The Nursing Management of the Snyder Robinson Syndrome

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Abstract

Snyder-Robinson syndrome is an X-linked condition that causes intellectual impairment. (sometimes referred to as spermine synthase deficiency) This condition is characterized by asthenic body habitus, facial dimorphisms, a wide-based gait, osteoporosis, and recurrent fractures. Here, we present the findings of a pediatric autopsy on a 4-year-old boy who had been previously identified as having Snyder-Robinson syndrome as a result of an SMS gene mutation and had a history of seizures, repeated fractures, and aberrant gait Hypoxic-ischemic encephalopathy brought on by persistent seizure activity was the cause of death.

Present complaint and investigation: The patient, a 50-year-old male with modest mental retardation with adaptive skills, gait problems necessitating the use of an On December 9, 2021, a patient with numerous fractures, treatment-resistant myoclonic epilepsy, and a walker or holding hands was brought to the Hospital. He showed no evidence of being able to speak with numerous words and some signals despite behavioral concerns. To identify the diagnosis, a blood investigation is used. analysis of the enzyme synthase. reduced or nonexistent SMS enzyme activity in cultured lymphoblast or fresh white cells testing for molecular genetics discovery of a harmful homozygous loss-of-function mutant. x-ray Bilateral peripheral infiltrates on a chest x-ray were alarming for a viral infection. a cerebral spinal fluid and blood culture-based sepsis workup analgesia was initiated because of the changed mental status. As the developmental delay worsens, many boys with SRS experience some motor impairment.

Main diagnosis and therapeutic interventions and outcome: The physician decide that the patient has Snyder Robinson syndrome after performing a physical examination and a general evaluation. An evaluation for sepsis using cerebral spinal fluid and blood cultures was prompted by the patient's changed mental status. Bilateral peripheral infiltrates on the chest x-ray were alarming for a viral infection.

Conclusion: Our results are consistent with the theories that poor bone density results from a failure to mineralize, and that tissue-specific changes in the tissue specificity of SRS features are influenced by metabolism. A new SMS mutation results in Snyder-Robinson syndrome. Our patient’s Additional indications that the digestive system is important to include severe failure to thrive, enteral feeding intolerance, hepatic fibrosis, pancreatic exocrine insufficiency, and jejunal stenosis.

Keywords: Spermine Syntheses, Snyder-Robinson disease, Intellectual Disability, Mental Retardation.

INTRODUCTION

The symptoms of Snyder-Robinson syndrome include intellectual incapacity, anomalies of the muscles and bones, and other developmental issues. Only men experience it.1 Early childhood is when intellectual disability and delayed development first appear in males with Snyder-Robinson syndrome. Mild to severe intellectual disabilities are possible. Speaking often takes a while to develop, and problems with speech are frequent. Some individuals with the condition never learn to speak.2 The majority of affected men have an asthenic habitus, which is characterized by thinness and poor muscular mass. Typically, weakness or “floppiness” (hypotonia) first appears in infancy, and as people age, they continue to lose muscular mass. Most sufferers of this illness have an unstable stride and frequently have trouble walking. Skeletal issues are brought on by Snyder-Robinson syndrome, particularly early-onset osteoporosis (thinning of the bones). 3 Osteoporosis makes the bones brittle and
prone to breaking, frequently during everyday tasks. Arms and legs are the most commonly shattered bones in Snyder-Robinson syndrome patients. The majority of those affected also experience aberrant spine curvature from the front to the back and the sides (scoliosis and kyphosis, often called kyphoscoliosis when they occur together). Affected people are typically shorter than their colleagues and family members. A Snyder-Robinson syndrome (facial asymmetry) is characterized by differences in the size and shape of the right and left sides of the face as well as a prominent lower lip, a high, narrow roof of the mouth, or a hole in the roof of the mouth (a cleft palate). Seizures that start in childhood as well as anomalies of the genitalia and kidneys are some of the documented indications and symptoms. Other names for the condition include supermini synthase deficiency and mental retardation syndrome caused by the X gene, Snyder-Robinson (MRXSSR) SRS only has an impact on men. Currently, just eleven households with SRS children. The X-linked recessive disorder the symptoms of Snyder-Robinson Syndrome (SRS) include mental retardation, skeletal abnormalities, hypotonia, and movement disorders. Spermine synthase (SMS), an essential enzyme of the polyamine pathway that catalyzes the conversion of spermidine into spermine, has been linked to mutations in all documented instances of SRS. Current research by Li et al. using a Drosophila SRS model revealed that lysosomal dysfunction and oxidative stress are consequences of SMS deficiency.

As of present, it is uncertain what causes weakened bones in SRS patients. They could involve abnormal parathyroid hormone regulation, abnormal calcium metabolism, or other systemic consequences. Alternately, osteoclast hyperactivity or osteo progenitor cell malfunction could be the root of weak bones. Albert assessed these characteristics in two patients with SRS and discovered that decreased lack of mineralization due to poor differentiation of MSCs into osteoblasts is likely the cause of low bone density. However, drawing more general findings was difficult due to the small sample size and inherent donor-to-donor differences.

Patient information

Specific patient information

A male patient, 50 years old, has been admitted. On the 12th of September 2021, I was admitted to the hospital with the primary complaints of several fractures, treatment-resistant myoclonic epilepsy, minor mental retardation with adaptive skills gait irregularities need Multiple fractures, walking with a walker, or holding hands. The vital signs changed to B.P. 122/70, pulse 82 beats per minute, and respiration 18 beats per minute. He displayed no behavioral problems and was able to communicate with a wide variety of words and signals. The patient seems alert and involved.

