A 38-year-old man was diagnosed, at the age of 18, with SCN4A-negative hyperkalaemic periodic paralysis. The diagnosis remained unchanged until his 8-year-old daughter suffered an exercise-induced syncope. Her EKG showed a polymorphic ventricular tachycardia. Patient’s hands and feet, previously overlooked, became “neurologically” relevant since they were characteristic of Andersen-Tawil syndrome (Figure). A pathogenic KCNJ2 mutation (Arg218Trp) was found.

Andersen-Tawil syndrome is an autosomal dominant disorder characterized by the triad of periodic paralysis, ventricular arrhythmias, and dysmorphic features. Phenotypical heterogeneity, even within a family, often delays the diagnosis which is necessary since cardiac assessment is warrant.

References
