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PROCEEDINGS

Multidisciplinary Treatment Care of a Patient With Microduplication Syndrome in the Background of Neurodevelopmental Delay

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BACKGROUND: We present a patient with microduplication syndrome who underwent multidisciplinary treatment care (MTC) at our department. Microduplication syndrome is characterized by distinctive craniofacial features that are mostly minor and as such often unrecognizable; intellectual disability; developmental delay; neurological and behavioral abnormalities; musculoskeletal problems; ascending aortic dilatation, and a peculiar higher threshold for pain perception.

AIM: To enhance the importance of genetic testing of children with multiple difficulties and malformations of organ systems.

METHOD: Our patient is the second child of nonconsanguineous healthy parents born at 39 weeks gestational age by spontaneous vaginal delivery from an orderly course pregnancy. Shortly after birth, she received phototherapy for neonatal jaundice. Due to early-onset neonatal sepsis, she was transferred to the intensive care unit (ICU). Pediatric physiatrist examination, at the age of 4 months besides hypotonia and asymmetry, assessed she did not meet age-appropriate developmental milestones. Her subtle phenotypic characteristics of craniofacial dysmorphism consistent with published literature, included brachycephaly, broad forehead, straight neat eyebrows, deep-set eyes, broad nasal tip, low columella insertion. Ultrasound brain findings verified the condition after intracranial bleeding grade II. Magnetic resonance imaging (MRI) of the brain revealed dysplasia of the corpus callosum genu and rostral body. Motor, speech, and social skills were prominently affected domains and therefore intensive comprehensive inpatient and outpatient MTC was pursued. Habilitation encompassed the Vojta principle due to chronic constipation and neurogenic bladder, which required intermittent catheterization. She has regularly been under cardiac surveillance due to ductus arteriosus persistens and ascendant aorta dilatation which is a common congenital malformation (46%). Throughout her infancy and toddlerhood difficulty feeding, expressive and receptive language skills, and selective mutism were observed.

RESULTS: MTC gradually optimized her communication skills by converting nonverbal behaviors into discrete words. She conquered wide-based independent ambulation at the age of 20 months with the persistence of balance and coordination disturbances. She had a history of frequent falls secondary to hypotonia. At the age of two chromosomal microarray (CMA) revealed a duplication at 7q11.23. DNA analysis discovered duplication that affects 36 genes, where genes ELN and NCF1 are considered responsible. It has been diagnosed worldwide in just over 150 patients with a prevalence estimated at 1:7500 to 1:20 000. Both parents tested negative for this duplication, confirming de novo change in the proband. The patient made gradual progress with therapies and the family was made aware of the diagnosis, prognosis, and future complications that might occur.

DISCUSSION AND CONCLUSION: Our experience and current knowledge about this syndrome indicate the necessity of genetic testing. In Croatia it is conditioned by the previous finding of neural axis MRI. Among the challenges in overall diagnostic evaluation, we would like to highlight the benefit of neurodevelopmental stimulation to minimize long-term sequelae within the underlying genetic disease. Our patient achieved optimal obtainable gross motor status in 5 years follow-up.

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