

Life-changing effect of L-Carnitine in a patient with KAT6A syndrome

Press release

Peter, was diagnosed with severe autism and global developmental delay at the age of 18 months. It wasn't till the age of 8 that he was diagnosed with KAT6A gene mutation, he was the 39th known patient in the world. Shortly after this diagnosis, we consulted Dr Richard Kelley, a World renown genetic and metabolic specialist, who, after checking his metabolic profile, recommended to start Peter on a mitochondrial cocktail that included L-Carnitine, B complex, and other supplements.

The effect of the cocktail was miraculous. Peter became potty-trained, he started riding his bike with no training wheels and playing soccer. Peter, who was non-verbal, started imitating sounds and then started saying few words.

Further research and a better understanding of the effect of the mitochondrial cocktail in the setting of a KAT6A gene mutation will help children and adults affected with the KAT6A syndrome.

By Natacha Esber and Paul Najm
Mother and brother of a child affected by KAT6A