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Symptoms of Mood Disorder in Undiagnosed Aicardi Syndrome: A Case Report

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Abstract

Aicardi syndrome is a rare neurological disorder diagnosed at an early age with the help from radiology imaging. Therefore, the sooner it is diagnosed, the sooner the interventions can be beneficial. However, as with mood disorders, it too has a spectrum of symptoms and can cause delays in treatments. In the following case report, a 26-year-old female's neurological symptoms were misdiagnosed as symptoms of Schizophrenia. Through this case report, the goal is to highlight the pathway to diagnose and treat an individual.

Keywords

Aicardi Syndrome, Schizophrenia, Behavioral Disturbances, Intellectual Disability

1. Introduction

The partial or complete absence of the corpus callosum, known as agenesis of the corpus callosum (ACC), was first observed in 1812 during an autopsy, and since then, with the advancement in technology, it has been identified using the various radiology machines [1]. Schizophrenia (SZP) was known as "dementia praecox" as Dr. Emile Kraepelin was the first to describe it in 1887. The term "schizophrenia", translated in Greek as "Split-Mind", was used in 1911 by Dr. Eugen Bleuler in order to distinguish Dr. Kraepelin's work from other neurological forms of dementia [2]. Aicardi Syndrome (AS) was first identified in 1965 by the French Neurologist Jean Aicardi. Aicardi Syndrome was initially diagnosed based on three distinct characteristics: chorioretinal lacunae, infantile spams, and partial or complete absence of corpus callosum; additional characteristics have since been further identified [3]. A connection has been identified between abnormalities of the corpus callosum and SZP [4], epilepsy, and psychosis [5], which may clarify the presence of psychotic symptoms in individuals with AS.

Since its discovery, ACC has been identified in numerous other neurological conditions, ranging from AS, Andermann Syndrome, Arnold-Chiari Malformation, and Dandy-Walker syndrome, all with different presentations [1]. Similarly, the symptoms of SZP have been identified within other mood disorders ranging from depression, anxiety, post-traumatic stress disorder, substance use disorder, or dementia, also all with different presentations. The key to diagnosing any of the above disorders has been to get a concise history and complete a thorough physical exam which includes bloodwork and imaging.

The following case report is that of an individual with AS who was unfortunately misdiagnosed with SZP and only received partial treatment as a result. There is much research from the neurological perspective of treating AS, but limited information on it for the psychiatric specialty. As this condition is rare, so is the psychiatric research on its diagnosis and treatment, hence indicating a need for understanding and managing the behavioral disturbances which emerge in this neurological disorder. The following is a unique case report which aims to shed light on the disorder.

2. Case Report

The patient was a 26-year-old female with a past medical history of Cerebral Palsy (CP) and past psychiatric history of Intellectual Disability and SZP. She was born premature with the umbilical cord wrapped around her and was placed in the NICU for one month before being discharged home with her biological parents. The patient was diagnosed with CP at age 3 and had always been developmentally behind. She lived at home with her biological mother and stepfather, did not graduate high school due to her underlying illness with CP and learning disability; she was, however, on social security disability and supplemental security income. At a young age, she witnessed her mother being physically abused by her biological father, but eventually he walked out of their life and stopped being involved in her life.

The patient started having seizures at age 10 but was never evaluated by a neurologist nor did she have any other workup completed for it. She was reported to be exhibiting behavioral disturbances since she was about 11 - 12 years old. She was first hospitalized to an acute inpatient mental health hospital at age 16, later at age 23 and then at age 26. She reported the start of hallucinations and increase in aggression when she was 16 years old, started treatment for psychosis, and was stable for about 6 - 7 years until she had her second mental health admission at age 23. When her behaviors become too challenging to manage-she was running away from home, hitting herself in the head repeatedly every 2 - 3 nights, threatening suicide, demanding her cochlear implant be taken out (as it turned out she did not have a cochlear implant), insisted that her parents are not her real parents and that she was "stolen," and was having auditory hallucinations, her mom brought her to the emergency department for another evaluation and assessment for acute inpatient admission for mood stability and her safety. By the time she was admitted to our facility, she had been tried on escitalopram,

quetiapine, valproic acid, benztropine, and perphenazine.

Early on in her admission, the patient was noncompliant with her medications and was observed to be engaged in self-dialogue often and would not have spontaneous conversations with the team, staff, or other patients on the unit. At one point the team considered her to be non-verbal with limited expressive speech. Within 3 days of her admission, she had a witnessed seizure on the unit prompting a Neurology consult. The witnessed seizure consisted of tonic-clonic convulsions followed by altered mental status, agitation, aggression towards self, and yelling and shouting but not at anyone in the room. When the convulsions subsided, she became calm and quiet, she was non-responsive to commands or sternal rubs. On her neurology exam she was observed to have her gaze shift towards the right side. After 4 - 5 minutes, she would have a repeat in the episode-convulsions, aggression, and then calm with the right sided gaze. In addition to medication treatment to break the seizures, an extensive workup was completed with laboratory examination, MRI, and EEGs. MRI Brain indicated partial congenital hypoplasia of the distal body of corpus callosum with mild periventricular hyperintense confluent T2 signal abnormality, likely demyelination and mild colpocephaly (Figure 1 and Figure 2). Overall, the MRI brain indicated Aicardi Syndrome, Mild. She went on to complete 3 EEGs, which were all negative for seizure activity.

