PAIN MEDICINE CASE REPORTS

ISSN 2768-5152
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Volume 7, Number 4, pp. 171-174

# Chronic Musculoskeletal Pain in a Case of Larsen Syndrome: Role of Myofascial Trigger Points

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**Background:** Larsen syndrome is an extremely rare congenital disorder characterized by craniofacial anomalies and multiple skeletal deformities such as short stature, kyphoscoliosis of the spine, cervical spine instability, clubfeet, and joint dislocations especially of the knee and hip joints. Musculoskeletal pain in a case of Larsen syndrome signals red flags as these patients have various congenital anomalies. However, myofascial trigger points could be a cause of chronic musculoskeletal pain in these patients. Myofascial trigger points are easy to diagnose and treat.

**Case Report:** We are presenting a 29-year-old man, diagnosed as a case of Larsen syndrome, who came to us with complaints of bilateral shoulder pain and right thigh pain for 4 years and 5 months, respectively. Examination of the patient revealed faulty joint mechanics along with the presence of trigger points in adjoining muscles. The trigger points were injected with local anesthetic followed by stretching and muscle-strengthening exercises, which led to pain relief.

**Conclusion:** In the present case report, we wish to highlight that myofascial pain could be a cause of pain and functional limitation in patients suffering from Larsen syndrome.

**Key words:** Larsen syndrome, myofascial trigger point

## BACKGROUND

Larsen syndrome is a rare, genetically heterogeneous (both autosomal dominant and recessive) genetic disorder characterized by craniofacial anomalies, joint dislocations, spine abnormalities, and other characteristically associated features (1). It was first described by Larsen and colleagues in 1950 and estimated to have an incidence of one in 100,000 live births (2). The characteristic features of the syndrome include multiple skeletal deformities such as short stature, kyphoscoliosis of the spine, cervical spine instability, clubfeet, joint dislocations especially of the knee and hip joints, and facial features comprised of marked frontal bossing, flattened nasal bridge, and wide-set eyes.

Patients usually seek medical care for problems arising from severe joint hypermobility and dislocation leading to reduction of function (3). Functional limitation occurs due to obvious mechanical factors, but also might

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Disclaimer: There was no external funding in the preparation of this manuscript.
Conflict of interest: Each author certifies that he or she, or a member of his or her immediate family, has no commercial association (i.e., consultancies, stock ownership, equity interest, patent/licensing arrangements, etc.) that might pose a conflict of interest in connection with the submitted manuscript.
Patient consent for publication: Consent obtained directly from patient(s).
Authors adhere to the CARE Guidlines for writing case reports and have provided the CARE Checklist to the journal editor.
Accepted: 2023-03-06, Published: 2023-07-31

be due to the pain accompanying such deformities. Conservative, medical, and surgical options addressing cervical spine instability (4), joint dislocations, and scoliosis have been described but issues regarding effective pain management and functional restoration have not been addressed.

Musculoskeletal pain in a case of Larsen syndrome signals red flags as these patients have multiple congenital anomalies. However, myofascial trigger points could be a cause of pain in these patients. Myofascial trigger points are a relatively common cause of chronic pain with a prevalence ranging from 10% to 13% in patients visiting pain medicine clinics. Myofascial trigger points are easy to diagnose and treat. In this report, we describe myofascial pain syndrome as a cause of chronic musculoskeletal pain and functional disability in a case of Larsen syndrome.

## CASE REPORT

A 29-year-old male patient diagnosed with Larsen syndrome was referred to the pain clinic from the department of medical genetics in our institute, a tertiary care center. He had short stature, flat facies, cylindrically shaped fingers, clubfoot, bilateral congenital dislocation of the tibia on femur, and kyphoscoliosis involving the thoracolumbar spine.

He was 78 cm tall and weighed 38 kg. The patient complained of bilateral shoulder

and right thigh pain for 4 years and 5 months, respectively. Shoulder pain on both sides was radiating into the lateral aspect of the arm up to the elbows and lateral aspect of the chest and was associated with difficulty in neck flexion. The pain was moderate in intensity with a Visual Analog Scale (VAS) score of 60 mm. The use of a hand pedal for a customized vehicle (hand-operated cycle) that required repeated circular motion at the shoulder girdle against resistance aggravated it. The pain was relieved by rest and analgesic intake, dropping to a VAS of 30 mm, although it never completely resolved. There were no accompanying neuropathic features.

The patient also had pain in the right anterior thigh radiating into the right anterior lower limb up to the dorsum of the foot with a VAS score of 70 mm, accompanied by occasional tingling. The pain increased on dorsiflexion of the foot and flexion of the knees. The patient scored 5 out of 27 on the Patient Health Questionnaire (PHQ-9) scales for diagnosis of depression, signifying mild depression.

He was not able to use his customized vehicle due to this pain, leading to functional limitation. On examination, trigger points in the interscapular region of both trapezius muscles and right thigh were detected. The myofascial trigger points were neutralized with wet needling using 10 mL of 0.2% lidocaine. Multiple twitches were elicited during needling of the trigger points. The patient was advised to do ice fomentation on the day of injection to reduce inflammation immediately following the needling. Thereafter, he was advised to follow a regular daily stretching and strengthening exercise program along with adjunctive therapy like hot fomentation and massage of the involved area. A short course of oral anti-inflammatory drugs for 2 days, followed by an oral combination of tramadol and paracetamol for use as and when required (SOS), was prescribed. Additionally, lab investigations such as vitamin D3 levels were ordered. A radiographic workup had been completed during the previous visit to the medical genetics department, which showed kyphoscoliosis of the thoracic spine.

The patient came for follow-up at one week and expressed significant relief in shoulder and thigh pain with a VAS score of 20 mm and 10 mm, respectively. He performed stretching exercises regularly and was now able to use his hand-operated cycle more effectively with minimal pain. Blood investigations revealed subnormal vitamin D3 levels with a value of < 20 ng/mL, which is the critical reference range. Thereafter, he was started on oral calcium and vitamin D3 supplementation.

Subsequent follow-up at 3 and 6 months showed a reduction of pain with the functional improvement compared to the first visit. He reported that pain was significantly lower, despite the use of the same vehicle. There was no continuous pain either in the shoulder or thigh. No other complications were noted. He was prescribed a set of stretching and strengthening exercises which were specially tailored according to his joints after analyzing the range of motion present at the shoulder, hip, and knee joints.

## DISCUSSION

The present case study describes the successful diagnosis and management of chronic musculoskeletal pain in a patient with Larsen syndrome. The pain relief resulted in improvement in the functional status of the patient.

Larsen syndrome is a rare congenital disorder with an estimated incidence of one in 100,000 live births and

nearly equal gender incidence (2). It is a genetically heterogeneous disorder with both autosomal dominant and autosomal recessive forms (5). The more frequent, autosomal dominant variety of Larsen syndrome is caused by mutations in the filamin B (FLNB) gene, which codes for a protein by the same name. This protein is important in the development of the skeleton, cell signaling, and normal cell growth (3). The less common autosomal recessive variety, however, is caused by mutations in the carbohydrate sulfotransferase 3 (CHST 3) gene (6). It is characterized by craniofacial anomalies and skeletal dysplasias (prominent forehead, depressed nasal bridge, malar flattening, and widely spaced eyes), club feet, congenital dislocation of hip, knee, elbow, and occasionally shoulder. Scoliosis and cervical kyphosis may also be present, which is associated with cervical myelopathy, short and broad phalanges, particularly of the thumb. Midline cleft palate, hearing loss, and pulmonary and cardiac involvement may also be present. People with Larsen syndrome can survive into adulthood. Intelligence is generally normal or mildly decreased, but not affecting function. Stature is mildly affected. The musculoskeletal involvement is in the form of major joint dislocations, hand and feet abnormalities, and spine malformations (3). Dysmaturity of collagen fibres and striking deficiency of dense mature collagen bundles is believed to be responsible for the musculoskeletal involvement in this syndrome.

We describe a case with a clinical phenotype of Larsen syndrome for the management of chronic musculoskeletal pain. The patient was a middle-aged man with short stature and normal intelligence leading a productive life; he presented with bilateral shoulder and thigh pain and was diagnosed to have trigger points (TrP). The etiology of myofascial trigger points is thought to be due to multiple factors such as body asymmetry, faulty posture, nutritional deficiencies, psychological factors, sleep disorders, endocrine dysfunction, and lack of exercise. TrPs result from muscle overuse injury, body asymmetry, or by keeping the muscle in disuse (7). Our patient had a history of hand-pedaled vehicle usage, which might have led to the formation of the said trapezius TrPs. Owing to the syndrome, he was also prone to joint dislocations, which caused severe malposition of the knee joint causing strain on the surrounding muscles and formation of TrPs.

Combination therapy including needling of TrP and medications, followed by physical rehabilitation, is beneficial and provides long-lasting relief in myofascial pain (7). TrP injections reduce pain and allow rehabilitation to be more successful. Our patient received TrP injection followed by a short course of medications and daily exercises, which resulted in a significant reduction of pain.

The role of vitamin D and calcium supplementation in skeletal dysplasias has been well established (8,9). Also, nutritional deficiencies play a big role in the causation of myofascial pain. Vitamin D deficiency has been implicated in a wide variety of muscular pains and myopathies. With a high index of suspicion, we evaluated the patient for vitamin D levels, which were found to be in the subnormal range and hence supplementation was started. Literature also reveals the significant improvement of muscular pains following calcium, bisphosphonate, and vitamin D supplementation (9-11). Our patient also received calcium and vitamin D supplementation, which probably helped early recovery and improvement in muscle function.

Review of the literature indicates the role of multidisciplinary management involving orthopedic surgeons, pediatricians, rheumatologists, and neurosurgeons in the holistic management of such conditions (9). However, we could not find previously reported literature on the presence of typical myofascial pain syndrome in Larsen syndrome. Conventional treatment options for myofascial pain include needling of the trigger point followed by stretching and strengthening exercises. But in the case of patients with such severe deformities, the ability to exercise may be restricted by the presence of malformed joints. Hence, exercises tailored to facilitate adequate stretching of the affected muscles need to be prescribed, as done in our patient. Efforts must be made to maximize muscle function and restore joint function to the range possible.

## CONCLUSION

Myofascial pain may be a cause of chronic musculoskeletal pain in patients with congenital dysplasias like Larsen syndrome. A combination of needling, medications, and physical rehabilitation is beneficial for the successful management of such patients.

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