Major Depressive Disorder mimicking Mental Retardation: A rare case of Joubert Syndrome

Mental Retardasyonu taklit eden Major Depresif Bozukluklu nadir bir olgu: Joubert Sendromu

Nazli Ece Karzan¹, Damla Eyuboglu²

¹M.D., ²Assoc. Prof. Department of Child and Adolescent Psychiatry, Eskişehir Osmangazi University, Eskişehir, Turkey

SUMMARY

Joubert Syndrome is a rare genetic and clinical disorder that affects many different parts of the body, especially the central nervous system, musculoskeletal system, kidneys, eyes, respiratory system, endocrine system and liver. In addition to all these affected organ systems, dysmorphic facial features can also be observed in people with Joubert Syndrome. Symptoms and clinical signs in individuals with Joubert Syndrome vary greatly in several ways, to the extent where afflicted individuals even from the same family might have completely different clinical presentations, when compared to one another. Although the clinical features of Joubert Syndrome appear in the neonatal period, the diagnosis can usually take years after the symptoms appear. A prevalence of between 1 per 80 000 and 1 per 100 000 live births has been reported by many investigators. Despite its heterogeneous symptom cluster, poor outcome in cases of Joubert Syndrome presenting predominantly with hypotonia and global developmental delay has been reported. Although mental retardation has been reported as the primarily anticipated psychiatric condition in cases with Joubert Syndrome, some other clinical sources within relevant literature identify autism spectrum disorders as another possible psychiatric diagnosis observed in affected individuals. It is known that there are a limited number of cases with Joubert Syndrome, it is a rare condition and the main psychiatric finding expected in the course of the syndrome is mental retardation. With this case report, we have aimed to discuss a case of Joubert Syndrome who had presented with symptoms of major depressive disorder that mimicked mental retardation.

Keywords: Joubert Syndrome, Major Depressive Disorder, Mental Retardation

(2022;25:.....)
DOI: 10.5505/kpd.2022.40326

OZET


Anahtar Kelimeler: Joubert Sendromu, Major Depresif Bozukluk, Mental Retardasyon

The arrival date of article:14.04.2021, Acceptance date publication: 27.08.2021
INTRODUCTION

With its estimated prevalence as between 1 per 80,000 and 1 per 100,000 live births, Joubert Syndrome (JS) is a rare genetic disorder with autosomal recessive inheritance, characterized by partial or total absence of cerebellar vermis (1,2). Making a formal diagnosis for the syndrome is challenging, due to phenotypical variance. No specific gene has been identified for the syndrome so far, as well as absence of pathognomonic biochemical findings (2). Mutations in over 30 genes responsible for protein synthesis of cellular structures, also known as primary cilium that have major role in detection of physical medium and chemical signals, might cause Joubert Syndrome (1,3). Clinical and neuroradiological characteristics need to be evaluated together, in order to make a formal diagnosis. Even though clinical features might emerge as early as during the neonatal phase, it generally takes years to make the diagnosis. Common clinical symptoms of the syndrome include ataxia, hypotonia, abnormal eye movements, nystagmus, episodes of hyperpnea-apnea, and global developmental delay (3). Specific facial features such as broad forehead, low-set ears, triangle-shaped mouth, hypertelorism, arched eyebrows, droopy eyelids might alert the clinician to further evaluate the child for a possible JS diagnosis (4). In most affected individuals, developmental delay and mental retardation that vary from moderate to severe forms are common, though in some studies, autism spectrum disorder has also been reported as another psychiatric condition accompanying the course of the syndrome (5,6). Despite its heterogeneous symptom cluster, poor outcome in cases of Joubert Syndrome presenting predominantly with hypotonia and global developmental delay has been reported (7). Distinctive neuroradiological feature of Joubert Syndrome has been identified as the combination of cranial abnormalities caused by problems in the development of rear structures of the brain, including cerebellar vermis and the brainstem, also known as the molar tooth sign that is visible via brain imaging studies, like Magnetic Resonance Imaging (MRI). This specific radiological sign was named after the characteristic brain abnormalities’ resemblance to the cross-section of a molar tooth, when seen on an MRI (1,3,7).