Primary concern symptoms

A 56-year-old male was hospitalized at A.V.B.R. Hospital on September 12, 2021, with the primary complaint that he was SR's symptoms, development, and severity vary a little bit within families and among patients. The "gestalt" of affected youngsters includes a large low muscular mass, kyphoscoliosis, lower lip, asthenic build, and abnormal speech. Males with SRS are born hypotonic (muscle tone is weak). Early symptoms, particularly those affecting the face, emerge. Early in life, developmental milestones are also not met.

Medical family and psychological history

For the past two years, the patient has had diabetes Mellitus, hypertension, and asthma. The Snyder Robinson syndrome results in that. Find the general examination, physical examination, and additional investigations with the diagnosis. And the patient is from a middle-class, nuclear household. The patient is in good mental health. He has a good relationship with his friends and family since he is focused on the date, time, and place.

Relevant historical intervention and result

Clinical findings: Significant physical and general examinations, as well as crucial clinical findings such as biopsy samples, blood tests, x-rays, and other types of CT scans, encephalopathy, and Molecular genetic testing. Etc.
Treatment

Snyder-Robinson syndrome has no effective treatment at this time. Initially, it was believed that spermine supplementation would help cure SRS because the illness is brought on by the body’s cells not producing enough spermine. However, this strategy hasn’t worked out well. Therefore, the goal of treatment is to reduce a few SRS signs and symptoms. There have been conflicting results from speech, physical, and occupational therapy. Calcium supplementation to boost bone mineral density has been tested as a treatment for osteoporosis. Results have, once more, been inconsistent. However, patients with SRS should undergo routine monitoring due to osteoporosis and a higher risk of fractures, and calcium supplementation should be started as soon as there is a decline in bone mineral density. Several medications can be used to try and treat seizures. The effectiveness of any one drug, however, has varied, and certain seizures have proven resistant to medication. Occupational treatment, physical therapy, or speech therapy may be used to help people with Snyder-Robinson syndrome. To treat seizures, anti-seizure drugs including carbamazepine, phenobarbital, and clobazam can be used, however, the choice of drug is frequently determined by the kind of seizure. A DXA scan can be used to determine bone density, and calcium supplements may help.

Nursing management

Administering the medication to treat seizures, anti-seizure drugs including carbamazepine, phenobarbital, and clobazam can be used, however, the choice of drug is frequently determined by the kind of seizure. A DXA scan can be used to determine bone density, and calcium supplements can increase it if a doctor prescribes them. Every day, check the vital signs. Correctly evaluate and record the patients. Additionally, speech, physical, or occupational therapy may be beneficial for Snyder-Robinson patients. Maintain the reports and records. If the patient's assessment of changes in consciousness is harsh because symptoms worsen quickly. Genetic Guidance To help people and families Genetic counseling involves teaching people about the nature, mode(s) of inheritance, and implications of genetic disorders so they can make informed medical and personal decisions.

Discussion

His recent episode of congestion and fever, along with his history of persistent seizures, most definitely played a role in the seizure that caused him to need hospitalization.11 His mortality was caused by diffuse acute hypoxic-ischemic neuronal damage caused by the prolonged seizure, which also caused cerebral edema and herniation. 12 Patients with SRS frequently experience atraumatic osteoporotic fractures, which reduce the quality of life. Therefore, it is crucial to identify the reasons why poor SMS expression reduces bone growth.13 Our findings show a link between inadequate SMS expression and a number of molecular alterations that could explain decreased osteogenesis in bone marrow-derived MSCs. While reducing differences that are inherent from donor to donor, the idea of The precise separation of SMS's participation was made possible by simulating the disease using MSCs derived from healthy donors and transducing them with a particular shrank. 14 Additionally, it made it possible to include more duplicates than would have been possible to obtain from the few SRS patients. However, it is crucial to confirm our findings in MSCs produced from SRS patients. 15 The observed effect of SMS insufficiency on osteogenesis is similar to the stated reduced mineralization seen in MSCs derived from SRS patients.5 Further evidence that the inhibition occurs during the osteoblasts’ development phase comes from bone sialoprotein (BSP), which was considerably reduced even before any mineralization was observed.6 Because it shows that SMS deficit impacts bone formation even in the presence of physiologically acceptable osteogenic stimuli, the decrease in mineralization in vivo is considerable. We hypothesized that the effect of SMS deficiency seems mild because a sizable section of the scaffold in our mouse model becomes populated by endogenous (murine) cells (see DAPI staining in Figure S2). This shows that cells other than the transplanted human MSCs are responsible for at least some of the ectopic bone growth. Our observations indicate that decreased bone volume in SRS patients may be associated with impaired osteoprogenitor cell proliferation, suggesting that future studies should consider a subpopulation of osteoprogenitor cells (MSCs) before any cell expansion.9 Due to the absence of SMS activity, spermidine and spermine levels in SRS patients rise and fall as is usual. Our metabolome investigation confirmed the rise in spermidine, however 15-25, spermine was below detection levels despite being examined. This discovery raises two concepts: First, the negative effects of SMS insufficiency could be brought on by an excess of spermidine rather than a scarcity of spermine (which is already very low). Second, MSCs may have extremely low quantities of spermine, making them highly susceptible to drops in spermine levels. Putrescine is a crucial spermidine precursor, and the fact that MSCs expressing schisms showed a drop in it10 implies a potential compensatory mechanism and supports the notion that high spermidine levels are dangerous. Additionally, recent studies have revealed that the putrescine and spermidine levels of lymphoblastic cell lines derived from SRS patients are different from those of healthy donors. Ornithine, a crucial precursor to putrescine, was also consistently reduced in all four
MSCs under study, even if the amount varied, preventing statistically significant differences. Surprisingly, the relative abundance of each polyamine in MSCs coincides with their order of synthesis, with ornithine being the most prevalent 26–40.

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