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Gene Section
Mini Review

CMKOR1 (chemokine orphan receptor 1)

Karin Broberg

Department of Occupational and Environmental Medicine, Lund University Hospital, Lund, Sweden (KB)

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Identity

Other names: CMKOR1; RDC1; GPRN159; G protein-coupled receptor; chemokine orphan receptor 1; G protein-coupled receptor RDC1 homolog

HGNC (Hugo): CXCR7

Location: 2q37.3

Local order: Telomeric to IQCA. Centromeric to COPS8.

Note: RDC1 was originally thought to be the receptor for VIP.

DNA/RNA

Description

The genomic size has been estimated to approximately 12.5-13.5 kb. RDC1 has previously been reported to contain only one exon of 1,09 kbp. However, the finding of a RDC1 transcript corresponding to four different regions with exon/intron boundaries in the BAC 514f21 suggests a more complex gene structure. The predicted amino acid sequence of exon 3 and 4 does not show any homology to the protein databases and, since they both contribute with stop codons, it could be questioned whether these sequences represent exons, or are part of an alternatively spliced 3' untranslated region of the gene.

Pseudogene

None.

Protein

Description

362 amino acids; 41522 Da.

Expression

RDC1 is expressed in embryological, juvenile as well as adult tissues. Expression has been reported in e.g. bladder, spleen, heart, skeletal muscle, peripheral nervous system and placenta.

Localisation

Integral membrane protein.

Function

Orphan receptor, but its endogenous ligand has not yet been identified. The protein is also a coreceptor for human immunodeficiency viruses (HIV). RDC1 belongs to a family of G-protein coupled receptors, which includes hormone, neurotransmitter and light receptors, all of which transduce extracellular signals through interaction with guanine nucleotide (G) binding proteins.

Homology

RDC1 displays homology to other members of the large family of G-protein coupled receptors.

Mutations

Germinal

Single nucleotide polymorphisms.

Somatic

Translocations involving RDC1 and HMGA2 has been reported in three lipomas (see below).

Implicated in

Lipoma

Disease

Benign adipocyte tumor.

Prognosis

Good.

Cytogenetics

Translocations involving 2q35-37 and 12q13-15 have been reported in six lipomas.

Hybrid/Mutated gene

Fusion between RDC1 and HMGA2 has been reported in three lipomas with rearrangement involving 2q35-37 and 12q13-15. The breakpoint occurred after the third exon of HMGA2, the most common breakpoint of this gene, and in a previously unknown 3' part of the RDC1 gene. The RDC1 part of the fusion was over 300 bp.

Abnormal protein

The functional impact of this fusion is most likely a truncation of HMGA2, since the RDC1 part contributes with a stop codon one amino acid downstream of the breakpoint.

Oncogenesis

Not yet established.

Tenosynovial giant cell tumours

Disease

Benign tumor of synovium and tendon sheath.

Prognosis

Good.

Cytogenetics

Translocations involving 1p11-13 and 2q35-37 have been reported in eight cases of tenosynovial giant cell tumours.

Hybrid/Mutated gene

Four out of seven cases of tenosynovial giant cell tumours with aberrations of 2q35-37 had breakpoints in a BAC probe 260J21 (BACPAC, Oakland), which contains the RDC1 gene.

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