WILLIAMS-BEUREN SYNDROME SHROUDED IN NEURODEVELOPMENTAL DELAY, SEVERE SCOLIOSIS, AND „COCKTAIL PARTY PERSONALITY"

Jelena Marunica Karšaj[1], Kristina Žgela Talan[2], Valentina Matijević[1]

[1] Sestre milosrdnice University Hospital Center, Croatia
[2] Istrian Health Centres, Croatia
e-mail: jelenamarunica@yahoo.com

Background

Williams-Beuren syndrome (WBS) is a rare congenital multisystem condition caused by a spontaneous heterozygous microdeletion at 7q11.23. It occurs in 1:7.500-20.000 live births. The syndrome manifests through characteristic dysmorphic facial features, described as an elfin visage, alongside cardiovascular anomalies, cognitive impairments, as well as endocrine, ophthalmologic, and orthopedic abnormalities. The course of treatment is contingent upon the particular abnormality; however, the paramount objective universally remains the alleviation of symptoms and the prevention of potential complications.

Case report

Our patient is the first child of nonconsanguineous healthy parents born at 36 weeks gestational age via spontaneous vaginal delivery from an orderly course pregnancy. He underwent phototherapy to alleviate neonatal jaundice. Ultrasound brain findings verified subependimal intracranial bleeding with multiple cystic formations. Phenotypic characteristics revealed craniofacial dysmorphism, including epicanthal folds, large ears, an upturned nose, a wide mouth, a small jaw, and small teeth. His cardiac anomaly in terms of bicuspid aortal valve has been regularly under cardiac surveillance. He has been under pediatric physiatrist follow-up since the time he was two months old until six years due to neurodevelopmental delay, trunkal hypotonia and increased muscle tone on extremities, and severe dextroconvex thoracolumbal scoliosis with punctum maximum at L4 and pectus excavatum. He has been toe walking since he was 15 months old. He has a very outgoing personality and empathy, has difficulty identifying strangers and therefore fits the description „cocktail party personality". As gross and fine motor skills and cognitive functions were affected, comprehensive inpatient and outpatient multidisciplinary habilitation is pursued encompassing Bobath concept and Vojta principle. The clinical exome sequencing has unveiled a microdeletion at locus 7q11.23.

Conclusion

We present this case to underscore the benefits derived from the timely referral to pediatric physiatrist, aimed at optimally promoting gross motor development and facilitating the resolution of scoliosis in a child afflicted with WBS.

Keywords: Williams-Beuren syndrome, child, scoliosis, neurodevelopmental