

Case Report

Bipolar and Goldenhar: A Case Report of Bipolar Disorder with Psychotic Features in a Man with Goldenhar Syndrome

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Abstract

Background: Goldenhar syndrome is a congenital condition primarily affecting eye, ear, and spine development. In some cases, it can also impact the heart, lungs, kidneys, and central nervous system. The exact cause of Goldenhar syndrome is not fully understood, and its incidence is estimated to range from 1 in 3500 to 1 in 5600 live births. While intellectual disability is associated with the condition, no established link exists between Goldenhar syndrome and psychiatric disorders.

Case presentation: In the presented case, a patient with Goldenhar syndrome has been diagnosed with an episode of mania with psychotic features in the context of bipolar disorder. The patient had no history of prior mental disorders and had an independent life.

Discussion: There are few case reports of Goldenhar syndrome patients with mental disorders. In all instances, including our own case, patients exhibited psychotic features during psychiatric examinations.

Conclusion: Based on the studies cited and our own research, it is recommended to further investigate the potential association between this syndrome and psychiatric disorders.

The following sections provide details on the examination, medical history, and treatment of this patient.

Introduction

The Oculo-auriculo-vertebral spectrum (OAVS) disorder, also known as Goldenhar syndrome, is a congenital condition characterized by its complexity and heterogeneity, primarily affecting the aural, oral, and mandibular regions. Maurice Goldenhar first described the characteristic features of the disease in 1952 [1]. The primary signs and symptoms of Goldenhar syndrome include facial asymmetry, microtia (partially formed ear) or anotia (absent ear), ocular dermoid cysts (non-cancerous growths in the eye), and spinal abnormalities. Additionally, Goldenhar syndrome can affect

various other organs, such as the heart, lungs, kidneys, and central nervous system. The condition is attributed to developmental issues during fetal formation within the mother's womb, particularly involving the first and second branchial arch structures [2]. The exact cause of Goldenhar syndrome remains unclear, and its incidence ranges from 1 in 3500 to 1 in 5600 live births, with a male-to-female ratio of 3:2. It is more commonly observed in children with congenital deafness. Although most cases are sporadic, there have been suggestions of an autosomal dominant genetic marker and multifactorial inheritance. While specific genetic abnormalities have not been consistently identified in chromosomal studies,

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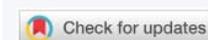
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Keywords: Goldenhar syndrome; Bipolar disorder; Oculo-auriculo-vertebral spectrum

Abbreviations: OAVS: Oculo-Auriculo-Vertebral Spectrum; IQ: Intelligence Quotient





it has been proposed that the 22q11 locus contains crucial genes that may influence regulatory signaling events during the development of the pharyngeal arch. 22q11 deletions or duplications could contribute to the craniofacial dysmorphism observed in Goldenhar syndrome [3].

The use of drugs such as thalidomide, retinoic acid, tamoxifen, and cocaine during pregnancy are possible risk factors for developing this syndrome. Other studies reported gestational diabetes, rubella, and influenza as potential etiological factors [2].

In a comprehensive study involving 566,255 individuals in Australia, Junaid, et al. [4] reported a 14.3% prevalence of intellectual disability among Goldenhar syndrome cases. Additionally, the same study found no instances of Goldenhar syndrome co-occurring with autism spectrum disorder. Yet, limited studies have reported comorbid psychiatric disorders. We present what seems to be the first published case of a patient with Goldenhar syndrome and bipolar disorder.

Case presentation

A 27-year-old Persian man diagnosed with Goldenhar syndrome arrived at the psychiatric emergency room for examination and treatment. He was accompanied by social emergency personnel, who complained about his aggressive behavior. Over the past few days, he has been experiencing symptoms such as insomnia, heightened energy, aggression, and suicidal thoughts. According to his parents, his behavior has noticeably changed over the past three months, oscillating between periods of depressed mood and elevated mood. He does not have any prior medical or psychiatric diagnoses on record. No reported prenatal or perinatal abnormalities were associated with his fetal stage and early development. The patient's childhood and adolescence were without any significant troubles or issues. The patient's school performance had been average up until a certain point. However, after completing two semesters at the university, he dropped out with a poor academic record. The patient was single and did not have any history of legal issues. He has been using opioid substances for a few months. Furthermore, he was working at a bakery and has been socially and financially independent.

