CASE REPORT OF A CHILD WITH POIRIER-BIENVENU NEURODEVELOPMENTAL SYNDROME

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Background

Neurological syndrome known as Poirier-Bienvenu neurodevelopmental syndrome (POBINDS) is characterized by early onset of epileptic seizures and delayed psychomotor development. It is a rare disease associated with mutations in the CSNK2B gene. This gene encodes the beta subunit of casein kinase CK2, which is involved in numerous processes and regulates metabolic pathways, signal transduction, transcription, translation and replication.

Case report

A firstborn boy from a first pregnancy that proceeded normally. Delivery at 41 weeks, vaginal, birth weight 4050 g, birth length 50 cm, Apgar score 10/10. At the age of 6 months, he manifested four generalized epileptic seizures. Hospitalized at the Children's Hospital Zagreb, where diagnostic workup was performed: neuroimaging methods CT and MRI of the brain were normal, laboratory and metabolic workup showed no abnormalities, lumbar puncture findings were normal. Genetic testing (Blueprint panel epilepsy) identified a pathogenic mutation in the CSNK2B gene (c.139C>T, p.(Arg47*)), with an autosomal dominant inheritance pattern. The clinical picture varies; some individuals have frequent and severe epileptic seizures, severe motor deficits and difficulties in language and speech development, others may have seizures that respond to treatment and have normal psychomotor development. Some children have symptoms from the autism spectrum, dysmorphic facial features (triangular face, hypertelorism, sparse eyebrows). Sporadic cases have vascular abnormalities and growth hormone deficiency. In most, this is a de novo mutation.

Conclusion

Treatment is symptomatic, based on epilepsy management and stimulation of psychomotor development. Given the significant phenotypic variability, the approach to POBINDS patients must be individualized. Multidisciplinary care involves neuropediatricians, geneticists, child physiatrists, speech therapists, and psychologists and is essential for optimal outcomes. Early recognition and timely treatment improve developmental outcomes.

Keywords: Poirier-Bienvenu, neurodevelopmental, syndrome;, child;, multidisciplinary