

CD64 deficiency

GENERAL INFORMATION

Description:

Lack of phagocyte expression of CD64 leads to CD64 deficiency. The monocytes of these patients are unable to support mouse IgG2a anti-CD3-induced T cell mitogenesis.

Classification:

- Defects of phagocyte function

Inheritance:

OMIM:

- *146760 Fc fragment of IgG, high affinity Ia, receptor for; FCGR1A

Incidence:

Incidence unknown.

CLINICAL INFORMATION

Description:

Patients were apparently healthy, suggesting that FCGR1 is dispensable for phagocyte functioning. Some patients had cancer.

Therapeutic options:

-

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies 2001-2004, coord.Edvard Smith.

GENE INFORMATION

Names:

HUGO name: FCGR1A

Alias(es): CD64 , FCRI , IGFR1, Fc fragment of IgG, high affinity Ia, receptor for, Fc-gamma receptor I A1 , High affinity immunoglobulin gamma Fc receptor I precursor

Localization:

Reference sequences:

DNA: X14356 (EMBL) X14355 (EMBL) L03418 (EMBL) M91555 (EMBL) , **cDNA:** X58957 (EMBL) , **Protein:** P12314 (SWISSPROT)
Other Sequences

Chromosomal Location:

1q21.2-q21.3

Maps:

FCGR1A (Map View)

Markers:

D1S3356, RH69162

Variations / Mutations:

- FCGR1Abase; Mutation registry for CD64 deficiency

Other gene-based resources:

Ensembl: ENSG00000150337, GENATLAS: FCGR1A, GeneCard: FCGR1A, UniGene: 3427836, Entrez Gene: 2209, euGenes: 2209, GDB: 135911, HomoloGene: 475

PROTEIN INFORMATION

Description:

Protein function:

Binds to the Fc region of Immunoglobulins gamma. High affinity receptor.

Subcellular location:

Type I membrane protein.

Tissue specificity:

Monocyte/macrophage specific.

Similarity:

Contains 3 Immunoglobulin-like C2-type domains.

Database:

Name=prow; note=cd guide cd64 entry;

Domains:

Extracellular domain: 17-292

Cytoplasmic domain: 314-374

Ig-like C2-type 1 domain: 22-101

Ig-like C2-type 2 domain: 95-184

Ig-like C2-type 3 domain: 190-277

Other features:

Signal peptide: 1-16

High affinity immunoglobulin gamma fc receptor i: 17-374

N-linked (glcnac...) glycosylation sites:

59,78,152,159,163,195,240

Disulfide bonds: 43-85, 124-168, 212-260

Other related resources:

PIR: A39878, PIR: A41357, InterPro: IPR007110; Ig-like, InterPro: IPR003598; Ig_c2, Pfam: PF00047; ig, SMART: SM00408; IGc2, PROSITE: PS50835; IG_LIKE

Expression pattern for human:

Tissue	Exp. (%)	Clones
vascular	86.00	1:18887
brain	10.17	2:319574
other	3.83	1:423795
vascular	86.00	1:18887
brain	10.17	2:319574
other	3.83	1:423795

Animal models:

Mouse:

MGD: ; Fcgr1, NCBI Gene: ; 14129 (70.88 % aminoacid similarity to human)

Rat:

NCBI Gene: ; 295279 (66.77 % aminoacid similarity to human)

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- European Society for Immunodeficiencies