Summary

Filippi syndrome is classified in the group of craniodigital syndromes. The patients have short stature, microcephaly, intellectual disability, developmental delay, characteristic face and syndactyly of fingers and toes as the prominent phenotypes. It is inherited as an autosomal recessive fashion with mutations known only in the *CKAP2L* gene. I ascertained a Filippi syndrome family from Italy and identified a de novo mutation, NM_001320.5;c.94G>C;p.Asp32His, in the *CSNK2B* gene, which I proposed as a second and novel gene of this disorder. *CSNK2B* encodes CK2β, which is the regulatory subunit of protein kinase CK2. This holoenzyme is a heterotetramer of the following compositions — αββα, αββα′ or α′ββα′, where two α and/or α′ subunits are structured around the obligate β dimers. To investigate the consequences of the mutation, lymphoblastoid cell lines (LCLs) were generated from patient and control. Initially, I studied the consequences of the identified mutation at the RNA level and revealed that the amount of mutant *CSNK2B* transcript was higher compared to control. This data was further corroborated by detecting increased levels of CK2β protein in the patient compared to control LCLs. Additionally, this mutation impairs its binding to CK2α compared to wild-type CK2β. At the cellular level, alterations in the localization of CK2β were not detected in the mutant LCLs. Rather, an altered Golgi morphology was observed in the patient LCLs compared to wild-type. I propose that an altered level of CK2β may impair the embryogenesis and produce drastic effects during development.