ABSTRACT

Cramp fasciculation syndrome is mildest among all the peripheral nerve hyperexcitability disorders, which typically presents with cramps, body ache and fasciculations. The diagnosis is based on clinical grounds supported by electrodiagnostic study. We report a case of young male with two months’ history of body ache, rippling, movements over calves and other body parts, and occasional cramps. His metabolic workup was suggestive of impaired fasting glucose, radiologic work up (chest X-ray and ultrasound abdomen) was normal, and electrodiagnostic study was significant for fasciculation and myokymic discharges. He was started on pregablin and analgesics. To the best of our knowledge this is report first of cramp fasciculation syndrome from Pakistan.

INTRODUCTION

Cramp fasciculation syndrome was first described in 1991 and is mildest among all the peripheral nerve hyperexcitability disorders, which include Issac’s syndrome (acquired neuromyotonia with hyperhidrosis) and Morvon’s syndrome (acquired neuromyotonia with autonomic dysfunction and encephalopathy) in addition to isolated cramp fasciculation syndrome. Cramps and fasciculations are hallmark features of the disease and other features include muscle stiffness and body ache. The diagnosis is clinical, supported by electrodiagnostic study that also helps to rule out underlying neuromuscular disorders which may secondarily lead to hyperexcitability. Membrane stabilization with medications like carbamazepine is mainstay of therapy but sometimes immunosuppression or pregablin are required.

CASE SUMMARY

A young male, resident of mountainous region of Balochistan, presented to neurology clinic with two months history of generalized body ache and discomforting involuntary rippling movements over calves and other body parts. He also gave history of occasional muscle cramps but he denied any symptoms to suggest autonomic dysfunction, weight loss, joint pain, joint swelling, cough, hemoptysis, hematemesis and bleeding per rectum. There was no history of mental status changes, seizures or memory impairment. He also denied any sensory symptom (apart from pain) and weakness or wasting. His past history was significant for vitamin D deficiency for which he received parenteral Vitamin D and his latest vitamin D level was 125. He was nonsmoker and there was no history of any other addiction either. His clinical examination was significant for continuous undulating movements over both calves and to lesser extent over upper arms. His CBC was significant for Hb of 17.2 with hematocrit of 51.6. ESR was 8, CRP 0.06, sodium 145, magnesium 2.1, calcium 8.9, creatinine 0.8, B12 334, TSH 1.7, free T4 1.31, ALT 71. HBsAg, anti HCV antibodies, ANA, AMA, ASMA and anti DsDNA were also negative. Ultrasound of whole abdomen and a chest X-ray were also normal. Routine nerve conduction studies were normal, a repetitive nerve conduction study from tibial nerve at 1, 2, 5 and 10 Hz revealed after potentials which lasted 80-100ms. Needle EMG revealed fasciculations and some myokymic discharges from gastrocnemius, FDI and deltoid. There was no other spontaneous activity including neuromyotonic discharges. The motor unit potentials morphology, recruitment and firing pattern were normal. A probable diagnosis of cramp fasciculation syndrome was made and he was started on pregablin. In about two weeks his symptoms including undulating movements over calves improved by 70%. He was referred to hematologist for evaluation of high hemoglobin, hematocrit and red cell count. He was advised for JAK-2 mutation analysis and erythropoietin levels. His JAK 2 mutation was negative and erythropoietin level was normal.

DISCUSSION

Pain and cramps are common symptoms and have variable
causative factors including neuromuscular disorders, electrolyte imbalance, systemic disorders, fibromyalgia. Fasciculations suggest lower motor neuron disorder including motor neuron disease. Additionally isolated syndrome of benign fasciculations is also well reported.\textsuperscript{3} Combination of cramps and fasciculations with myalgia, stiffness especially with features of autonomic dysfunction is very suggestive of peripheral nerve hyperexcitability.\textsuperscript{2} These disorders are considered to be secondary if there is underlying neuromuscular disorder and primary if there is no such underlying disorder. Primary hyperexcitability disorders have been found to be associated with autoimmunity especially presence of voltage gated potassium channel (VGPC).\textsuperscript{2,4} Recently 50% of the patients in a cohort of patients with chronic pain found to have antibodies to VGKC complex.\textsuperscript{5} Our patient had subacute history of myalgia, muscle stiffness, occasional cramps and undulating movement over calves and upper arms which were bothering him. His work up was significant for impaired fasting glucose and high hematocrit. There was no evidence of malignancy, thyroid dysfunction or autoimmune disorders. JAK mutation was negative and erythropoietin levels were normal. Nerve conduction studies did not reveal any evidence of neurogenic or myopathic disorder. Absence of neuromyotonic discharges on EMG and absence of autonomic dysfunction and normal higher mental functions argue against Isaac’s or Morvan’s syndrome. Significant fasciculation and syndrome discharges were noted in absence of other neurogenic or myopathic features suggest cramp fasciculation syndrome a strong possibility in our case. RNS of tibial nerve, in our case, did not reveal significant after potentials. Recently a series from mayo clinic also did not determine its utility in diagnosing cramp fasciculation syndrome, however, it might be useful to discriminate seronegative from seropositive cases.\textsuperscript{2} this may have therapeutic implications as immunotherapy may be helpful in seropositive patients. Mainstay of treatment for CSF is membrane stabilizing agents like carbamazepine, gabapentin or pregabalin.\textsuperscript{2} We started our patient on pregablin and within two weeks his symptoms including the rippling movement in calves improved by 70%. To the best of our knowledge this is first report on cramps fasciculation syndrome from Pakistan. This should be considered in patients with myalgia, cramps and fasciculations as appropriate treatment can reduce patient’s agony.

**REFERENCES**


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**Bhojo Khealani:** Study concept and design, protocol writing, data collection, data analysis, manuscript writing, manuscript review