

Gene Section

Mini Review

CLTCL1 (clathrin heavy polypeptide-like 1)

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Identity

Other names: CLTCL; CLTD; CLH-22

HGNC (Hugo): CLTCL1

Location: 22q11

Note

May have been confused with CLTC (clathrin heavy chain gene), a partner gene of ALK.

DNA/RNA

Transcription

5564 bp mRNA

Protein

Description

1640 amino acids, 187 kDa; is composed, from N-term to C-term, of: a globular domain (amino acids 1-479), a linker (480-523), and the heavy chain arm (524-1640); properties: binding site for ATPase in N term, binding of the light chain in the C-term, and trimerization domain in the C-term. Subunit of clathrin, a structural protein composed of 3 heavy chains (CLTC, CLTCL1), and 2 light chains (CLTA, CLTB), which assembly is mediated by CALM. Form cages. Component of the vesicles matrix originated from the plasma membrane or the Golgi

Localisation

Vesicles.

Function

Mediate endocytosis of transmembrane receptors.

Implicated in

? t(2;22)(p23;q11.2)- ALK

Note

CLTCL1/ALK may in fact be cases of CLTC/ALK

Disease

? found in a case of ALK+ anaplastic large cell lymphoma.

Abnormal protein

? 2197 amino acids, 248-250 kDa; 1634 (nearly all the CLTCL1 protein) N-term amino acids from CLTCL1, fused to the 562 C-term amino acids from ALK (i.e. the entire cytoplasmic portion of ALK with the tyrosine kinase domain); cytoplasmic localization restricted to granules.

Oncogenesis

Constitutive autophosphorylation.

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