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Case Report OMICS International

Major Depressive Disorder: A Case of an Adolescent Female with Russell-Silver Syndrome

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Abstract

RSS is a congenital genetic disorder characterized by discrete aberrations in genes that account for growth and development. A number of genetic configurations may play a formative role in the development of RSS. The syndrome may result from: (1) Maternal uniparental disomy of chromosome 7 (matUPD7) - the child essentially inherits two copies of chromosome 7 from the maternal parent. This defect is found in 10% of cases.

(2)Imprinting - a hypomethylation process for chromosome 11p15 is responsible for paternal expression of gene. Imprinting is responsible for 38% of cases. The genes that are known to be affected are H19 and IGF2. The H19 gene correlates with the phenotypic expression of skeletal abnormalities; the patient case presented below had corrective surgery for her scoliosis. The diagnosis of RSS is primarily based on clinical presentation. Prenatal genetic confirmation is available for intrauterine growth retardation (IUGR). Furthermore, researchers have elaborated upon the tangible association that exists between Russell-Silver Syndrome and Asperger Syndrome. Major depressive disorder often coincides with RSS; therefore it is of utmost importance for the clinician to adequately identify and treat depression in RSS children before it progresses to suicidal thoughts and/or psychosis.

Objective: Our goal is to assess the clinical presentation of RSS within the context of comorbid conditions, in particular, MDD and Asperger's Syndrome (AS). The purpose of this case report is to highlight the importance of early recognition and management of individuals afflicted with RSS - to raise awareness for a congenital disorder that often coincides with serious symptoms of depression, including suicidal ideation and/or psychosis.

Method: A literature search via PubMed and Google on the topics of RSS.

Conclusion: RSS can present with clinical depression which also affects medical management, and patients should be carefully monitored for the development of new symptoms and/or side effects.

Introduction

Russell-Silver Syndrome (RSS) is a congenital genetic disorder characterized by discrete aberrations in genes that account for growth and development. A number of genetic configurations may play a formative role in the development of RSS. The syndrome may result from:

(1) Maternal uniparental disomy of chromosome 7 (matUPD7) – the child essentially inherits two copies of chromosome 7 from the maternal parent. This defect is found in 10% of cases [1,2].

(2) Imprinting – a hypomethylation process for chromosome 11p15 is responsible for paternal expression of gene. Imprinting is responsible for 38% of cases [1].

The genes that are known to be affected are H19 and IGF2 [3]. The H19 gene, in particular, correlates with the phenotypic expression of skeletal abnormalities; the patient case presented below had corrective surgery for her scoliosis [4]. However, due to the non-homogenous nature of genetic contribution with respect to RSS development, patients with RSS are generally evaluated based on salient symptom expression rather than presumed genetic make-up [5]. Due to the multi-faceted nature of RSS, therapeutic management may be further complicated by the existence of comorbid conditions.

The diagnosis of Russell-Silver Syndrome is primarily based on clinical presentation. Prenatal genetic confirmation is available for intrauterine growth retardation (IUGR) [4]. However, there is a caveat; namely, reliable measurement of IUGR by means of ultrasound is not feasible until the third trimester [4]. The diagnosis of RSS is considered if patients have three major criteria or two major and two minor criteria; Major criteria encompass IUGR, postnatal deceleration in height and weight, head circumference on par with normal population, and/or the presence of a limb asymmetry of sorts. The list of minor criteria includes short arm span, clinodactyly (curvature of the 5th finger) and triangular facies, perhaps, in conjunction with forehead prominence. In addition to the aforementioned diagnostic criteria, RSS patients may also share the following symptoms: café au lait spots, visible psychomotor disturbances, gastroesophageal reflux disease or hypoglycemia due to poor appetite [4,6]. Furthermore, researchers have elaborated upon the tangible association that exists between Russell-Silver Syndrome and Asperger Syndrome. Major depressive disorder often coincides with RSS; therefore it is of utmost importance for the clinician to adequately identify and treat depression in RSS children before it progresses to suicidal thoughts and/or psychosis.

Objective

Our goal is to assess the clinical presentation of Russell-Silver Syndrome within the context of comorbid conditions, in particular, Major Depressive Disorder (MDD) and Asperger's Syndrome (AS). The case involves an adolescent RSS patient with Asperger's Syndrome and new onset MDD coupled with psychotic features. The purpose of this case report is to highlight the importance of early recognition and management of individuals afflicted with RSS – to raise awareness for a congenital disorder that often coincides with serious symptoms of depression, including suicidal ideation and/or psychosis. The paper also addresses psychosocial issues concerning the families of RSS children.

