Functional Decline in Patient with Sjogren-Larsson Syndrome During COVID-19: A Case Report.

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Patient

13 M with PMH of cerebral palsy, global developmental delay, ichthyosis, spastic diaparesis, and Sjogren-Larsson syndrome and a PSH of BL Achilles tendon lengthening, adductor release, femoral VDRO, hamstring lengthening and rectus femoris transfer.

Case

Patient presented to outpatient pediatrics rehabilitation clinic with new complaint of constant bilateral thigh pain, with radiation of pain to bilateral hips for two weeks. Patient denied trauma/falls. Patient's mother had tried to give Tylenol, which patient did not tolerate. Imaging revealed bilateral coxa valga/femoral neck anteversion and slight lateral uncoverage of the left femoral head that reduced with abduction. Per mother, patient ambulated with posterior rollator and AFO's, with a total of 150 feet with breaks on rollator. She reported that patient had outgrown his AFOs and had not worn them since March. Additionally, she denied any red marks in the area. Patient was previously receiving PT and OT 2x a week with speech therapy each day for 10 minutes. Due to COVID-19, patient's therapy had since stopped. During this time, patient resumed with swimming and riding a stationary bike everyday. According to mother, patient was also able to stand up in the pool. He had also been wearing corrective lens for 1 month.

Assessment/Results

Upon examination, hyperplastic and hyperpigmented skin, thoracic kyphosis, R internal rotation due to femoral anteversion, with a R popliteal angle of 30 degrees and L popliteal angle of 45 degrees, and BL toe flexion and pronation were noted at rest. TTP of BL quads and BL sustained clonus was also elicited.

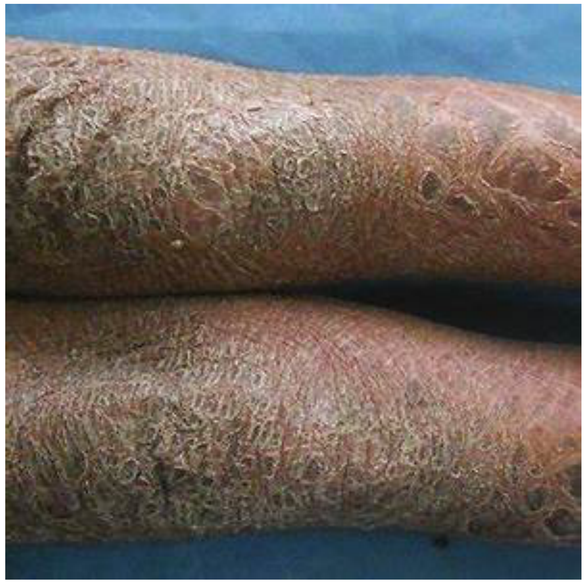

Figure 1: Lamellar type of scaling over extremities in a patient with Sjogren-Larsson Syndrome. Retrieved from Sjögren-Larsson syndrome: A study of clinical symptoms in six children - Scientific Figure on ResearchGate. Available from: https://www.researchgate.net/figure/Case-5-Lamellar-type-of-scaling-over-extremities_fig1_262916880 [accessed 25 Jan, 2021]

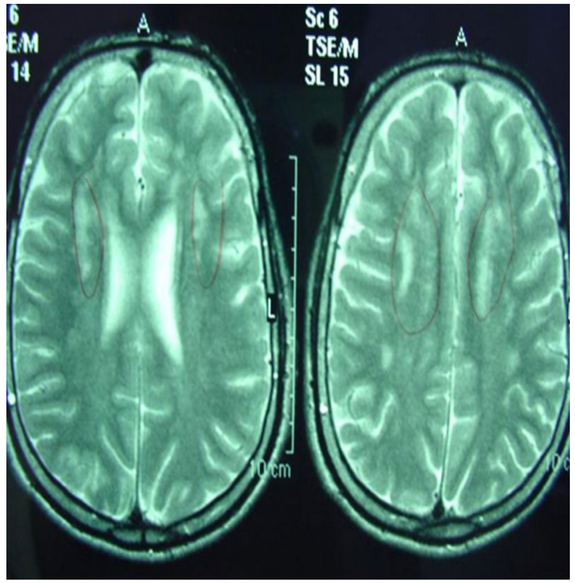

Figure 2: T2-weighted MR images of a 25 yo Sjögren-Larsson syndrome patient. MRI revealed dysmyelination in the deep periventricular white matter and reduced brain volume in frontal lobe. Retrieved from: licensee BioMed Central Ltd. 2009, CC BY 3.0 <https:/creativecommons.org/licenses/by/3.0>, via Wikimedia Commons

References

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Conclusion

The review of literatures supports that patients with Sjogren-Larsson syndrome experience cutaneous, motor, cognitive, speech and language, and ocular manifestations. In our patient, findings of BL radicular thigh pain, with changes in passive ROM, ambulation and gait contributed to his functional decline. BL solid AFOs with inner boot was prescribed, increase in therapy was requested and kinesio taping to correct internal rotation at the hips was considered to address these concerns.

Discussion

Sjogren-Larsson syndrome is a rare autosomal recessive inborn error of metabolism due to deficiency of fatty aldehyde dehydrogenase. The gene that is altered is the aldehyde dehydrogenase 3A2 (ALDH3A2) gene. This causes an accumulation of lipids in the body, especially in the corticospinal tracts of the brain and stratum corneum of the skin, resulting in a triad of ichthyosis, spastic diplegia/diplegia and cognitive and intellectual deficits. Other symptoms may be variable, but the triad will always be present.

The first sign of Sjögren-Larsson syndrome is often preterm birth. After infancy, the skin loses its redness and dark scales often appear on the neck and under the arms. Additionally, larger plate-like thick scales may develop on the lower legs. Spastic paresis will also be present and more often affects the legs greater than the arms. This can lead to wheelchair dependence. Finally, these patients will have intellectual disability with most patients reaching an average developmental age of 5-6 years old. Other symptoms that may be present but vary from person to person include seizures, speech difficulty, short stature, spinal abnormalities (kyphoscoliosis), microcephaly, peripheral neuropathy, widely spaced teeth, under-formed enamel of the teeth, widely spaced eyes, and nystagmus.

Treatment is symptomatic and multidisciplinary. Skin manifestations are treated with topical creams of 2-10% urea. Diets low in long-chain fats, such as oils, fish, nuts, avocados, and meat, and supplemented with medium-chain fats (triglycerides), such as dairy products and coconut oil, have been associated with skin improvement for some patients.

Spasticity may be treated with benzodiazepines, muscle relaxants or anticholinergics, with resulting contractures managed with bracing. Additionally, physical therapy will assist with ambulation for those that are able and transfers for those that cannot.

Speech and language therapy has shown some benefit in improving language performance and augmented communication skills.