CASE REPORT

Munchmeyer's disease—a rare case report and review of literature

¹S B Sheth, ^{1,2}S N Santhosh Kumar, ²S Sabhlok and ²M Singh

¹Gujarat Cleft and Craniofacial Research Institute (The GCCRI), Ahmedabad, Gujarat, India; ²Dr D Y Patil Dental College and Hospital, Pune, Maharashtra, India

Munchmeyer's disease is a rare variety of myositis ossificans. It is a significant clinical entity, diagnosing and understanding its pathophysiology can save a patient from spurious medical workups and anxiety of a suspected neoplasm. The number of authentic cases of this disease in the literature is small. The purpose of this article is to present a case of Munchmeyer's disease and review the literature for the same to date. The rare incidence, obscure aetiology, unknown definitive treatment plan and poor prognosis make this condition an enigma.

Dentomaxillofacial Radiology (2014) **43**, 20140022. doi: [10.1259/dmfr.20140022](https://doi.org/10.1259/dmfr.20140022)

Cite this article as: Sheth SB, Santhosh Kumar SN, Sabhlok S, Singh M. Munchmeyer's disease—a rare case report and review of literature. *Dentomaxillofac Radiol* 2014; **43**: 20140022.

Keywords: Munchmeyer's disease; myositis ossificans; myositis ossificans progressiva

Introduction

Myositis ossificans refers to a condition in which ectopic bone is formed within muscles and other soft tissues. Three types of myositis ossificans are distinguished: myositis ossificans circumscripta, myositis ossificans traumatica and myositis ossificans progressiva (Munchmeyer's disease), a rare widespread syndrome.¹

Munchmeyer's disease is a rare disease of the locomotor system, which occurs during the period of growth and insidiously but inevitably brings about immobilization. This condition is characterized by the ossification of muscles, tendons, fasciae and aponeuroses and by the occurrence of exacerbations and remissions in the course of the disease and of congenital bony anomalies.²

The disease usually begins during the first two decades of life and is gradually progressive, resulting ultimately in the complete immobilization of nearly every joint of the body.³ Treatment of this disorder has so far proved unsuccessful. The prognosis of the disease is poor.³ The rare incidence, obscure aetiology, unknown definitive treatment plan and poor prognosis make this condition to be presented and help to disclose the mystery behind this disease.

Thus, this article presents a brief review and a case report of a 12-year-old male child suffering from Munchmeyer's disease.

Case report

A 12-year-old male came with a complaint of difficulty in opening the mouth for 4 months. There was no history of consanguineous marriage in the family. The birth and gestation history were normal, except that the parents noticed a small-sized great toe in both feet. No other member of the family was similarly affected. Before the age of 6 years, the patient had a completely uneventful childhood.

The patient's parents attributed the onset of the illness to an accident of a minibus in which he was travelling 6 years ago. There was a single hard swelling and pain in the neck on the left side. He also had difficulty in opening his mouth that was noticed by his parents as he was struggling to eat. Eventually, the neck swelling became soft and disappeared within 2 years; however, the difficulty in opening his mouth worsened. He further developed multiple new bony hard swellings in his back, of which few regressed.

On examination, the patient was moderately built and poorly nourished, and the general physical examination revealed pallor. Local examination of the face showed facial asymmetry with the chin deviated to the right (Figure 1a). The mouth opening was reduced to about 1.5 mm (Figure 1b). A worm's eye view revealed a scar over the chin, suggestive of trauma. Temporomandibular joint movements were reduced on the right side. The patient was provisionally diagnosed as having temporomandibular joint ankylosis of the right side.

Routine blood and urine investigations were performed, and all the values were within normal limits. The radiological investigations were performed with the chest radiograph being normal, and a CT scan of the temporomandibular joints showed normal joints bilaterally. However, a radiodense mass extending from the lower border of the right zygoma to the angle of the mandible was seen, suggestive of ossified masseter muscle (Figure 2). This finding along with a history of trauma led us to the diagnosis of myositis ossificans traumatica.

On thorough probing into the history, the patient's father said that the patient also had swelling and pain in the lower back region for 1 month followed by limitation of movements in the same region. The patient was referred to the orthopaedic surgeon for further management.

A full body examination revealed ossifications in the lumbosacral, thoracic and bilateral scapular regions (Figure 3). Slightly restricted neck movement towards the right side and restricted extension of the neck were noted. Also, microdactyly of the great toe on both the feet was seen (Figure 4a). The thumb of the right hand was shorter relatively.