In the psychiatric examination, the patient was dressed appropriately, and his physical appearance aligned with his reported age. The patient had a cooperative attitude during the interview. In a psychomotor examination, he exhibited signs of hyperactivity. He was talkative and expressed strong religious beliefs. Additionally, he exhibited delusions of grandeur. The patient's mood was irritable, yet his affect was congruent with his mood. He experienced both auditory and visual hallucinations, with no signs of illusions. The patient demonstrated full orientation to person, time, and place. However, their attention, comprehension, and concentration were severely impaired. In the IQ test, the patient achieved

a variable IQ score of 102, with a performance IQ of 84 and an overall IQ of 94, placing them within the average range of intelligence. The Bender-Gestalt test assessing the maturation of visuomotor perceptions did not reveal any disturbances.

The physical examination revealed several notable findings, including microtia (underdeveloped ear), micrognathia (small jaw), zygomatic hypoplasia (underdeveloped cheekbones), mild scoliosis (abnormal curvature of the spine), and partial deafness. However, routine blood tests, electrolyte levels, renal and liver function tests, and electrocardiography all returned normal results. The patient received a treatment regimen of Risperidone (6 mg/day), Sodium Valproate (500 mg/day), and psychotherapy. Following the treatment, the patient experienced a complete recovery in both affective and psychotic symptoms, accompanied by notable improvements in attention and concentration.

Discussion

We presented a case of a man with Goldenhar syndrome presenting symptoms of bipolar depression. He has been experiencing the symptoms for three months and was referred only after an episode of psychotic mania. The patient had an average IQ with no signs of intellectual disability and average educational performance before university. He has had an acceptable performance in society and did not was not involved in crime or substance abuse. Following treatment, the patient experienced a full recovery, with significant improvements observed in both mood and cognition.

Goldenhar syndrome is a congenital condition primarily impacting eye, ear, and spine development. Its main symptoms include facial asymmetry, microtia (underdeveloped ear), non-cancerous lipodermoid tumors, and spinal abnormalities. Intellectual disability is probable in individuals with Goldenhar syndrome, and there is currently no evidence suggesting a clear association between Goldenhar syndrome and psychiatric disorders [5]. In the following paragraphs, we examined only two case reports detailing psychiatric disorders associated with Goldenhar syndrome.

In their study, Brieger, et al. [6] described the case of a 27-year-old man diagnosed with Goldenhar syndrome. The patient was referred to a psychiatrist by his general practitioner. Since birth, he had exhibited facial asymmetry, dysplasia of the right auricle, and dysplasia of the right thumb. During the evaluation, the patient demonstrated a loosening of associations in his formal thought processes. Mild impairments in attention, comprehension, and concentration were also noted. Additionally, he experienced olfactory hallucinations and held delusional ideas, although he did not have autochthonous delusions. Ultimately, the patient received a diagnosis of schizophreniform disorder [6].

In their article published in Russian, Kovaleva [7] reported an 18-year-old male patient with Goldenhar syndrome



admitted to a psychiatric ward. A retrospective analysis of his medical records revealed childhood psychopathological disorders, including delayed psycho-verbal development, emotional immaturity, and social behavior delays. He was diagnosed with an affective disorder with psychotic features, an organic personality disorder attributed to Goldenhar's syndrome, and chronic fatigue syndrome. Kovaleva [7] further emphasized that the clinic of genetic pathology (Goldenhar syndrome) had underestimated the presence of psychopathological disorders, resulting in inadequate diagnostic and treatment support for the child. This oversight ultimately contributed to mental disorders and the development of an organic personality disorder, highlighting the importance of recognizing and addressing such conditions in a comprehensive manner [7].

We conducted a review of two additional case reports of Goldenhar syndrome with coexisting mental disorders. In line with our own case, all of these reports revealed that the patients exhibited psychotic features within the context of major depression, bipolar disorder, and schizophreniform disorder. This finding holds potential for further exploration.

Conclusion

Sharing personal experiences, engaging in discussions about diagnoses, and exploring treatment plans have proven to be effective strategies for disseminating knowledge among professionals. Based on the studies cited and our own research, it is recommended to further investigate the potential association between this syndrome and psychiatric disorders.

Acknowledgment

We would like to express our sincere gratitude to the patient for his valuable cooperation and written consent.

Ethical consideration

Our research adheres to the principles outlined in the Helsinki Declaration. These include obtaining informed consent from participants, ensuring their privacy and confidentiality, and minimizing any potential risks.

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