

κ light-chain deficiency

GENERAL INFORMATION

Description:

κ light-chain deficiency is produced by mutations in IGKC located on chromosome 2p11. The transmittance is autosomal recessive and have been point mutations described in one family. Pathogenesis include failure to express kappa chains but the reason is not known.

Alternative names:

- Kappa-chain deficiency

Classification:

- Deficiencies predominantly affecting antibody production
 - Light-chain deficiency

Inheritance:

Autosomal recessive

OMIM:

- +147200 Immunoglobulin kappa constant region; IGKC

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Kappa deficiency has little if any effect on health, gamma chains seem to be able to substitute effectively. The symptoms can vary including recurrent respiratory infections and diarrhea.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Agammaglobulinemia, eMedicine

Therapeutic options:

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

Alias(es): HCAK1, Ig kappa chain C region

Localization:**Reference sequences:**

DNA: J00241 (EMBL) V00557 (EMBL) ,
cDNA: , **cDNA:** AB004304 (GeneBank) ,
Protein: (SWISSPROT)

Chromosomal Location:

2p12

Maps:

IGKC (Map View)

Other gene-based resources:

Ensembl: ENSG00000163245, GENATLAS:
 IGKC, GeneCard: IGKC, UniGene: 259236,
 Entrez Gene: 3514, euGenes: 3514, GDB:
 120088, IMGT: IGKC

PROTEIN INFORMATION**Description:****Other features:****Other related resources:**

PIR: K3HU, InterPro: IPR003006; Ig_MHC,
 InterPro: IPR003597; Ig_c1, Pfam: PF00047;
 ig, SMART: SM00407; IGc1, PROSITE:
 PS00290; IG_MHC

Expression pattern for human:

Tissue	Exp. (%)	Clones
ovary, surface epithelium	18.76	5:38
brain, corpus collosum	17.82	1:8
breast, normal duct	13.60	33:346
prostate, stroma	12.05	24:284
lymph node	4.56	4:125
lymph node, pool of 10	3.87	11:405
lymphomas		
adipose	2.52	15:849
head/neck	2.46	9:522
nasopharynx	2.21	10:646
B-cells	1.91	221:16533

Animal models:**Mouse:**

MGD: ; Igk-C

R.norvegicus :

NCBI: ; Igk-C

OTHER RESOURCES**Societies:****General:**

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