



The Stone Man: Myositis Ossificans Progressiva, About a Case

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Abstract

Progressive ossifying myositis is a rare genetic disease. A 10-year-old boy is reported to have multiple painful swellings of spontaneous and progressive onset in the back and upper limbs. These swellings were associated with a febrile. The radiological and computed tomography aspects were largely sufficient to confirm the diagnosis. The treatment was purely medical. The development was marked by the appearance of other ossifications of fascias and muscles leading to very disabling joint stiffness. Circumstances of discovery, epidemiological, etiopathogenic, diagnostic, evolutionary and therapeutic aspects are discussed through a literature review.

Keywords: Congenital; Child; Myositis ossificans; Radiology

Introduction

Progressive ossificans myositis (MOP), also known as progressive ossificans fibrodysplasia (FOP) or Munchmeyer's disease or stone man's disease, is a rare genetic disorder with autosomal dominant inheritance characterized by a congenital malformation of the big toes and progressive postnatal development of heterotypic ossifications of tendons, ligaments, fasciae, connective tissues and skeletal muscles. Initially described by Patin in 1648, then by Munchmeyer in 1869. Beginning generally before the age of 10, this heterotypic ossification evolves by pushing along a cranio-caudal and dorso-ventral path leading to a disabling joint stiffness. Patients see themselves trapped in a "second skeleton" [1].

ACVR1 (active in receptor type 1A) is the gene responsible for FOP, identified in 2006 by KAPLAN and SHORE. It is located on the long arm of chromosome 2 and it encodes the receptor for BMP (Protein Bone Morphogenetic) protein that induces the formation of bone and cartilage. ACVR1 is common to any individual but the disease only occurs if one of the two copies of this gene contains an abnormality [2].

We report a case of MOP observed in 10-year-old boys to show the epidemiological, etiopathogenic, diagnostic and therapeutic aspects.

Submitted: 10 May 2019 | **Accepted:** 15 June 2019 | **Published:** 18 June 2019

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Citation: Boukhalit H, Lahfidi A, Allali N (2019) The Stone Man: Myositis Ossificans Progressiva, About a Case. JSM Radiol Radiat Ther 5: 4.

Observation

Unjeune garçon de 10 ans sans antécédents familiaux particuliers, présentant depuis l'âge de 4 ans des tuméfactions douloureuses para vertébrales négligées, évoluant vers l'installation d'une cyphose dorsale, l'ankylose des deux membres supérieurs et une impotence fonctionnelle invalidante à l'examen clinique.

Le scanner thoraco-abdomino-pelvien mettait en évidence des ossifications extra-squelettiques de la musculature para vertébrale depuis l'occiput jusqu'au coccyx, ces ossifications présentant des ramifications bilatérales et asymétriques passant par les régions sous scapulaires, fusionnant avec les extrémités supérieures des humérus, arrivant à la paroi thoracique antérieure, réalisant l'aspect de l'homme en pierre, le diagnostic de myosite ossifiante progressive a été retenu et a été confirmé par l'étude génétique.

10-year-old boy with no consanguineous parents and none of the family members problem, presenting since the age of 4 neglected paravertebral painful swelling, moving towards the installation of a dorsal kyphosis, the ankylosis of both upper limbs and disabling functional impotence, Laboratory blood tests for calcium, phosphorus, alkaline phosphatase, urea, creatinine and parathyroid hormone found normal values.

The thoraco-abdomino-pelvic CT scan was performed without IV contrast (Figure 1) and showed extra skeletal ossifications in the paravertebral musculature from the occiput to the coccyx, these ossifications having bilateral and asymmetrical ramifications through the sub-scapular regions, merging with the upper extremities of the humerus, arriving at the anterior chest wall, realizing the appearance of the stone man, the diagnosis of progressive myositis ossificans was retained and was confirmed by the genetic test (Figure 2).

Discussion

MOP extremely rare genetic condition, affects on average 1 person in 2 million. Described for the first time in 1648 by PATIN [3]. This condition is mainly described in young children and affects both sexes equally: 41% of MOP is screened before the age of 2 years, 80% before 10 years and 95% before 15 years [4].



