GAD ANTIBODIES NEGATIVE TYPE 1 DIABETES AND DRAVET SYNDROME

Sara Ciccone¹, Romana Marini¹, Lucia Fusco², Alessandra Terracciano³, Riccardo Schiaffini¹, Marco Cappa¹

¹Endocrinology and Diabetes Unit, Bambino Gesù Children’s Hospital, IRCCS, Rome, Italy
²Neurology Unit, Bambino Gesù Children’s Hospital, IRCCS, Rome, Italy
³Unit of Molecular Medicine for Neuromuscular and Neurodegenerative Diseases, Department of Neurosciences, Bambino Gesù Children’s Hospital, IRCCS, Rome, Italy

• An association between T1DM and idiopathic generalized epilepsy is reported. Some authors suggest an autoimmune mechanism mediated by antibodies to glutamic-acid-decarboxylase (GAD), that is an enzyme involved in the synthesis of the neurotransmitter GABA.
• Dravet syndrome (DS) is a rare, severe epilepsy disorder characterized by febrile hemiconic seizures or generalized status epilepticus starting at 6 months of age. In classical DS, a delayed development and a motor impairment are often described. Mutation or deletions of SCN1A account for 85% of DS cases. SCN1a mutations alter sodium channel activity that can predispose the SNC to abnormal excitability.
• We report the case of a 9-year-old boy with T1DM and DS.
• No familial history of epilepsy or diabetes. First-born at the 37th week from a normal pregnancy, with a normal adaptation at birth.
• At 8 months, he developed febrile seizures, then at 2.5 years he presented afebrile generalized tonic-clonic seizures. A DRAVET SYNDROME was clinically diagnosed, confirmed by a positive test for a SCN1A gene mutation (heterozygous c.560_563inv). Epilepsy has proved to be drug-resistant (valproate, gardenal, topiramate, levetiracetam and then stiripentol). A mild improvement of seizures was reported with stiripentol treatment.
• At the age of 7, the boy developed a T1DM.

CASE REPORT

• A concordance between GAD-antibody titres and clinical manifestations of myoclonic encephalopathy was reported in some patients, in whom a pathogenetic role of GAD autoimmunity was suggested.
• In the presented case, we can hypothesize an autoimmune etiology but not GAD-antibodies mediated.

REFERENCES

Saturday, 3 October

Diabetes - P3-721