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Severe evolution of Thevenard's disease: Case report

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Abstract: Thévenard's disease is a rare familial ulceromutilative acropathy of autosomal dominant inheritance. It starts at the level of the feet where it remains the most often the only manifestation, to extend exceptionally and then later at the level of the hands. Its evolution is generally towards aggravation by successive shoots, separated by phases of remission more or less long (sometimes several years). Nerve osteoarthropathy and infectious complications are the main complications of Thevenard's disease. We report the case of a 31-year-old woman, followed since the age of 6 years for Thévenard's disease, complicated by plantar perforations with repetitions of both feet and an osteoarthropathy of interest to both lower limbs, the upper limbs are intact.
Keywords: Thevenard's disease, Sensory neuropathy, Ulcero-mutilating acropathology**.**

INTRODUCTION

Thevenard's disease is a rare familial ulceromutilative acropathy, affecting both sexes. It is a pathology of the peripheral nervous system which starts at the level of the feet where it remains the most often the only manifestation, to extend exceptionally and then later at the level of the hands. It causes skin ulcers that progress slowly to bone deformities and then osteoarticular destruction due to thermoalgesic anesthesia of distal topography [1,2].

CASE REPORT

This is a 31-year-old woman with two sisters followed for Thévenard's disease and a 29-year-old brother with no symptomatology. At the age of 6, the patient was hospitalized for a plantar perforating disease of the right foot developed following a wound with a dirty nail. The radiographic assessment had not shown any bone involvement (Figure 1). The patient had debridement in the operating room and the biopsy of the ulcer had failed to show any specific lesions. The evolution was marked by the persistence of the lesion which indicated the establishment of a plantar flap a year later.

At the age of 7, our patient consulted for the appearance of a deep ulcer on the right foot with the bare bone and another at the outer edge of the left heel. Electrophysiological examination showed an alteration of sensory evoked potentials, and anatomopathological study of the long saphenous nerve showed severe axonal loss of myelinated fibers associated with less loss of amyelinic fibers.

The evolution was marked, 3 years later, by the installation of an osteoarthritis of the right ankle (Figure 2). At 9 years, and without any notion of straight trauma, the patient had a closed fracture of her left leg, for which she received orthopedic treatment (Figure 3).

At 14 years, a marked deformation of the two ankles was established, with development of a plantar perforating right and another opposite the left external malleolus, associated with a deformation of the left leg. On the right side, the radiographic examination showed complete destruction of the bones of the tarsus and the lower extremity of the two bones of the leg, and on the left side, the appearance of neglected pseudarthrosis of the malleolar fracture, as well as the shin producing a flexion of 15 ° (Figure 4). The patient benefited from a reaxation of the leg and osteosynthesis by an external fixator, with a good evolution (Figure 5).

At 15 years old, significant offsetting of the left foot was partially corrected by an external based valgus fixation osteotomy with two staples.

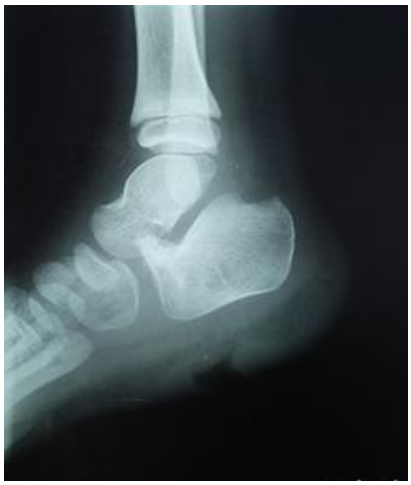
At 16 years old, the patient underwent arthrodesis of her right ankle.

At 17, she presented a right varnished genu, corrected with a valgus osteotomy (Figure 6).

At 28 years old, our patient was hospitalized for an important plantar perforation on the right (Figure 7).

Accompanied by a bone destruction interesting the bones of the back foot and the lower quarter of the two bones of the leg (Figure 8), requiring the realization of an amputation of the right leg. Healing of the amputation stump was obtained. 2 years later, our patient was rehospitalized and operated for an infection in the amputation stump related to the artificial limb (Figure 9).

At present, the patient does not present malformations in the hands. The neurological examination is without particularity at the level of the upper limbs, whereas it objectifies, at the level of the lower limbs, an abolition of the osteo-tendinous reflexes (patellar and achilles), the surface sensitivity thermoalgic and tactile as well as deep arthrokinetic sensitivity. The higher functions are retained.



Fig−1 *:* Standard X-rays of the right ankle (at 6 years old) showing a plantar perforating disease with no bone lesions.