Figure 1 Clinical appearance of the trunk showing bone deformity of dorsal kyphosis, stiffness of the whole trunk with skeletal and extra-skeletal subcutaneous castings..

Our case is in harmony with the literature, the first symptoms having appeared at the age of 4 years. Ethnicity, race, gender, or geography do not appear to be predisposing factors, however a genetic factor is likely with the occurrence of a spontaneous mutation of the ACVR1 gene on chromosome 4 of autosomal dominant inheritance. Encoding a BMP4 protein receptor involved in the growth and modeling of bone [5]. The symptomatology of the MOP associates a fever with the appearance of inflammatory swellings affecting the connective tissues and the muscles. These will ossify and gradually spread to all areas of the body. This process initially affects the neck and paravertebral muscles with extension in a proximo-distal and cranio-caudal pattern [6]. The attacks will then touch the shoulders and then the lumbar spine and hips. Later distal joints can be reached by the 3rd decade of life. However according to Munchmeyer, muscles that do not fit on the skeleton by their two ends are spared. This explains the respect of the ocular muscles, the diaphragm, the tongue, the pharynx, the larynx and the smooth muscles (Figure 3) [6].

Our case is similar to the target muscle and lesion progression literature and tends to confirm Munchmeyer's hypothesis. On the other hand, 70-100% of the cases show mainly bone defects in the thumbs and the big toes [6]. They are clinically translated into a micro typing and a bilateral Hallux valgus. Other more rare and varied malformations are described such as vertebral, epiphyseal, rotulian, calcaneous spines, the femoral cervix, the fifth fingers or thickening of the inner cortical of the tibia. Does not exist in our case. Patients with atypical forms of FOP have been described, either the classic signs of FOP are present, with one or more atypical signs (for example: inters current aplastic anemia, craniopharyngioma, infantile glaucoma or stunting), either one or both cardinal signs of FOP show major variations for example, large normal toes or severe reduction of fingers

(FOP variants). Due to ankylosis of the costo-vertebral joints and vertebral deformation, patients end up developing restrictive respiratory insufficiency with atelectasia. Pneumonia and right heart failure can be fatal. Biological examinations are part of the diagnostic elements of the MOP but they have little therapeutic interest, evolution and prognosis [7]. The minimum number of biological examinations to be requested is related to the bone metabolism including the phosphocalcic balance and the level of PAL and that relating to the inflammatory process including the VS and the blood count [7].

Conventional radiology is the key to diagnosis by showing evocative images such as ectopic cortical calcification of affected and advanced muscles, bone bridges between different parts of the skeleton with a true ectopic skeleton. It also shows typical congenital bone malformations. Other imaging means are not necessary for diagnosis, especially at an advanced stage. The scanner makes it possible to better analyze the ossifications as well as their extent thanks to the multi planarian reconstructions as observed in our patient. MRI and scintigraphy may show lesions that are not yet ossified [8]. The diagnosis of FOP is radio-clinical and does not require a biopsy that may be the starting point for ectopic ossification and may be misleading because of the heterogeneous nature of the lesions [8].

Several differential diagnoses can be mentioned especially at the beginning of the disease and when the toes and fingers are not examined. Ossifying metastases, ossified hematomas, calcifying tendinopathies, exotic diseases, circumscribed ossifying posttraumatic myositis that is usually limited to a



Figure 2 Thoracic CT scan with reconstruction: significant extra skeletal and skeletal ossification engaging the posterior face of the trunk, merging with the upper ends of the humerus, associated with other parasternal and paravertebral cortical ossifications bilaterally and a dorsal kyphosis..

