Igβ deficiency

GENERAL INFORMATION

Description:

Ig# mutations is involved in a failure of B cell development and can provide valuable insight into the assembly and function of BCR. The majority of patients with the early onset of infection, panhypogammaglobulinemia, reduced or absent B cells (over 85%) are males with mutations in BTK. 5% of patients have defects in the μ H chain, #5, Igα or BLNK.

Alternative names:

- CD79bD
- Immunoglobulin-associated beta; IGB
- Immunoglobulin-associated b29 protein; B29

Classification:

- Deficiencies predominantly affecting antibody production
  - Agammaglobulinemia

Inheritance:

Autosomal recessive

OMIM:

- *147245 Cd79b antigen; CD79B

Cross references:

Phenotype related immunodeficiencies:

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

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

The patient had early onset of infection, profound hypogammaglobulinemia, and markedly reduced but not absent B cells.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Agammaglobulinemia, autosomal recessive, ORPHANET
- Agammaglobulinemia, eMedicine

Genetic:

- IDdiagnostics

Therapeutic options:

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

Research programs, clinical trials:

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

GENE INFORMATION

Names:

HUGO name: CD79B

Localization:

Reference sequences:

DNA: IDRefSeq: D0126 (IDRefSeq) , **cDNA:** BC030210 (EMBL) , **Protein:** Q6PIS4 (SWISSPROT)

Chromosomal Location:

17q23

Maps:

CD79B (Map View)

Variations / Mutations:

- CD79Bbase; Mutation registry for Ig# deficiency

Other gene-based resources:

Ensembl: ENSG00000007312, GENATLAS: CD79B, GeneCard: CD79B, UniGene: 89575, Entrez Gene: 89575, euGenes: 89575, GDB: 133786, HomoloGene: 521

PROTEIN INFORMATION

Description:

Other features:

Expression pattern for human:

Tissue	Exp. (%)	Clones
tonsil	42.45	25:17800
lymph	23.64	34:43471
lymph_node	15.46	41:80139
spleen	5.54	9:49068
pituitary_gland	2.07	1:14599
uncharacterized_tissue	1.83	13:214464
mixed	1.68	16:287479
thymus	1.19	3:76122
blood	1.14	3:79803
prostate	1.03	4:117863

Animal models:

Mouse:

MGD: ; Cd79b

OTHER RESOURCES

Societies:

General:

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

Other information sources:

- Immunodeficiencies