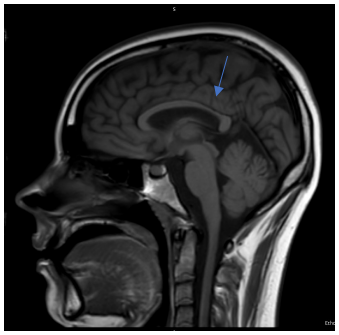

Figure 1. Sagittal T1-WI MRI shows partial congenital hypoplasia of the distal body of corpus callosum.

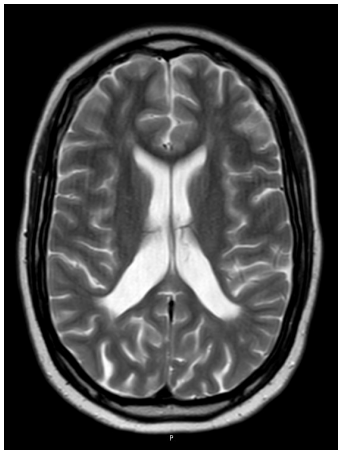

Figure 2. T2-WI MRI shows mild periventricular hyperintense confluent T2 signal abnormalities, likely dysmyelination.

The patient was started on Keppra 1 mg twice a day, but, unfortunately, she was selectively complaint and continued to have further seizure activity. As a result, Ativan 1 mg four times a day was started prophylactically to help break unwitnessed seizures and she was placed on 1:1 observation for concerns of seizures and falls. She required much redirection to take medicines at times and was inconsistent with them. She was then started on Depakote Sprinkles and Risperidone 1 mg to address behaviors secondary to the seizures. Once she was more compliant with the medications, Keppra 1 mg two times a day, Depakote Sprinkles 500 mg as two times a day, and Risperidone 2 mg oral solution two times a day, she did not experience any side effects during the remainder of her stay. The combination of treatment did decrease the frequency of her seizures and from there the aggressive behaviors and hallucinations decreased as well. She started attending groups although could not fully participate or benefit from them due to her low IQ. She was also able to engage in conversations with less paranoia and psychosis. As she continued to improve, the 1:1 was discontinued. The patient noted to show significant improvement when she was complaint with her medications and noted to be verbally assaultive when she missed a dose, indicating the importance to comply with her seizure medications. She was discharged home with the diagnosis of Aicardi Syndrome, intellectual disability, and mental health disorder due to another medical condition and treatment continued with Keppra 1 mg two times a day, Depakote Sprinkle 500 mg two times a day, and Risperidone 2 mg two times a day, with plans to follow up with both neurology and psychiatry for medication management.

Since her hospital discharge, the patient has had a stable mood, has not had any new seizures reported, and regularly follows-up with her neurologist and psychiatrist. The patient continues to the medications prescribed at the time of discharge with the exception for having added escitalopram for concerns of depression. She has not had any re-admissions to an acute inpatient behavioral health unit either.

3. Discussion

Abnormalities related to the corpus callosum are the most common brain malformations found, and with that the most common clinical findings include intellectual delays, developmental delays, seizures, and behavioral disturbances [6]. In comparison, the most common findings in SZP include decrease in total brain volume, enlarged ventricles, reduced medial temporal, superior temporal, prefrontal, thalamic and hippocampal volumes, and increased globus pallidus volume [7] [8] [9].

Research has shown reports of both mood disorders with neurological symptoms on exam as well as neurological disorders with mood disorders. Moreover, altered mood and hallucinations have been seen with both complete and partial agenesis of the callosum as well as abnormalities with the prefrontal and temporal cortices and white matter [4] [7]. Studies have also demonstrated reports

of neurological disorders with similar cortical abnormalities found on MRI after the chief complaint was that of worsening seizures in early adulthood [10].

4. Conclusion

As in this case, the patient presented with a chief complaint of behavioral concerns and was thought to have an acute decompensation of SZP. However, with the help of medical staff who were able to witness a seizure led to the Brain MRI in which she was found to have hypoplasia of the distal body of corpus callosum and colpocephaly, giving her the diagnosis of Aicardi Syndrome, mild form. Although the concern of seizures was reported in early childhood, the fact that it was not investigated nor treated effectively in a timely manner, leads one to the conclusion that her seizures were possibly worsening and resulting in the aggressive behaviors. There have been similar cases where the initial diagnosis of a mental health disorder does not match the diagnosis at the time of discharge. The purpose of this case report is to increase awareness of the similarities of two very different diagnoses with different treatment approaches.

Conflicts of Interest

The author declares no conflicts of interest regarding the publication of this paper.

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