Igα deficiency

GENERAL INFORMATION

Description:

Igα deficiency is an autosomal recessive disorder caused by mutation in the Igα (CD79α) leading to B cell defect. Mutations in the gene for Igα as well as in other pre-B cell receptor complex as λ 5/14.1 (IGLL1) and in the the gene for μ heavy chain (IGHM), can cause a disorder clinically similar to XLA.

Alternative names:

- CD79A deficiency, IGA deficiency

Classification:

- Deficiencies predominantly affecting antibody production
  - Agammaglobulinemia

Inheritance:

Autosomal recessive

OMIM:

- #601495 Agammaglobulinemia, non-bruton type
- *112205 CD79A antigen; CD79A

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for X-linked agammaglobulinemia
- IDR factfile for X-linked hypogammaglobulinemia with growth hormone deficiency
- IDR factfile for BLNK deficiency
- IDR factfile for μ heavy-chain deficiency
- IDR factfile for λ5 surrogate light-chain deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Ig# deficiency is characterised by early onset recurrent bacterial infections of lower respiratory tract and otitis media, profound hypoglammaglobulinemia, and a lack of circulating mature B cells. One patient developed recurrent diarrhea and failure to thrive in the first month of life.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Agammaglobulinemia, autosomal recessive, ORPHANET
- Agammaglobulinemia, eMedicine

Therapeutic options:

- (Intravenous) immunoglobulins and antibiotic therapy. Oral poliovaccine should not be given because of the risk of paralytic disease.
- Agammaglobulinemia, eMedicine

Research programs, clinical trials:

- Improved Healthcare for Patients with Primary Antibody Deficiencies through new Strategies Elucidating their Pathophysiology (IMPAD), IMPAD
- European Initiative for Primary Immunodeficiencies
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name: CD79A
Alias(es): IGA, MB-1, MB1, CD79A antigen, B-cell antigen receptor complex associated protein alpha-chain precursor, MB-1 membrane glycoprotein, Surface-IgM- associated protein, Membrane-bound immunoglobulin associated protein, CD79a

Localization:

Reference sequences:

DNA: U05259 (EMBL) , **cDNA:** M86921 (EMBL) X13451 (EMBL) S75217 (EMBL) S46706 (EMBL) M80462 (EMBL) X83540 (EMBL) M74721 (EMBL) , **Protein:** C79A_HUMAN (SWISSPROT)

Chromosomal Location:

19q13.2

Maps:

CD79A (Map View)

Variations / Mutations:

- CD79Abase; Mutation registry for Ig# deficiency

Other gene-based resources:

Ensembl: ENSG00000105369, GENATLAS: CD79A, GeneCard: CD79A, UniGene: 79630, Entrez Gene: 973, euGenes: 973, GDB: 133778

PROTEIN INFORMATION

Description:

Protein function:

Associated to surface IgM-receptor; may be involved in signal transduction.

Subunit:

Heterodimer of # and # chains, disulfide-linked.

Subcellular location:

Type I membrane protein.

Post-translational modification:

Phosphorylated on tyrosine as a result of B-cell activation.

Protein function:

2 isoforms; a long form and a short form; are produced by alternative splicing.

Other features:

Other related resourses:

PIR: A46477, InterPro: IPR003110; ITAM, InterPro: IPR003006; Ig_MHC, InterPro: IPR003598; Ig_c2, Pfam: PF00047; ig, Pfam: PF02189; ITAM, SMART: SM00408; IGc2, SMART: SM00077; ITAM

Expression pattern for human:

Tissue	Exp. (%)	Clones
lymph node	40.61	1:125
B-cells	16.87	55:16554
B cells from Burkitt lymphoma	9.48	4:2143
normal head/neck tissue	7.84	2:1295
lymph	7.21	94:66155
leukocyte	3.94	7:9008
spleen	2.03	5:12529
osteoarthritic cartilage	1.68	1:3016
thymus, pooled	1.60	1:3169
lymph, T-cell	1.19	2:8536

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