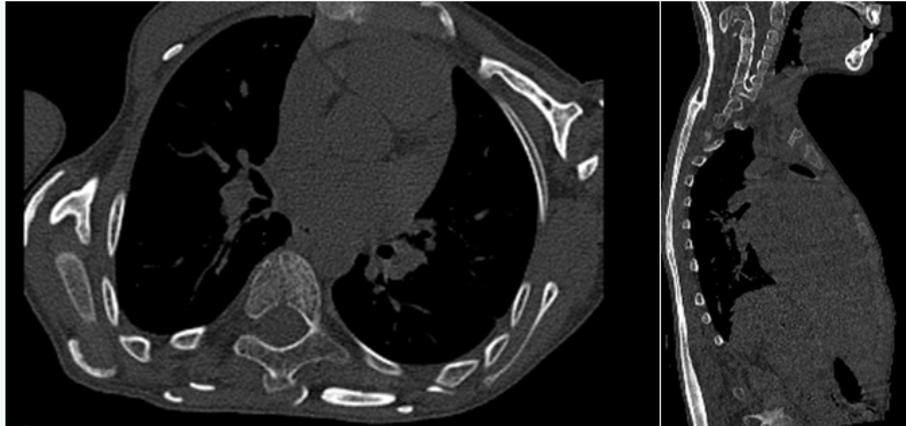


Figure 3 Thoracic CT scan with the axial and sagittal planes: show extra-skeletal ossifications of the para-vertebral musculature, with bilateral and asymmetrical ramifications through the sub-scapular regions, merging with the upper ends of the humerus, reaching the anterior chest wall.

single location, delimited, and painful, that occurs as a result of trauma. Soft tissue sarcomas may be discussed on biopsy results of early lesions [9,10]. Hereditary osteodystrophy of ALBRIGHT is characterized by ossification in muscle and connective tissue, but is more marked in subcutaneous fat and associated with pseudohypoparathyroidism [11]. Ankylosing spondylitis may be referred to ankylosis of the spine and sacroiliac joints [12]. The natural history of this condition is characterized by inflammatory outbreaks of 2 to 3 weeks, interspersed by periods of latency more or less long. The development may be enmeshed by the occurrence of several thromboembolic, neurological, skin, infectious or respiratory complications. Its prognosis depends mainly on the occurrence of respiratory failure at a late stage.

To date, no treatment has proven effective. However, several therapeutic means are proposed in the management of MOPs [3]. Mild physiotherapy is proposed for analgesic purposes during surges. Drug treatment includes corticosteroids, mastocyte inhibitors, cyclooxygenase 2 inhibitors, non-steroidal anti-inflammatory drugs, amino biphosphonates, BMP antagonists, anti-angiogenic agents or retinoids with still random and unsatisfactory results. Their use must be weighed against the potential severity of their side effects. There is little room for surgery, especially since anesthesia in these patients is difficult because of spinal rigidity and mandibular fixation [13,14]. Surgical removal of these ossifications to mobilize the joints is a new trauma that promotes the development of additional heterotopic ossifications. The sole purpose of the surgery is to correct the vicious attitudes to give stiffness in a most favorable position possible. In front of the limits of medical and surgical treatments, prevention becomes very important. Several measures can limit the evolution of the disease: prevention of trauma, mandibular blockage, rehabilitation medicine and physiotherapy with respiratory physiotherapy [15].

Conclusion

An extremely rare condition, MOP should be reported in any child with an unexplained inflammatory mass, associated with heterotopic ossification of the soft parts and congenital bone

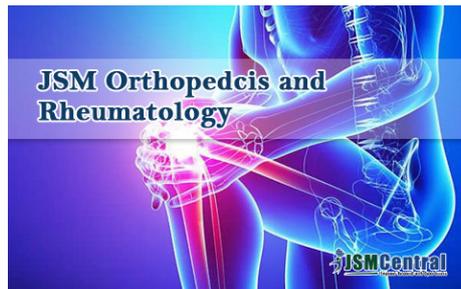
abnormalities. His diagnosis is radio-clinical. The development is marked by the appearance of particularly disabling joint stiffness. It can also involve the vital prognosis of general complications. No effective preventive or curative treatment is available to date. Nevertheless, several molecules are currently being tested.

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