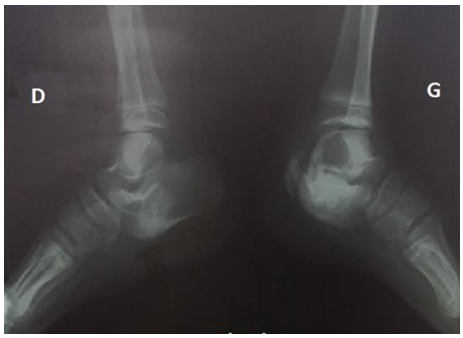


Fig-2: Standard radiography at the age of 8 showing

- **Left: an image of osteoarthritis of the left ankle with bone involvement of the hind foot;**
- **Right: a straight MPP.**

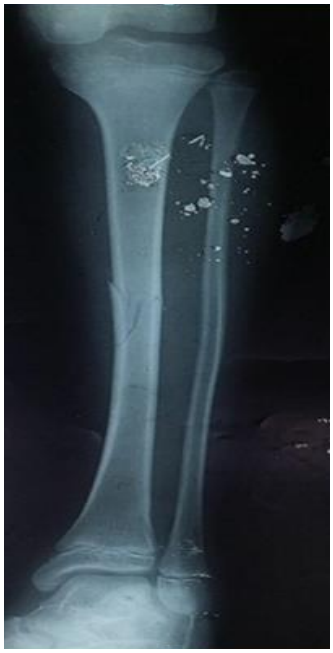
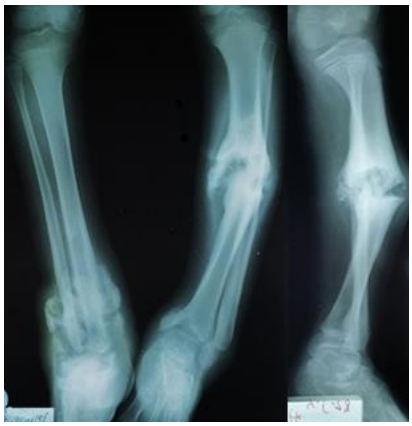
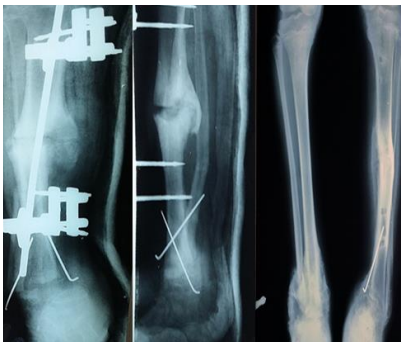


Fig-3: Standard frontal radiograph showing a pathological fracture of the left tibia at the age of 9 years

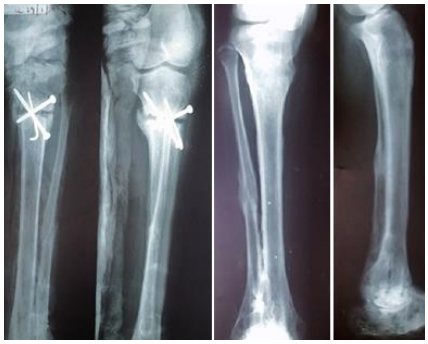


Fig−4: Standard X-rays at the age of 12 showing:

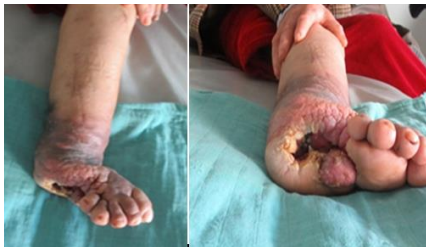
- **Right: the sequelae of osteoarthritis of the right ankle,**
- **Left: Non-union and tibial dislocation with a bimalleolar pathological fracture.**



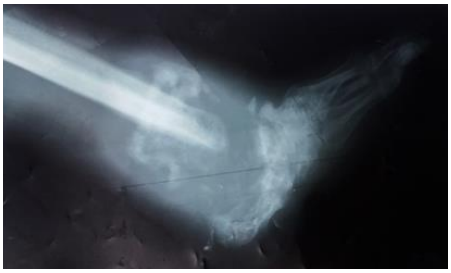
Fig−5 : Standard radiographs showing tibia repositioning and placement of an external fixator and racking of the bimalleolar fracture at age 13 and evolution after 2 years



Fig−6 : Standard radiographs showing right valgus osteotomy and osteosynthesis with vices and pins at age 17, with a good evolution over 6 years



Fig−7 : Trophic disorders of the right ankle with soft tissue loss and bone destruction



Fig−8 : Destruction of bone interesting bones of the hind foot and the lower quarter of the two bones of the leg

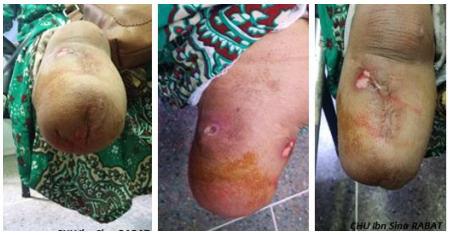


Fig-9: Images showing cutaneous lesions in the stump of the amputation related to the prosthesis

DISCUSSION

Thevenard's disease is a type 1, autosomal dominant, inherited sensory and dysautonomic neuropathy that affects both sexes and may begin at birth or later in childhood, adolescence or adulthood [1,2]. It evolves most often slowly from the second and third decades. The symptoms are not specific since they are described in other Charcot-Marie-Tooth hereditary neuropathies or in neuropathies acquired with severe sensory disorders [1−4].

