54th Annual Meeting 1-3 October
ESPE 2015 BARCELONA
European Society for Paediatric Endocrinology

GAD ANTIBODIES NEGATIVE TYPE 1 DIABETES AND DRAVET SYNDROME

Sara Ciccone[1], Romana Marini[1], Lucia Fusco[2], Alessandra Terracciano[3], Riccardo Schiaffini[1], Marco Cappa[1]

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

BACKGROUND

- 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 hemiclonic 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.

CASE REPORT

- 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**.

Serum glucose 536 mg/dl, HbA1c 86 mmol/mol (n.v.20-38), venous pH 7,29, HCO3- 10.7 mmol/l, BE -17,1 mmol/l;

PHOSPHOTYROSINE ANTIBODIES + ANTI-GAD AND ANTI-INSULIN ANTIBODIES - (CONFIRMED AFTER 2 YEARS)

CONCLUSION

- 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

1. Lenti C, Bognetti E, Bonfanti R, Bonifacio E, Meschi F. Myoclonic encephalopathy and diabetes mellitus in a boy. Dev Med Child Neurol. 1999 Jul;41(7):489-90.
2. Yun C, Xuefeng W. Association between seizures and diabetes mellitus: a comprehensive review of literature. Curr Diabetes Rev. 2013 Jul;9(4):350-4.
3. Verrotti A, Scaparrotta A, Olivieri C, Chiarelli F. Seizures and type 1 diabetes mellitus: current state of knowledge. Eur J Endocrinol. 2012 Dec;167(6):749-58
4. McKnight K, Jiang Y, Hart Y, Cavey A, Wroe S, Blank M, Shoenfeld Y, Vincent A, Palace J, Lang B. Serum antibodies in epilepsy and seizure-associated disorders. Neurology. 2005 Dec 13;65(11):1730-6.
5. Vincent A, Crino PB. Systemic and neurologic autoimmune disorders associated with seizures or epilepsy. Epilepsia. 2011 May;52 Suppl 3:12-7.

Saturday, 3 October **Diabetes - P3-721**

ESPE 2015 721-P3
Diabetes 1
Romana Marini
DOI: 10.3252/pso.eu.54espe.2015
Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology
PosterSessionOnline.com