DOI: 10.1590/0004-282X20140168

Clinodactyly and syndactyly – diagnostic clues for Andersen-Tawil syndrome

Clinodactilia e sindactilia – pistas diagnósticas da síndrome de Andersen-Tawil

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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[1]. Phenotypical heterogeneity, even within a family, often delays the diagnose which is necessary since cardiac assessment is warrant[2].

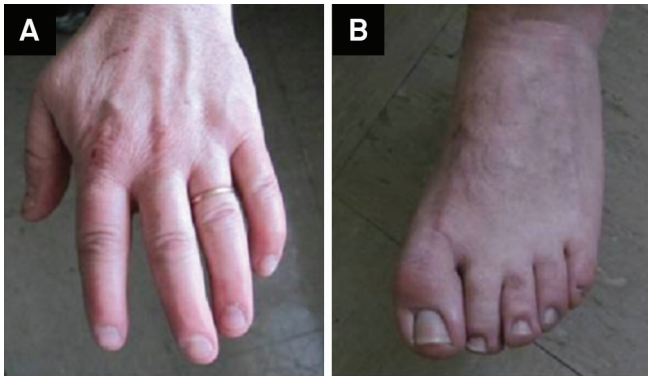

Figure. (A) Fifth digit clinodactyly and (B) syndactyly of the toes 2 and 3, highly suggestive of Andersen-Tawil syndrome. The face (not shown) had only mild phenotypical characteristics.

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Conflict of interest: There is no conflict of interest to declare.

Received 14 June 2014; Received in final form 31 July 2014; Accepted 19 August 2014.