The first symptoms appear at the distal ends of the lower and upper limbs. They combine skin and osteoarticular lesions of the trophic type as well as neurological signs located at the level of the distal part of the upper and lower limbs [5].

The sensory attack is constant in Thevenard's disease, it initially affects the surface sensitivity thermo-algic. The attack is most often bilateral and symmetrical. Its topography is distal, and it can extend in a systematic way (anesthesia root or truncal) or not systematized "in sock". The search for neuropathy is better with the mono-filament test [6,7].

The osteo-articular reflexes can be preserved at the beginning, diminished or abolished from the start. This attack can be uni or bilateral. The Achilles reflex is the first disturbed and its attack is the most common. The distal reflexes of the upper limbs (pen-radial, cubito-pronator) are affected in case of involvement of the upper limb [1].

Plantar perforating disease begins abruptly in a week or so. It is a painless, sluggish, greyish ulcer, without fibrinous deposit or necrosis, surrounded by a halo of hyperkeratosis. Its origin is mechanical, and it usually starts on a zone of hyper support, physiological or pathological, or on a zone of friction. Secondary secondary infection causes firm edema, local hyperthermia and hyperhidrosis. The location of the upper limb is much rarer than the lower limb [8].

The electroneuromyographic confirms the essentially sensory polyneuropathy, neuromuscular biopsy can rule out other polyneuropathies resulting in an alteration of thermoalgic sensitivity, such as diabetic neuropathy, amyloid, para-amyloid and leprosy. The genetic study confirms the diagnosis by highlighting a mutation in the SPTLC1 gene [7,9].

The evolution of Thevanard's disease is generally progressing to aggravation by successive shoots, separated by periods of remission that are more or less long (sometimes several years). Nerve osteoarthropathy and infectious complications are the main complications of Thévenard's disease [1,4].

The onset of nerve osteoarthropathy is characterized by the appearance on a foot, previously unresponsive due to neuropathy, pain, increased volume and local heat, redness with ligamentous hyperlaxity. General signs are absent (no fever) and there are no biological signs of infection. The diagnostic delay is extremely deleterious because the patient will continue to support his weakened foot, leading to worsening of osteoarticular and ligamentous lesions, with the risk of secondary onset of permanent major deformities. In the absence of immobilization, this acute phase will be complicated by osteolysis with risk of occurrence of fractures and a dislocation of foot architecture.

These deformities can occur one month after the acute phase, in the absence of foot discharge. In the absence of treatment or complications, after weeks of evolution in the destructive mode, a phase of chronic progressive repair appears. This is characterized by a decrease in edema and cutaneous temperature, associated with fracture consolidation. X-rays show the formation of dense bone, particularly in the midfoot, with osteophytes, exostosis and ossification of ligaments and articular cartilages [10]. This consolidation leads to a major reduction in joint mobility, while stabilizing the new abnormal foot architecture. In the absence of a discharge, these restorative phenomena may not be triggered and the foot then remains in a chronic destructive phase. When the deformity is stabilized, the foot remains at high risk of ulceration.

Infectious complications (perforating ulcer secondary rinfected, osteitis and osteoarticular infections) occur in a context of chronic wound often poorly supported, including poor compliance with the discharge of the wound, which is not strictly applied by the patient, because it is experienced as an unjustified constraint in the absence of support pain. This infection aggravates the risk of amputation.

The surgical treatments, in case of osteoarticular damage, will aim, with the help of small local gestures, to limit amputations: Curettage of an osteitis lesion, elimination of bone sequestra. Experience shows that amputations should be considered as late as possible and practiced as economically as possible [11]. These amputations cause indeed displacements of the support axes of the foot on the ground, with increase of the pressures on the opposite limb, provoking or favoring the formation of new lesions, or the recurrence of trophic disorders on the amputation stump [11, 12]

CONCLUSION

Thevenard's disease is a rare familial ulceromutilative acropathy. It is necessary to evoke it before any ulceration of the foot associating with a decrease of the thermoalgesic sensitivity, a family context, or even an attack of the hands. The involvement of the hands is later and therefore more rare. Once the diagnosis is made on a bundle of clinical, electrophysiological or even anatomopathological arguments, the patient must be forewarned of the evolution. No curative treatment is available and the evolution of the disease is inevitable. However, the preventive treatment of cutaneous lesions is essential.

Conflicts of interest

The authors do not declare any conflict of interest.

Contributions of the authors

All authors have read and approved the final version of the manuscript.

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