t(2;17)(p23;q23)

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Clinics and pathology

Disease
Inflammatory myofibroblastic tumors.

Clinics
Rare soft tissue tumor found in children and young adults.

Pathology
Spindle cell proliferation with myofibroblastic differentiation and an inflammatory infiltrate.

Prognosis
Low malignant potential and good prognosis.

Genes involved and proteins

ALK
Location
2p23
Protein
1620 amino acids; 177 kDa; glycoprotein (200 kDa mature protein) ; membrane associated tyrosine kinase receptor.

CLTC (clathrin heavy polypeptide)
Location
17q23
Protein
1675 amino acids, 191 kDa; mediate endocytosis of transmembrane receptors.

Result of the chromosomal anomaly

Hybrid Gene
Description
5' CLTC - 3' ALK

Fusion Protein
Description
2196 amino acids. 1634 N-term amino acid from CLTC, containing nearly all of CLTC, including the triskelion assembly domain, fused to the 562 C-term amino acids from ALK (i.e. the entire cytoplasmic portion of ALK with the tyrosine kinase domain).

References


This article should be referenced as such: