ICOS deficiency

GENERAL INFORMATION

Description:

ICOS deficiency it is a form of common variable immunodeficiency caused by mutation in the ICOS gene, the "inducible costimulator". ICOS has critical involvement in T cell help for late B cell differentiation, class-switching and memory B cell generation.

Alternative names:

- ICOSD
- Inducible costimulator deficiency

Classification:

- Deficiencies predominantly affecting antibody production
  - Common variable immunodeficiency

Inheritance:

Autosomal recessive

OMIM:

- #607594 ICOS deficiency
- %240500 Common variable immunodeficiency
- *604558 Inducible costimulator; ICOS

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Common variable immunodeficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

The ICOS-deficient patients have recurrent bacterial infections of the respiratory and digestive tracts characteristic of humoral immunodeficiency. They lack other complicating features of CVID such as splenomegaly, autoimmune phenomena, or sarcoid-like granulomas and do not present with clinical signs of overt T cell immunodeficiency. ICOS deficient patients develop an adult-onset immunodeficiency characterised by low number of B cells, lack of memory cells and low serum Ig.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Common Variable Immunodeficiency (CVID), eMedicine

Genetic:

- ICOS, IDdiagnostics

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:

- European Initiative for Primary Immunodeficiencies
- Immune System and Gut Abnormalities in Patients with Common Variable Immunodeficiency with and without Gastrointestinal Symptoms, ClinicalTrials.gov
- Improved Healthcare for Patients with Primary Antibody Deficiencies through new Strategies Elucidating their Pathophysiology (IMPAD), IMPAD
- The Genetics of IgA Deficiency and Common Variable Immune Deficiency, Comprehensive Cancer Center, University of Albama at Birmingham, USA
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name: ICOS
Alias(es): AILIM, MGC39850, Activation-inducible lymphocyte immunomediatory molecule, Inducible T-cell co-stimulator, AILIM precursor

Localization:

Reference sequences:

DNA: AF411058 (EMBL) , **cDNA:** AB023135 (GenBank) , **Protein:** Q9Y6W8 (SWISSPROT)

Chromosomal Location:

2q33

Maps:

ICOS (Map View)

Markers:

RH103964, G54507

Variations / Mutations:

- ICOSbase; Mutation registry for ICOS deficiency

Other gene-based resources:

Ensembl: ENSG00000163600, GENATLAS: ICOS, GeneCard: ICOS, UniGene: 56247, Entrez Gene: 29851, euGenes: 29851, GDB: 10450295, HomoloGene: 8097

PROTEIN INFORMATION

Description:

Other features:

Signal peptide: 1-20

Other related resourses:

PIR: S78540, InterPro: IPR007110; Ig-like

Expression pattern for human:

Tissue	Exp. (%)	Clones
leukocyte	66.01	3:8838
moderately-differentiated endometrial adenocarcinoma, 3 pooled tumors	15.97	1:12174
aveolar macrophage	14.83	1:13110
mixed	2.30	1:84528
unclassified	0.88	4:879462
leukocyte	66.01	3:8838
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mixed	2.30	1:84528
unclassified	0.88	4:879462

Animal models:

Mouse:

MGD: ; Icos, NCBI Gene: ; 54167 (69.70 % aminoacid similarity to human)

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