

Gene Section

Mini Review

CLTCL1 (clathrin heavy polypeptide-like 1)

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Identity

Other names: CLTCL; CLTD; CLH-22

Location: 22q11

Note: Must not be confused with CLTC (clathrin heavy chain gene), inasmuch as both are involved in translocations with 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.

Expression

Vesicles.

Function

Mediates endocytosis of transmembrane receptors.

Implicated in

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