CLTC (clathrin heavy polypeptide)

Identity

Note	must not be confused with CLTCL1 (clathrin heavy polypeptide-like 1).
Other names	**clathrin heavy chain**
	KIAAOO34
	CLH-17
Hugo	**CLTC**
Location	17q23

DNA/RNA

Transcription 32 exons, 6111 bp mRNA

Protein

Description	Clathrin is the major protein constituent of the coat that surrounds organelles (cytoplasmic vesicles) to mediate selective protein transport. Clathrin coats are involved in receptor-mediated endocytosis and intracellular trafficking and recycling of receptors, which accounts for its characteristic punctate cytoplasmic and perinuclear cellular distribution. Structurally, clathrin is a triskelion (three-legged) shaped protein complex that is composed of a trimer of heavy chains (CLTC) each bound to a single light chain. CLTC is a 1675 amino acid residue protein encoded by a gene consisting of 32 exons. Its known domains include a N-terminal globular domain (residues 1-494) that interacts with adaptor proteins (AP-1, AP-2, b-arrestin), a light chain-binding region (residues 1074-1552), and a trimerization domain (residues 1550-1600) near the C-terminus.
Localisation	Cytoplasmic vesicles
Function	mediate endocytosis of transmembrane receptors.

Implicated in

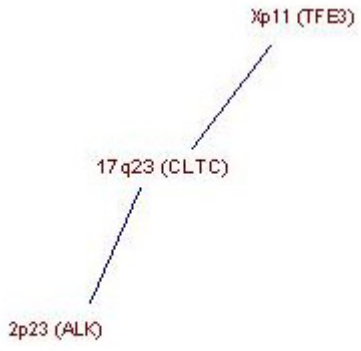
Entity	Anaplasic large cell lymphoma (ALCL) with t(2;17)(p23;q23) --> ALK - CLTC
Disease	ALCL are high grade non Hodgkin lymphomas; ALK+ ALCL are ALCL where ALK is involved in a fusion gene; ALK+ ALCL represent 50 to 60 % of ALCL cases (they are CD30+, ALK+;); belong to the "cytoplasmic ALK+" subset.
Prognosis	Although presenting as a high grade tumour, a 80% five yr survival is associated with this anomaly

Hybrid/Mutated Gene	5' CLTC - 3' ALK
Abnormal Protein	NH2 CLTC - COOH ALK

Entity	Inflammatory myofibroblastic tumors with t(2;17)(p23;q23)
Note	In these tumors, the fusion point in CLTC is identical, being at amino acid 1634 (corresponding to the 3' end of exon 31 of CLTC), such that almost all of CLTC is included in the fusion protein, including its trimerization domain . As a fusion partner, CLTC has been postulated to provide CLTC-ALK with deregulated expression driven by its constitutively activated promoter and constitutive oligomerization of the chimeric protein via the CLTC multimerization domains normally used for clathrin coat assembly. Since ALK is a tyrosine kinase that is activated by cross-phosphorylation following ligand binding, CLTC-ALK-induced oligomerization may result in a constitutively activated ALK tyrosine kinase domain. In this sense, CLTC is likely to function in CLTC-ALK as other prototypical "dimerizing translocation partners" in fusions involving tyrosine kinase genes.
Disease	rare soft tissue tumour found in children and young adults
Prognosis	good prognosis
Hybrid/Mutated Gene	5' CLTC - 3' ALK

Entity	Xp11 renal translocation carcinoma with t(X;17)(p11;q23)
Note	In the CLTC-TFE3 fusion, the fusion point on CLTC is at amino acid 932 (corresponding to the end of exon 17), thereby excluding the CLTC trimerization domain from the predicted fusion protein. As in other TFE3 gene fusions, the nuclear localization and DNA binding domains of TFE3 are retained in CLTC-TFE3. Based on these features and existing data on other TFE3 fusion proteins, CLTC-TFE3 may act as an aberrant transcription factor, with the CLTC promoter driving constitutive expression.
Disease	rare renal carcinoma (single case report)
Prognosis	Unknown prognosis
Hybrid/Mutated Gene	5' CLTC 3[1]TFE3

Breakpoints



CLTC and partners. Editor 06/2005; last update 08/2005.

External links

	Nomenclature
Hugo	CLTC
GDB	CLTC
Entrez_Gene	CLTC 1213 clathrin, heavy polypeptide (Hc)
	Cards
Atlas	**CLTCID360**
GeneCards	CLTC
Ensembl	CLTC
CancerGene	CLTC
Genatlas	CLTC
GeneLynx	CLTC
eGenome	CLTC
euGene	1213
	Genomic and cartography
GoldenPath	CLTC - 17q23 chr17:55052038-55127254 + 17q23.2 (hg17-May_2004)
Ensembl	CLTC - 17q23.2 [CytoView]
NCBI	Genes Cyto Gene Seq [Map View - NCBI]
OMIM	Disease map [OMIM]
HomoloGene	CLTC
	Gene and transcription
Genbank	AK127134 [SRS] AK127134 [ENTREZ]
Genbank	BC015854 [SRS] BC015854 [ENTREZ]
Genbank	BC036430 [SRS] BC036430 [ENTREZ]
Genbank	BC051800 [SRS] BC051800 [ENTREZ]
Genbank	BC054489 [SRS] BC054489 [ENTREZ]
RefSeq	NM_004859 [SRS] NM_004859 [ENTREZ]

RefSeq	NT_086883 [SRS] NT_086883 [ENTREZ]
AceView	CLTC AceView - NCBI
TRASER	CLTC Traser - Stanford
Unigene	Hs.491351 [SRS] Hs.491351 [NCBI] HS491351 [spliceNest]
	Protein : pattern, domain, 3D structure
SwissProt	Q00610 [SRS] Q00610 [EXPASY] Q00610 [INTERPRO]
Interpro	IPR008938 ARM [SRS] IPR008938 ARM [EBI]
Interpro	IPR001473 Clathrin_propl_N [SRS] IPR001473 Clathrin_propl_N [EBI]
Interpro	IPR000547 Clathrin_repeat [SRS] IPR000547 Clathrin_repeat [EBI]
Interpro	IPR008941 TPR-like [SRS] IPR008941 TPR-like [EBI]
CluSTr	Q00610
Pfam	PF00637 Clathrin [SRS] PF00637 Clathrin [Sanger] pfam00637 [NCBI-CDD]
Pfam	PF01394 Clathrin_propel [SRS] PF01394 Clathrin_propel [Sanger] pfam01394 [NCBI-CDD]
Smart	SM00299 CLH [EMBL]
Blocks	Q00610
	Polymorphism : SNP, mutations, diseases
OMIM	118955 [map]
GENECLINICS	118955
SNP	CLTC [dbSNP-NCBI]
SNP	NM_004859 [SNP-NCI]
SNP	CLTC [GeneSNPs - Utah] CLTC [SNP - CSHL] CLTC] [HGBASE - SRS]
	General knowledge
Family Browser	CLTC [UCSC Family Browser]
SOURCE	NM_004859
SMD	Hs.491351
SAGE	Hs.491351
Amigo	function|binding
Amigo	component|clathrin vesicle coat
Amigo	component|coated pit
Amigo	process|intracellular protein transport
Amigo	function|structural molecule activity
PubGene	CLTC
	Other databases
	Probes
Probe	CLTC Related clones (RZPD - Berlin)

PubMed

PubMed **9 Pubmed reference(s) in LocusLink**

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