Cognitive Impairment, Scoliosis and Renal Parenchymal Disease with Isolated Rhombencephalosynapsis: A Case Report

İzole Rombensefalosinapsis ile Birlikte Kognitif Bozukluk, Skolyoz ve Renal Parankimal Hastalık: Bir Olgu Sunumu

Şule BÜYÜK¹, Ebru ARHAN², Betül DERİN KUYU³, Şemsə ÇAYCI⁴, Esra GÜNEY⁵

¹Ankara Child Health and Diseases Hematology Oncology Training and Research Hospital, Ankara, Turkey
²Ankara Child Health and Diseases Hematology Oncology Training and Research Hospital, Department of Child Neurology, Ankara, Turkey
³Ankara Child Health and Diseases Hematology Oncology Training and Research Hospital, Department of Radiology, Ankara, Turkey
⁴Ankara Child Health and Diseases Hematology Oncology Training and Research Hospital, Department of Pediatric Nephrology, Ankara, Turkey
⁵Ankara Child Health and Diseases Hematology Oncology Training and Research Hospital, Department of Child Psychiatry, Ankara, Turkey

ABSTRACT

Rhombencephalosynapsis (RS) is an uncommon cerebellar malformation defined by vermian agenesis with fusion of the hemispheres and of the dentate nuclei. Very few cases have been reported in the literature. While the literature suggests that RS is often associated with behavioral and/or intellectual impairment, very few previous reports have described neuropsychological functioning. We report a 12-year-old male who was diagnosed with RS. The neurological examination revealed general hypotonia, brisk deep tendon reflexes, ataxic gait and dysarthria while neuropsychological evaluation revealed low verbal learning, attention and working memory subtest scores. He also had scoliosis and renal parenchymal disease, both conditions being reported very rarely in association with this anomaly. Our findings suggest that RS is associated with cognitive impairment and often accompanied by other abnormalities.

Key Words: Cerebellar abnormality, Intellectual disability, Kidney disease, Scoliosis

ÖZET


Anahtar Sözcükler: Serebeller anormalı, Bilişsel bozukluk, Böbrek hastalığı, Skolyoz

INTRODUCTION

Rhombencephalosynapsis is an uncommon cerebellar malformation characterised by vermian agenesis with fusion of the hemispheres and of the dentate nuclei. Embryologic and genetic mechanisms are still unknown. Clinical presentation is variable and primarily determined by the presence of systemic malformations. A number of pediatric and adult cases (less than 80) have been previously reported in the literature (1,2). Associated anomalies within and outside the central nervous system have been reported in some cases.

It is well known that the cerebellum is essential for the control and integration of movement and that cerebellar disorders often are associated with a motor syndrome that can include ataxia, dysmetria, disordered eye movements, dysarthria, dysphagia and tremor (3). In recent years it has been hypothesized that there is also a strong cerebellar contribution to cognition and mood.

This is one of the very few published reports of RS presenting with cognitive impairment, and rare anomalies outside the central nervous system.
CASE REPORT

A 12 years old boy was referred to our department with developmental delay and mental retardation. He was born at term following a normal pregnancy and delivery. He showed a consistent delay in acquisition of psychomotor skills (head control at 6 months, sitting alone at 12 months, walking at 3 years of age, speaking at 3.5 years of age). He experienced two simple febrile seizures when he was three years old. He had an operation for scoliosis at the age 3.5 years. He had recurrent urinary tract infections during infancy. Subsequent ultrasound study of the urinary tract revealed renal parenchymal disease with a mildly irregular renal contour. Further dimercapto succinic acid study revealed scarring of the left side of the single kidney. He has developed chronic renal insufficiency. In the ensuing years, he was found to have global developmental delay and cerebellar ataxia.

At admission, physical examination disclosed normal head circumference and cranio-facial dysmorphism: microcephaly (head circumference <3p), low set ears, hypertelorism, prominent forehead, protruding tongue and midface hypoplasia. On neurological examination, general hypotonia, brisk deep tendon reflexes in all four limbs, ataxic gait and dysarthria were noted. His social relations and behavioural capabilities were largely impaired, and he has developed no independence. The patient's Wechsler Intelligence Scale-revised showed full scale IQ fell within the moderately low range (WISC-R FSIQ = 40). The estimated verbal (VIQ = 38) and performance (PIQ = 42) IQs fell within the moderately low average range. Verbal learning, attention and working memory subtest scores assesses by Stanford Binet test were in low range. Cranial magnetic resonance imaging showed agenesis of the cerebellar vermis with the fusion of the cerebellar hemispheres (Figure 1). Fusion of the middle cerebellar hemispheres and probable fusion of the dentate nuclei along the midline were also present (Figure 2). He was referred to a special child care centre, where he received intensive training. There was a steady improvement and satisfactory progress in his development.

DISCUSSION

Cerebellar disorders often are associated with disturbances of movement. Many individuals with RS have motor impairments, albeit of varying range and severity (4). In recent years it has been hypothesized that there is also a strong cerebellar contribution to cognition and mood. Schmahmann suggested that cerebellar lesions can manifest as a “cerebellar cognitive affective syndrome” consisting of disturbances in executive functioning, spatial cognition, linguistic abilities and personality (5). The association areas that are connected to the cerebellum include regions of the prefrontal cortices, posterior parietal lobes, superior temporal lobe, and parahippocampus.

Figure 1: Axial T2-weighted magnetic resonance image showing absence of the cerebellar vermis with the fusion of the cerebellar hemispheres.

Figure 2: Axial T1-weighted magnetic resonance image showing the fusion of the dentate nucleus, agenesis of the vermis and cerebellar fusion.
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In conclusion, RS occurs most of the time as a complex condition associated with a variety of intra- and/or extraneural malformations, as seen in our patient. We postulate a link between the cognitive disturbance of this patient and his cerebellar malformation, suggesting that this case may represent another example of the role of cerebellum in cognitive functioning.

REFERENCES

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