-348 and/or Tyr-378. Interacts with Plcg2 when phosphorylated on Tyr-409.

Subcellular location:

Membrane; single-pass type I membrane protein.

Protein function:

Ref.1 sequence differs from that shown due to a frameshift in position 396.

Similarity:

Contains 2 Ig-like C2-type (immunoglobulin-like) domains.

Domains:

Ig-like C2-type 1 domain: 20-113

Ig-like C2-type 2 domain: 176-277

Other features:

Signal peptide: 1-19

B-lymphocyte antigen cd19: 20-556

N-linked (glcnac...) glycosylation sites: 86,125,138,181,265

Disulfide bonds: 38-97, 200-261

Other related resourses:

UniProt: CD19_HUMAN, InterPro: IPR007110; Ig-like, InterPro: IPR013783; Ig-like_fold, InterPro: IPR003599; Ig_sub, SMART: SM00409; IG, PROSITE: PS50835; IG_LIKE

Expression pattern for human:

Tissue	Exp. (%)	Clones
spleen	33.40	33:47318
tonsil	31.90	12:18016
rectum	8.32	1:5759
lymph_node	8.07	14:83039
lymph	7.68	7:43667
thymus	2.79	4:68776
uncharacterized_tissue	1.78	8:215582
connective_tissue	1.50	3:96057
bone_marrow	1.16	1:41353
testis	0.85	5:282747

Animal models:

Mouse:

MGD: ; Cd19

OTHER RESOURCES

Societies:

General:

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

Other information sources:

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