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Neuropsychiatric Manifestations of Skraban-Deardorff Syndrome: Coprophagic Behavior, Progressive Catatonia, and Psychotropic Hypersensitivity

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Abstract Skraban-Deardorff Syndrome (SDS) is an extremely rare neurodevelopmental disorder caused by a mutation in the WDR26 gene, leading to delayed psychomotor development. Along with this, patients present with different levels of intellectual disability, characteristic dysmorphic features like coarse facies with prominent maxilla and upper lip, and early-onset seizures. They also exhibit neurodevelopmental features such as hypotonia, failure to thrive, and feeding difficulties, minor structural brain anomalies - like enlarged ventricles, white matter volume loss, cerebellar hypoplasia, and microcephaly. Behaviorally, individuals with SDS are elated and socially engaging, with a preference to engage with adults and children rather than pursue solitary activities. In this case report, we present a unique case of SDS in which the patient presented with coprophagic and self-mutilating behavior, progressive catatonia with a change in baseline, and decreased appetite following the loss of a loved one who was also his primary caretaker. The patient exhibited increased sensitivity to psychotropic medications, including aripiprazole, and developed extrapyramidal side effects, further adding to his decompensation from baseline. The patient experienced marked resolution of his coprophagic and catatonic behavior after discontinuation of Aripiprazole, and continuation of Lorazepam daily. This case highlights how patients with neurodevelopmental disorders present differently when faced with changes in their routine and life circumstances, which can lead to unusual behaviors and physiological responses. Additionally, individuals with SDS may require complex care from various specialists. Therefore, a multidisciplinary approach is crucial for diagnosing and improving patient outcomes when presented with rare diseases such as Skraban-Deardorff Syndrome.

Keywords: *Skraban-Deardorff Syndrome, Coprophagia, Catatonia, Psychotropic Hypersensitivity, Neurodevelopmental disorder, Intellectual disability*

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1. Introduction

Skraban-Deardorff Syndrome (SDS) is a rare genetic and neurocognitive disorder that was first discovered in 2017. To date, fewer than fifty cases have been reported worldwide, with a range of 24 months to 34 years [1]. The diagnosis is made in combination with the physical symptoms and molecular testing for the heterozygous pathogenic WDR26 gene variant [1,2]. Molecular testing has shown the involvement of chromosome 1q42.13, encoding a subunit of the multiprotein c-terminal LIS homology E3 ubiquitin ligase complex [2]. Furthermore, WDR26 is expressed in abundance in the human brain and skeletal muscle during the fetal stage, as well as in the skeletal muscle and heart in adults, influencing vital processes such as neuronal and cardiomyoblast proliferation, apoptotic signaling, and leukocyte activation and signal transduction [2,3]. Phenotypically, diagnosed individuals demonstrate varying degrees of developmental delay, intellectual disability, distinct facial features like a prominent upper lip, wide-spaced teeth, and a broad forehead [2]. Additionally, it is reported that almost all individuals will present with speech delay or present as nonverbal [1]. Patients also have been found to have a predisposition to possible seizures, difficulty with motor coordination, and gait abnormalities. Due to the limited number of cases, the clinical spectrum and medical management of this neurocognitive disorder is poorly understood. We report a case of a patient with SDS who presented with quite a few of the symptoms aforementioned, but also with characteristic coprophagic behavior - the ingestion of feces.

2. Case Description

Our patient is a 13 year old male with a past medical history of SDS diagnosed in 2022 who presented to the Emergency Department (ED) with increased agitation and patient not acting like himself. The patient was recently hospitalized a week prior due to his abnormal behavior of picking at his rectal skin leading to rectal bleeding and eventually iron deficiency anemia. During his prior hospitalization, a psychiatric evaluation was done and it was recommended that the patient be started on Abilify. A few days prior to the patient's presentation in the ED, the patient visited a psychiatric clinic, at which point his Abilify was increased. His uncle, who is his legal guardian, noticed that the patient was not behaving like himself. He stated that the patient's appetite decreased and he was not following commands at baseline. The uncle stated that he found the patient picking at his rectal skin and consuming his feces for a period of ten minutes. The uncle attested that the patient remains stiff at all times. He was having normal bowel movements and no nausea, vomiting, diarrhea or rectal bleeding on presentation.

On examination, the patient was slightly tachycardic, normotensive and afebrile. Abdomen was soft, nondistended, nontender to palpation. CBC showed leukocytosis of 14.5 without associated bandemia. Hemoglobin was 8.1 which was stable from previous visits, in which hemoglobin was 6.8 for which he received 1 unit of PRBC transfusion. He was found to be in flexed posture, dysmorphic features, increased muscle tone in all four limbs with tongue protrusion, and tremorous but otherwise at his baseline. The patient was initially meant to be transferred to the pediatrics floor from the emergency department but was found to be jittery and suddenly became stiff at which point his oxygen saturation dropped below 60%. After 2-3 seconds, the O2 saturation came back up. The patient was given 1mg of Ativan for his agitation and for concern of extrapyramidal side effects. The patient was then transferred to the PICU where he was given Benadryl 25 mg IV for his extrapyramidal symptoms. After monitoring of respiratory status and no further episodes, the patient was transferred to the pediatric floor.