The radiographs of the hand, the foot and lumbosacral regions were taken. The radiograph of the foot

confirmed the clinical examination, showing microdactyly of hallux (Figure 4b). The radiograph of the lumbosacral vertebrae showed calcification of the intervertebral disc region (Figure 5). The patient's father refused to undergo further investigations.

All these features led us to conclude the diagnosis as myositis ossificans progressiva. We have advised regular physiotherapy to the patient and kept him under observation. The other parts of the body are not yet affected.

Review of literature and discussion

Munchmeyer's disease is a rare variety of myositis ossificans. Myositis ossificans is an extra-osseous non-neoplastic but pathological cartilage and bone formation that occurs in soft tissues, especially within the muscles that normally do not ossify. Munchmeyer's disease is a disabling genetic condition that leads to the formation of a second (heterotopic) skeleton and is the most catastrophic disorder of heterotopic ossification in humans.⁴ Throughout childhood and early adult life, this disease progressively immobilizes all of the joints of the normotopic skeleton, rendering movement impossible. This condition has been described in horses, dogs and pigs.^{3,5}

According to Pack and Braund,⁶ this condition was first described by Guy Patin in 1692 when he wrote about "the woman who turned into wood". Von Duschin 1868 first used the term "myositis ossificans progressiva".⁵ Munchmeyer in 1869 first gave a comprehensive description of the disease with an account of 12 cases; hence, the eponym "Munchmeyer's disease".³ Rosenstirn⁵ thought that a better name would be "fibrocellulitis ossificans progressiva". Fairbank⁷ suggested the

Figure 1 (a) Frontal facial view showing facial asymmetry and (b) difficulty in opening the mouth.

Figure 2 Three-dimensional CT scan showing an ossified masseter muscle. RAF, relative attenuation function; RP, right posterior.

term “fibrositis ossificans progressiva” and McKusick,⁸ following Bauer and Bode, favoured “fibrodysplasia ossificans progressiva”.³

The aetiology of Munchmeyer's disease remains obscure. Trauma may be a precipitating factor, as was reported in our case. However, in view of the fact that 90% of the cases is associated with congenital anomalies, most frequent being microdactyly of the great toe, it has been suggested by many authors that the disease is either congenital or inherited. Koontz mentioned that Rosenstirn reports three instances in which heredity appears to be a factor.⁹ In one instance, the grandfather, father and three sons all had microdactyly of hallux. The other evidence is the report of a hereditary case by Burton-Fanning and the occurrence of the disease in

Figure 3 Ossifications seen in the lumbosacral, thoracic and bilateral scapular regions.

homozygotic twins reported by Vastine, Vastine and Aaron and Eaton, Conkling and Daeschner.^{4,9}

Koontz mentions some theories of causation, *viz.* retention of calcium; congenital pre-disposition towards ossification of intramuscular connective tissue, fascia and aponeurosis; extravasation of blood into tissue; inflammatory process of intermuscular connective tissue; and disturbances of embryonic development.⁹ Rolleston in 1901 believed the condition to be a congenital defect of the mesoblast.² However, this was disputed by Rosenstirn⁵ who postulated the condition to be a congenital defect of small blood vessels and Lutwak¹⁰ who considered the defect in the osteoblastic enzymatic activity to be the culprit. It is suggested that in Munchmeyer's disease, the process is a metaplasia of fibrous tissue to cartilage and bone.²

The pathophysiology of soft-tissue calcification is not completely understood, but the location and appearance of the calcification may suggest the diagnosis. The genetic mutation responsible for this has recently been identified. The mutation affects a bone morphogenic protein receptor called activin receptor type IA and has been mapped to chromosome 2q23–24.¹¹ Bone morphogenic proteins are regulatory proteins that play a critical role in embryology, bone formation and fracture healing.¹² Activin receptor type IA is an important signalling control protein in chondrocytes of the growth plate and cells of the skeletal muscle. Enhanced activation of activin receptor type IA is seen in fibrodysplasia ossificans progressiva.¹¹

It has been shown that this disease affects the interstitial tissues of muscles, fascia, tendon, ligaments and aponeuroses and not the muscle fibres themselves. Owing to these factors, it has been suggested by McKusick⁸ that the degeneration of muscle fibres seen in the advanced stages of the disease is actually a secondary atrophy owing to pressure and disuse, the major defect being the dysplasia of fibrous and connective tissue. In view of these pathological findings, it has been suggested by McKusick⁸ and Mair¹³ that the term “myositis ossificans progressive” is a misnomer and that the disease should be more correctly referred to as “fibrodysplasia ossificans progressiva”.

