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Andermann Syndrome in a Pakistani Family Caused by a Novel Mutation in SLC12A6.

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Abstract

Agenesis of the corpus callosum with peripheral neuropathy (ACCPN) or Andermann syndrome is an autosomal recessive condition caused by mutations in *SLC12A6*. The neurodegenerative features are characterized primarily by severe and progressive polyneuropathy, with eventual loss of ambulation and limited lifespan. We report two siblings with Andermann syndrome from a consanguineous Pakistani family with severe global developmental delays, sensory-motor polyneuropathy, and complete agenesis of the corpus callosum, associated with a homozygous c.745+2T>A mutation in *SLC12A6*. This sequence change is predicted to inactivate the donor splice site and abolish correct splicing of intron 6, yielding an abnormally truncated protein. This is the first report of Andermann syndrome in patients of Pakistani origin, which supports the pan-ethnic incidence of this condition.