CSII MANAGEMENT IN AN INFANT WITH DEND SYNDROME

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Aim:

Some *KCNJ11* gene mutations result in DEND syndrome (Developmental delay, Epilepsy, Neonatal Diabetes). The diabetes management of the KCNJ11 mutations with severe seizures and insensitivity to glibenclamide is troublesome.

Case:

A 7 days old boy was referred with hyperglycemia

History: Term/2340gr/vaginal delivery

No consanguinity

At presentation, glucose: 202 mg/dl, insulin: 0.5 mIU/ml, autoantibodies: (-)

NPH was started

First genetic report showed no mutation in ABCC8 and KCNJ11 genes.

Infantile spasms were noticed when the patient was switched from MDI to CSII at 4 months of age.

The electroencephalogram (EEG) was showed hypsarrythmia and suppression bursts.

The seizures were refractory to several antiepileptics (phenobarbital, levetiracetam, vigabatrin) and his neurological condition deteriorated slowly.

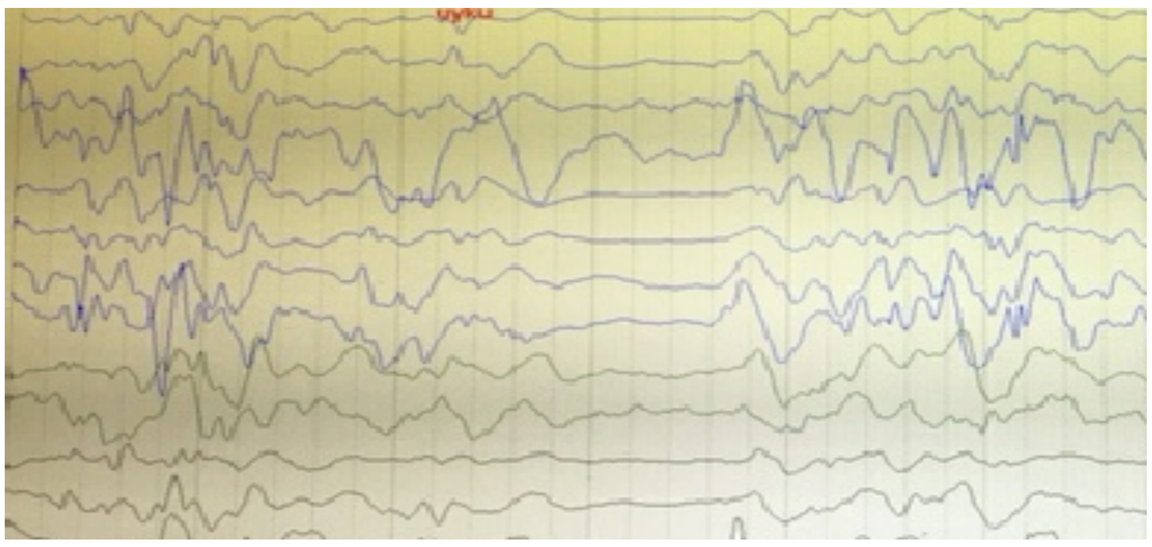
The patient had unpredictable hypo-hyperglycemia due to contractions and swallowing / feeding problems.

During this period, the CSII overcame dysglycemia but the contractions due to seizures and physiotherapy caused frequent infusion blockage. Massage or set change was performed to cope with this problem.

Glibenclamide (max 2 mg/kg/d) was administered at 10 months of age, since a known denovo KCNJ11 gene mutation (c.497G>T) was detected at NGS. However, he didn't respond the sulphonylurea.

During follow-up, HbA1c was between %7.0-8.3

Currently, he is 3 years old and has severe mental-motor retardation. The convulsions are still uncontrolled.



Conclusion:

CSII is the most physiologic way for a good metabolic control in diabetes. However, CSII in diabetic patients with neurologic problems can be quite troublesome.