Method

A literature search via PubMed and Google on the topics of RSS, AS, MDD and medical management of patients with Russell-Silver Syndrome has been performed.

Case Report

The patient is a 15 year old Caucasian female with a significant history of Russell- Silver Syndrome and Asperger Syndrome who presented with Major Depressive Disorder for one month. She is a student in the 10th grade at a local high school and currently lives at home with her mother. The patient lacks a psychiatric history of hospitalizations. Moreover, the patient has no history of suicidal or homicidal ideations.

It has been reported that the patient became progressively depressed after her father was diagnosed with cancer 5 years ago. After the patient's father was placed in hospice, her preoccupation with intrusive thoughts exacerbated. The patient's father passed away on Oct 4, 2013; this event was influential in bringing forth a series of visual hallucinations in the patient. She claims to have seen visual representations of herself alongside dead people.

The patient was asked to follow-up with the nurse and was compliant with the medications. She was referred to a local mental health center where she was seen by a psychiatrist. The patient stated that she had begun having suicidal thoughts with a plan to hang herself. The patient expressed concerns regarding her new ideations and wanted to be hospitalized. Therefore, she was sent to Bergen Regional Medical Center (BRMC) for evaluation.

Upon evaluation, the patient reported a number of typical symptoms of depression which also included significant weight loss of 15-20 lbs within the last few months. Furthermore, the patient recently experienced suicidal ideations for a period of one day as well as a corresponding plan to hang herself. The patient denied the presence of other psychotic symptoms, homicidal ideations, or ongoing substance abuse. Her blood alcohol level was less than 25 and her urine toxicology screening was negative. Despite reporting an irritable mood, the patient experienced no other manic symptoms. She reported compliance with her medications which included Quetiapine 50 mg PO QHS, Lansoprazole 30 mg PO daily, Meclizine 12.5 mg PO T.I.D., Docusate 100 mg PO at midnight, Cetirizine 10 mg PO QHS PRN, Ondansetron 4 mg PO every 8 hours PRN, calcium 1 tablet PO daily, and multivitamin 1 tablet PO daily.

The patient reported having a past psychiatric history of Major Depressive Disorder with a single episode that started in December of 2013. The patient has no history of aggressive behavior or property destruction. She has never had any legal charges or access to firearms. She reported a family history of depression in her father and bipolar disorder in one cousin on the paternal side. She denied any history of suicidal attempts in the family. The patient's past medical history includes an allergy to banana, lactose, Amoxicillin/clavulanic acid, and Pregabalin. Her medical problems include Russell-Silver Syndrome, fibromyalgia, irritable bowel syndrome, GERD, hypoglycemia, and dizziness. The patient had corrective surgery for scoliosis several years ago. As per mother, the patient had the less common matUPD7 genetic aberration of RSS.

On exam, the patient appeared younger than stated age, wearing glasses, and was appropriately groomed. She possessed a triangular facies and appeared thinly built. The patient maintained poor eye contact and seemed visibly anxious, but she was cooperative nonetheless. She displayed psychomotor retardation during the course of the exam. Her speech was soft with normal thought process and intact thought associations. Her judgment was poor with minimal to moderate insight. She was also oriented to person, place, and time. Her memory and fund of knowledge were all intact. The patient's language was articulate and intact. Her mood was depressed and her affect was appropriate and full.

The patient was admitted with a single episode of MDD coupled with severe psychotic features as well as comorbid Asperger Syndrome. The patient was initiated on Hydroxyzine 50 mg PO every 6 hours PRN for her anxiety, Acetaminophen 650 mg PO every 6 hours PRN and Quetiapine 50 mg PO at night. During her hospital course, she was integrated into individual psychotherapy, group psychotherapy, recreational therapy, and family therapy. Her medication regimen was continued and adjusted in accordance with her mother's consent. The Quetiapine was titrated to the dosage of 100 mg PO QHS daily. Escitalopram 10 mg PO was also started every morning daily for persistent depressive symptoms and obsessive features. The patient was successfully integrated into all aspects of her psychotherapeutic regimen. Furthermore, she was compliant with her medications.

As the treatment plan progressed, some of the patient's psychotic tendencies subsided. However, there were times when the patient became increasingly uncomfortable, even, suspicious, displaying the presence of referential thinking. She expressed the typical symptoms of MDD including problems with maintaining concentration at school. The patient also exhibited multiple stressors, namely, school workload, her mother's current state of sadness, and her friends' ongoing curiosity.