This disease has an onset usually in the first 10 years of life; it may begin *in utero*, but rarely after 20 years of age. In the above case, the onset was said to be by 5–6 years of age. The first symptom is commonly torticollis, and the sternomastoid muscle is usually involved; concurrently or soon after, there is involvement of the masseter muscle. Even in our case, a single hard swelling and pain in the neck and difficulty in opening the mouth were noted. This is usually followed by involvement of the muscles of the shoulder girdle, upper arm and then the pelvic girdle. The heart, diaphragm, larynx, tongue, sphincter and eye muscles are not involved. The abdominal muscles are usually spared.³

The first sign of involvement is often heat, oedema and pain in a muscle; this may subside in a few weeks, followed by a doughy swelling, which in a few months

Figure 4 (a) Microdactyly of the great toe bilaterally and (b) a radiograph of the foot showing microdactyly of hallux.

may ossify. Eventually, tendons, fascia and ligaments are replaced by bone. The characteristic remissions and exacerbations, as described by Garrod,¹⁴ are being demonstrated in this case.

There is almost always pathognomonic microdactyly of the great toe, with suppression of the proximal phalanx. Helferish in 1879 first noted the association of this disease with microdactyly of hallux.¹⁶ Although this pathognomonic feature was missed in our initial examinations, it was later diagnosed by discussing the case with an orthopedician. There may be other anomalies,

exostoses, a broad neck of the femur, hypogenitalism, the absence of lobules of the ears and deafness.^{10,15}

The prognosis of the disease is grave. Death may be due to respiratory infection or starvation. In patients with Munchmeyer's disease, constriction of chest wall movements is compounded by abdominal wall calcification in the late stages and therefore any minor infection of the respiratory tract becomes a hazard to life. Starvation is caused by trismus owing to ossification in the masseter muscles. Fairbank⁷ wrote that severe crippling occurs in "a few years" to 30 or 40 years. Campbell¹⁶ quoted a case of a male who died of the condition at 70 years, and Rosenstirn⁵ quoted a case of a male dying in 1744 at 60 years. According to Rosenstirn,⁵ most die before the age of 15 years.

The absence of any abnormality of blood chemistry is in accordance with the findings of the majority of investigators.^{3,4}

There is no treatment for the Munchmeyer's disease.¹⁷ However, knowledge of the genetic mutation will now provide hope for drug development and gene therapy for these patient's misery.¹⁸ Innumerable forms of treatment have been tried all to no avail. They include local blisters, iodine, mercury, colchicum, local bleeding, sarsaparilla, nitric acid, lactic acid, phosphoric acid, thyroid extract, thiosamine, sodium citrate parathyroid extract, low calcium diet, vitamin B1, vitamin E, disodium hydrogen phosphate, beryllium, androgens, ethylenediaminetetraacetic acid, ketogenic diet, ultrasound, antimitotic agents, oestrogens, thymectomy, radiotherapy, radium implants and surgical removal of plaques.^{5,19-22} Ethane-1-hydroxy 1, 1-diphosphate showed beneficial effects on two acute cases of this disease.²³ The role of corticosteroids is uncertain.²⁴⁻²⁷ Because of such great uncertainties regarding the treatment, we have just placed the patient under observation and regular physiotherapy.⁴

Surgery, till now, does not seem to have any role in the management of this condition.

Conclusion

A case of Munchmeyer's disease is reported in a boy aged 12 years old, with notes on theories of aetiology, clinical features and differential diagnosis, and a mention

Figure 5 A radiograph of lumbosacral vertebrae showing calcification of the intervertebral disc region. R, right.

is made of the numerous but disappointing varieties of treatment. Research into the aetiology and

physiopathology of this disorder should be continued to search for causal therapies.

