The protein encoded by this gene belongs to the Kank family of proteins, which contain multiple ankyrin repeat domains. This family member functions in cytoskeleton formation by regulating actin polymerization. This gene is a candidate tumor suppressor for renal cell carcinoma. Mutations in this gene cause cerebral palsy spastic quadriplegic type 2, a central nervous system development disorder. A t(5;9) translocation results in fusion of the platelet-derived growth factor receptor beta gene (PDGFRB) on chromosome 5 with this gene in a myeloproliferative neoplasm featuring severe thrombocythemia. Alternative splicing of this gene results in multiple transcript variants. A related pseudogene has been identified on chromosome 20. [provided by RefSeq, Dec 2014]

GeneCards Summary for KANK1 Gene

KANK1 (KN Motif And Ankyrin Repeat Domains 1) is a Protein Coding gene. Diseases associated with KANK1 include Cerebral Palsy, Spastic Quadriplegic, 2 and Inherited Congenital Spastic Tetraplegia. Gene Ontology (GO) annotations related to this gene include beta-catenin binding. An important paralog of this gene is KANK2.