Upon discharge, the patient's overall condition improved and was devoid of acute symptoms of personal harm. She no longer endorsed suicidal thoughts. Her final diagnosis was Major Depressive Disorder, recurrent, severe without psychotic features in partial remission; her home medications were to be continued, adding Quetiapine 50 mg PO QHS in combination with Escitalopram 10 mg PO every morning daily. Therefore, the patient was discharged on HD 7 to care with her biological mother with good family support. She was advised to continue outpatient treatment with her previous psychiatrist and commence an intake referral made for partial hospitalization.

Management Strategies

The management of RSS is contingent upon symptom expression. The complex nature of RSS warrants an interdisciplinary approach to treatment that includes the use of growth hormone, physical therapy,

speech and language therapy, antacids, dietary supplementations and careful glucose monitoring [4]. Escitalopram was an age-appropriate anti-depressant medication indicated for our patient. However, anti-depressants are well known for gastrointestinal side effects; this patient appeared to suffer from symptoms of hypoglycemia, gastrointestinal complications and poor appetite. Thus, the patient's diet and symptoms were closely monitored [7]. Her age and BMI were also of importance when choosing the proper medication and corresponding dosage. Once the therapeutic dosage of the medication was achieved the patient's MDD symptoms markedly improved. Her sleep disturbances resolved and her concentration improved with time. Although, the patient's anxiety concerning her school assignments persisted, her mother provided ample support leading to an improvement in overall self-esteem. The medication effectively resolved all psychotic symptoms during her hospital course. Her treatment plan was chosen by deliberately balancing the risk and benefit of her symptoms of RSS.

The aforementioned case report included a patient that exhibited adjunctive criteria for RSS, in particular, gastrointestinal symptoms of GERD and hypoglycemia. Due to the presence of gastrointestinal side effects, anti-depressants are used with caution in patients with Russell Silver Syndrome. Comorbid conditions such as clinical depression also affect medical management, and patients should be carefully monitored for the development of new symptoms and/or side effects.

Discussion

The author of a study determined that there was a significantly greater incidence of Asperger's Syndrome in RSS children; it has been proposed that AS and RSS may actually share a common pathway [5]. Individuals with Asperger's Syndrome are reported to be small for gestational age (SGA), therefore experiencing notable complications that include, hypoglycemia as well as diminished access to essential nutrients and oxygen during the overall development process [5]. Thus, environmental trauma may contribute to dysfunctional brain growth in these patients. Likewise, patients with RSS have risk factors in common with their AS counterparts and are therefore exposed to parallel insults [5]. Thus, it is not surprising to anticipate the co-occurrence of these conditions in patients. Interestingly enough, the matUPD7 defect shows an association with Autism Spectrum Disease. Genetic interplay between RSS and AS has been suspected [5]. Ultimately, deficits in language acquisition and/or cognitive development portend significant impairment in social areas of functioning during adolescence [2,8]. Furthermore, these developmental deficits and/or delays may contribute towards an emerging clinical presentation of a progressively depressed individual with RSS.

According to DSM-IV, an individual that meets the diagnostic criteria for Major Depressive Disorder must experience at least a single depressive episode; the observed or self-reported episode(s) must occur in the absence of manic, hypomanic, or mixed episodes [9]. The evaluation of depressive symptoms in adolescent and pediatric population proves to be challenging due to the presence of varying contextual demands (academic failures, school avoidance, etc.) as well as ongoing identity and/or social issues, namely, the lack of interpersonal coordination, public withdrawal and negative attributions regarding perceived intent of peers [10].

Conclusions

There have been noticeable advancements in medical genetics and Russell-Silver Syndrome research is still underway. The current literature reveals that the matUDP7 mutation in RSS is associated with a greater number of AS cases than the general population. Our case study highlights the same genetic anomaly (matUDP7) in accordance with the available research. It is important to note that the clinician must implement an interdisciplinary approach to properly address the symptoms of RSS, especially within the context of comorbid conditions. The patient's family should consult with nutritionists, experts in speech and language therapy as well as psychotherapists. The caregiver's role is instrumental in affecting the well-being of the patient. Furthermore, plans should be made to facilitate the child's academic endeavors and family support should be readily encouraged; teachers should be advised to take an active role in the child's academic curriculum. Lastly, various combinations of psychotherapy should be explored. Individual psychotherapy, group psychotherapy, recreational therapy, and family therapy were beneficial in addressing our patient's social and academic issues. Future research can expound upon the role of depression in other imprinting disorders and whether or not the management strategies discussed here are transferable across congenital disorders.

The authors do not have any competing interests to declare.

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