

Thevenard Disease: Rare Association of 3 Cases in the Same Family (3 Sisters)

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Abstract

Original Research Article

Thevenard's disease is a rare familial ulceromutilative acropathy, responsible for sensory peripheral neuropathy associated with dysautonomous syndrome. The delay in diagnosis and major comorbidities make this condition disabling. We report here a case of 3 sisters, with a history of parental consanguinity, received in Orthopedic and Traumatology Department for the management of a sensitive polyneuropathy, associated with pressure ulcers and ulcers arthropathies of the feet. Functionally, walking was possible without dodging lameness. The electromyogram was in favor of a sensory polyneuropathy of the four limbs. An artisan shoe footfeeding shoe has been proposed, in the face of delayed healing of the wound, associated with local care and sensorimotor rehabilitation. Thévenard's Disease is a rare nosological entity that requires multidisciplinary management.

Keywords: Acropathy, Thevenard, Genetics, Autosomic, Suppuration, Neuropathy.

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1. INTRODUCTION

Thevenard's disease is a rare family ulceromutilating acropathia, responsible for a sensory peripheral neuropathy associated with a dysautonomic syndrome. The diagnostic delay and significant comorbidities make this condition disabling.

Ulceromutilating acropathies (AUM) correspond to affections of the limbs, mainly the feet, characterized by painless ulcerations of the skin associated with bone mutilation [1].

We report the case of three sisters, F.H, S.H and H.H, aged on average 38 and a half years, followed in our training for Thevenard's disease. No patient was diabetic. The clinical presentation common to the three patients corresponded to plantar perforating pain associated with bone mutilation [2].

AUM are complications of many sensory neuropathies. Treatment is also based on etiological treatment if possible and on symptomatic treatment [3].



Figure 1: Swelling and abduction of the interphalangeal joints of the toes

2. RESEARCH METHODS

Our work focuses on the study of the clinical history and the evolution of 3 sisters followed at the Traumatology-Orthopedics Department of the CHU Ibn Sina in Rabat for Thevenard's disease since 2005 [4]. These patients were initially followed at the Children's Hospital of Rabat before their files were transferred to our service. These three patients H.H, F.H and S.H had numerous ulcerations in their lower limbs with an abundant issue of pus, which motivated their consultations and then their hospitalizations on numerous occasions [4]. As their symptoms worsened, two of them were able to benefit from a limb amputation (mid-leg and disarticulation of the knee) while the third still benefits from medical follow-up with antibiotic treatment adapted to cytobacteriological examination and numerous trimmings with excision of infected and suppurated tissues [5]. F.H was recently able to benefit from an arthrodesis of the ankle due to the significant osteitis, which developed there at the origin of numerous episodes of dislocations [5].



Figure 2: Ulcerations localized in the foot



Figure 3: Ulcerations localized in the knee and the ankle



Figure 4: Ulcerations localized in the knee

3. RESULTS AND DISCUSSION

Thevenard's disease corresponds to type 1 of hereditary sensory and autonomic neuropathies, transmitted in an autosomal dominant mode. It evolves slowly from the second and third decades and is manifested by thermoalgesic sensitivity disorders, leading to painless ulcerations at the pressure points [6].

The evolution is usually towards the extension of ulcerations and the installation of perforating plantar pain, a dislocation of the tarsus with a "cubic foot" aspect [10], iterative superinfections in the form of analgesic paronychia and osteoarthritis of the extremities leading to mutilations of the phalanges and variable alterations of the skeleton of the foot [6]. Foot and leg amputations reflect the failure of systemic antibiotics often due to the emergence of bacterial resistance [10].

The diagnosis is made in practice on clinical, electrophysiological and family data. Neuromuscular biopsy is only useful for differential diagnosis, ruling out other polyneuropathies responsible for impaired thermoalgesic sensitivity, such as diabetic, amyloid, para-amyloid and leprosy neuropathies [7].

Diagnostic confirmation requires the detection of a mutation in the SPTLC1 gene. Preventive treatment of skin lesions is the mainstay of the management of these patients because no curative treatment is available [8].



Figure 5: Deformation of the fingers and loss of nail relief

5. CONCLUSION

Thevenard's Disease is a rare familial ulceromutilating acropathy. It is a pathology of the peripheral nervous system which begins at the level of the feet where it most often remains the only manifestation, to extend exceptionally and then later at the level of the hands. Management is difficult and long and requires long-term follow-up.

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