Major depressive disorder is an episodic psychiatric disorder that presents with depressed mood, loss of interest, restlessness, decreased energy, impaired cognitive functions, and a combination of vegetative symptoms such as alterations in sleep and appetite (8). Symptoms of the disorder might vary based on the age, gender, education level and cultural characteristics of the child. Prevalence studies of major depressive disorder in school aged children have reported an estimated value of approximately 2%, with no gender dominance for given ages (8). Even though cases with JS were mainly expected to meet the criteria for intellectual disability as the index psychiatric condition, following an in-depth assessment, we have determined that our case had age-appropriate cognitive functioning and symptoms observed were actually reflective of another psychiatric disorder underneath, that was major depressive disorder. With this case report, we aim to discuss a case of Joubert Syndrome who is presented with symptoms of major depressive disorder that mimicks mental retardation. Secondary purpose of this case report is emphasizing the importance of performing an elaborative clinical evaluation and taking a detailed history from the patient and their parents in the psychiatric evaluation and diagnosis process.

CASE

The case was a 7 year 3 months old male who had been brought to child and adolescent psychiatry unit in order to formally apply for a request to renew his disability report given by the health board of the institution. Gathered from the anamnesis obtained by interviewing the mother, it was learned that the case was being followed up by child neurology and child nephrology units due to conditions involving developmental delay starting from birth, disruptions in the EEG, a history of seizures and chronic renal failure. In his last disability report, the case was also diagnosed with mild mental retardation and speech problems. The case had born at 38 weeks with a birthweight of 4750 grams, and had a history of deoxygenation at delivery and difficulties in sucking while being breastfed, later on. When his developmental history was interrogated in detail, it was learned that the case had managed to hold his head up at 2 years old, sit without any support at 3, walk at 4,5, and...
Major Depressive Disorder mimicking Mental Retardation: A rare case of Joubert Syndrome

The case was brought to the child and adolescent psychiatry unit for the treatment of his speech disorder and motor retardation and for the renewal of his disability report which was previously pre-

DISCUSSION

The case was brought to the child and adolescent psychiatry unit for the treatment of his speech disorder and motor retardation and for the renewal of his disability report which was previously pre-
pared with the diagnosis of mental retardation. The patient’s dysmorphic facial appearance, motor retardation, the history of deoxygenation following delivery and difficulties in breast-feeding were consistent with the defined Joubert Syndrome clinic. (7,9). As a result of, regular outpatient follow-up and clinical assessments, it was understood that even though the case had a history of speech delay, his language skills had developed by age and his current speech problem was articulation disorder. Additionally, it was observed that the case actually refused to speak in social outlets since his speech was not understood by other people and his mental capacity was compatible with his peers and developmental level. Among predominant symptoms of depression in children are social withdrawal, avolition and loss of interest (8,10). Symptoms of our case at the time would fit this profile. Since the mental capacity of the case is considered within normal limits, the patient was aware of his speech disorder, motor retardation, that he needed his mother’s help to continue his daily functions, and his father’s neglectful attitudes. It is thought that all these paved the way for the decrease in self-esteem, social withdrawal and depression in the patient.

Even though, no research has been found in the relevant literature evaluating the possibility of a link between major depressive disorder and JS, we believe it would be important to underline the need to undergo a detailed psychiatric examination for individuals diagnosed with JS. Although many cases of JS are presented with mental retardation as the primary psychiatric condition, our case had normal intellectual functioning, and the delay in his developmental milestones as well as limited functioning, were due to his underlying neurological motor retardation tied to the syndrome. It is especially hard to diagnose depressive disorder in small children, mainly because of the natural and expected underdevelopment of their verbal skills and emotion regulation etc, as well as the negative impact depressive mood exerts on cognitive functions (9). For all these reasons, major depressive disorder might mimic cognitive delay and intellectual disability in children. Symptoms of our case at the time of application mimicking mental retardation and since intellectual disability was a frequent and anticipated component of JS, clinicians that had evaluated the case before might have diagnosed him with mental retardation. However cases like this one; shows us the importance of carrying out an elaborate clinical assessment and history, obtaining information about the behavior patterns and human relations in the various environments, and making detailed observations in each case that applied for us. We would like to emphasize the importance of regular outpatient follow-ups and evaluation interviews in order to complete these necessary processes in order to make the correct diagnosis in all cases who applied to us (11). It is only possible to create the most appropriate treatment plan once we are able to make the right diagnosis, and provide optimum levels for resuming psychiatric well being of our patients.

Correspondence address: M.D. Nazl Ece Karzan, Department Of Child And Adolescent Psychiatry, Eskisehir Osmangazi University, Eskisehir, Turkey, n.eceeej@hotmail.com

KAYNAKLAR

5. Takahashi TN, Farmer JE, Deidrick KK, Hsu BS, Miles JH, Maria BL. Joubert syndrome is not a cause of classical autism.
Major Depressive Disorder mimicking Mental Retardation: A rare case of Joubert Syndrome