References

1. Bossche LV, Vanderstraeten G. Heterotopic ossification: a review. *J Rehabil Med* 2005; **37**: 129–36.
2. Griffith G. Progressive myositis ossificans: report of a case. *Arch Dis Child* 1949; **24**: 71–4.
3. Illingworth RS. Myositis ossificans progressiva (Munchmeyer's disease) brief review with report of two cases treated with corticosteroids and observed for 16 years. *Arch Dis Child* 1971; **46**: 264–8.
4. Letts RM. Myositis ossificans progressiva: a report of two cases with chromosome studies. *Can Med Assoc J* 1968; **99**: 856–62.
5. Rosenstirn J. A contribution to the study of myositis ossificans progressiva. *Ann Surg* 1918; **68**: 485–520.
6. Pack G.T, Braund RR. The development of sarcoma in myositis ossificans. *J Am Med Assoc* 1942; **119**: 776–9.
7. Fairbank HAT. Myositis ossificans progressiva. Synonyms-fibrosis ossificans progressiva. *J Bone Joint Surg* 1950; **32B**: 108–16.
8. McKusick VA. *Hereditary disorders of connective tissue*. 2nd edn. St. Louis: Mosby; 1960.
9. Eaton WL, Conkling WS, Daeschner CW. Early myositis ossificans progressiva occurring in homozygotic twins. *J Pediatr* 1957; **50**: 591–8.
10. Lutwak L. Myositis ossificans progressiva. Mineral, metabolic and radioactive calcium studies of the effects of hormones. *Am J Med* 1964; **37**: 269–93.
11. Fiori JL, Billings PC, de la Peña LS, Kaplan FS, Shore EM. Dysregulation of the BMP-p38 MAPK signaling pathway in cells from patients with fibrodysplasia ossificans progressiva (FOP). *J Bone Miner Res* 2006; **21**: 902–9. doi: 10.1359/jbmr.060215
12. Chen D, Zhao M, Mundi GR. Bone morphogenic proteins. *Growth Factors* 2004; **22**: 233–41.
13. Mair WF. Myositis ossificans progressiva. *Edin Med J* 1932; **39**: 13–36.
14. Garrod AE. The initial stage of myositis ossificans progressiva. *St Bartholomew's Hospital Reports* 1908; **43**: 43–9.
15. Ludman H, Hamilton EBD, Eade AWT. Deafness in myositis ossificans progressiva. *J Laryngol Otol* 1968; **82**: 57–63.
16. Campbell RK. Myositis ossificans progressiva. *Radiol Rev* 1933; **55**: 153–6.
17. Kitterman JA, Kantanie S, Rocke DM, Kaplan FS. Iatrogenic harm caused by diagnostic errors in fibrodysplasia ossificans progressiva. *Pediatrics* 2005; **116**: e654–61. doi: 10.1542/peds.2005-0469
18. Kaplan FS, Glaser DL, Pignolo RJ, Shore EM. A new era for fibrodysplasia ossificans progressiva: a druggable target for the second skeleton. *Expert Opin Biol Ther* 2007; **7**: 705–12. doi: 10.1517/14712598.7.5.705
19. Klemperer FW, Miller M, Hill CJ. The inhibition of alkaline phosphatase by beryllium. *J Biol Chem* 1949; **180**: 281–8.
20. Liberman UA, Barzel U, De Vries A, Ellis H. Myositis ossificans traumatica with unusual course. Effect of EDTA on calcium, phosphorus and manganese excretion. *Am J Med Sci* 1967; **254**: 35–47.
21. Esenwein SA, Sell S, Herr G, Gaissmaier C, Bamberg M, Mollenhoff G, et al. Effects of single-dose versus fractionated irradiation on the suppression of heterotopic bone formation—an animal model based follow-up study in rats. *Arch Orthop Trauma Surg* 2000; **120**: 575–81.
22. Sautter-Bühl ML, Liebermeister E, Nanassy A. Radiotherapy as a local treatment option for heterotopic ossification in patients with spinal cord injury. *Spinal Cord* 2000; **38**: 33–6.
23. Bassett CAL, Donath A, Macagno F, Preisig R, Fleisch H, Francis MD. Diphosphonates in the treatment of myositis ossificans. *Lancet* 1969; **2**: 845–6.
24. Riley HD, Jr, Christie A. Myositis ossificans progressiva. *Pediatrics* 1951; **8**: 753–67.
25. Evans PR. Myositis ossificans progressiva treated with cortisone and ACTH. *Great Ormond St J* 1952; **2**: 56.
26. Lockhart JD, Burke FG. Myositis ossificans progressiva. *Am J Dis Child* 1954; **87**: 626–35.
27. Beco V. An attempt at treatment of progressive myositis ossificans with the disodium salt of EDTA. *Hum Genet* 1967; **4**: 320–51.