The patient was evaluated by Psychiatry and was held off all psychotropic medications due to heightened sensitivity in neurodevelopmental patients and continued with the Ativan 1 mg IV. The patient was trialed on uptitration of Ativan to 3 mg PO TID for catatonic features. He showed improvement in symptoms as the patient was more participatory in encounters, and demonstrated less rigidity and hypervigilance. The patient was held at Ativan 3 mg PO TID because there was a concern for potential oversedation and disinhibition that might lead to recurrence of self-gratifying and coprophagic behavior if Ativan was augmented to 4 mg po TID. Social work was able to reach out for aftercare treatment, consisting of returning to home, attending his developmental school and resuming physical therapy.

3. Results/Outcomes

The patient diagnosed with SDS had marked resolution of his coprophagic and catatonic behavior after discontinuation from psychotropic medication like Aripiprazole and continuance of Lorazepam daily.

4. Discussion

Patient's condition is exceedingly rare in the world and is known to have intellectual and developmental disabilities, coarse facial features, microcephaly, stereotypic/autistic features, swallowing/feeding deficits, gait abnormalities like forefoot varus secondary to skeletal and bone dysplasia, and overall failure to thrive [1]. This patient demonstrates all these characteristics. However, unlike classic presentation, our patient and case are unique in that he also presented with coprophagic behavior and progressive catatonia. Catatonia is a neuropsychiatric state that can commonly occur in conditions like schizophrenia, depression, mania, autism, medication-induced, and withdrawal states. It is not a known side effect of SDS in the literature, which makes this a unique presentation [4]. What is intriguing is how much of a factor genetics plays into the behavior that was demonstrated by this patient. In addition, it also begs the question of the level of sensitivity that this patient must be experiencing from psychotropic medications such as Abilify, since his behavior was worsened by the augmentation of his Abilify. This is in stark contrast to what has been proven in literature. There has been success found in the use of Abilify (Aripiprazole) for the treatment of coprophagia in autism spectrum disorder [5]. That unique case highlights the symptoms of a high-functioning autistic individual with motor stereotypies and significant impairment in social interactions at baseline who displayed a marked reduction in coprophagic behavior after 8 weeks of treatment. Thus, evidence-based medicine allowed for our case to potentially improve with the use of Aripiprazole as well.

According to the uncle, the patient was never fully accommodated with the proper social support in his community and that could also be a reason for his decompensated presentation. Additionally, the patient recently experienced hardship with the passing of his mother just three months prior. The patient, who is unable to express his emotions, might have been enduring a tough adjustment phase. Losing a mother, especially one who was a primary caretaker, must have been an incredibly destabilizing experience. It is more than just grief—it's the loss of a foundation, a sense of security, love, and belonging. Now, being under the care of someone he hasn't grown up with adds another layer of discomfort and disorientation. It may have felt like being placed in an unfamiliar world where the routines he once knew were gone.

Neurodevelopmental disorders pose unique challenges not only for patients but also for their loved ones. Due to the complexity of developmental conditions like SDS, patients often require complex care from various specialists, ranging from dentists to cardiologists. This emphasizes how critical it is to have effective collaboration in a multidisciplinary setting. The idea of multidisciplinary healthcare teams was created to improve patient care by ensuring continuous communication between the patient, their families, and various

subspecialties. This approach allows all healthcare providers to share insights, and align treatment plans, thus allowing comprehensive support tailored to the patient's needs. The importance of this approach allows for better patient outcomes, innovation and knowledge sharing, and increased support for families. It has been noted that “poor communication and ineffective teamwork in seemingly routine interactions have been identified as contributory factors in 70% of serious incidents” [6]. The use of multidisciplinary teams will lead to the healthcare workers being able to rapidly identify and aggressively treat patients leading to more favorable outcomes, especially in patients with such rare disorders like SDS.

5. Conclusion

This case emphasizes unique and complex manifestations of SDS, specifically coprophagia and catatonia, which have been rarely documented in medical literature. By detailing this presentation, we aim to expand clinical awareness of the diverse symptoms associated with this syndrome, facilitating earlier recognition and improved management in affected individuals. Given the psychotropic hypersensitivity often observed in SDS patients, this report also underscores the importance of careful selection of medication and multidisciplinary collaboration in treatment planning. Further research is needed to explore the underlying pathophysiology, refine diagnostic criteria, and develop targeted interventions for managing neuropsychiatric symptoms. By sharing this case, we hope to enhance the understanding, diagnosis, and treatment of SDS.

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