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Recommended Citation

Dentino, Philippe J., "Clinical Management of Larsen Syndrome in Inpatient Rehabilitation: A Case Report" (2024). *Research Symposium*. 33.

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Title

Clinical Management of Larsen Syndrome in Inpatient Rehabilitation: A Case Report

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Key Words: Larsen Syndrome, Inpatient Rehabilitation, Pain Management, Filamin 1, Osteochondrodysplasia

Background:

Larsen Syndrome is a rare osteochondrodysplastic disorder associated with Filamin 1 (FLN) gene mutations and clinically presents with frequent large joint dislocations, osseous disturbances, and craniofacial abnormalities. To date, no studies have assessed the medical and rehabilitative management of an individual living with Larsen Syndrome in an inpatient rehabilitation care setting.

Case Presentation:

A 30 year-old biological female presented to the emergency department after developing worsening right-sided low back pain while bending forward. The patient's past medical and surgical history was significant for Larsen Syndrome complicated by chronic hip, knee, and back pain, Celiac disease, major depressive disorder, post-traumatic stress disorder, obstructive sleep apnea, gastroesophageal reflux disease, cholecystectomy, and multiple falls. Radiological imaging uncovered cervical spinal cord instability with multiple vertebral abnormalities, of which were managed non-surgically via a cervical stabilization orthosis and mobility precautions. Throughout the 13-day course of care, the patient's pain and functional mobility decline were managed with pharmaceutical, rehabilitative, psychological, and recreational interventions. Upon discharge, the patient's pain had improved from 7/10 to 0/10 on the Numerical Pain Rating Scale and Functional Independence Measure Motor subset scores improved from 55 to 75.

Discussion:

Primary goals of inpatient rehabilitation focused upon pain management and progressive return toward the patient's prior level of function to promote a safe discharge to outpatient therapies and home-based activities. Physical, occupational, speech, and recreational therapies were a crucial adjunct to the management of the patient's chief complaints, including addressing functional mobility impairments, activities of daily living safety, supported locomotion, and spinal protection. Spinal instability is considered the most emergent pathology in patients living with Larsen Syndrome, in light of the neurological consequences that may

follow mechanical injury during spinal loading or high-velocity movements. Cervical spinal instability without thecal sac compression was uncovered during rehabilitation, necessitating cervical bracing and non-operative measures to mitigate progression of cervical myelopathy. Although non-operative management of spinal instability was pursued, the patient ultimately improved in global pain ratings and Functional Independence Measure scores, indicating higher likelihood of meaningful return to the desired level of function.

Conclusions:

Larsen syndrome is a debilitating, yet rarely encountered condition that requires the specialized management of a multi-disciplinary medical and rehabilitative team. FLN 1 gene mutations are suspected to be underdiagnosed and will continue to present with a variety of unique phenotypic impairments requiring treatment in the inpatient rehabilitation setting. We present this case of Larsen Syndrome to highlight the importance of diagnostic familiarity and approaches to rehabilitation of individuals living with FLN 1-associated conditions.