SKIN MANIFESTATIONS OF INTERNAL DISEASE

TRANSTHYRETIN-RELATED FAMILIAL AMYLOID POLYNEUROPATHY: A CASE REPORT WITH SKIN MANIFESTATIONS

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Background: Transthyretin-related familial amyloid polyneuropathy (TTR-FAP) is a rare autosomal dominant disease caused by mutations on the thyroxine and retinol transport protein transthyretin, leading to tissular amyloid deposits. The symptoms usually start at the 4th decade of life with dysautonomia, weight loss and progressive reduction of peripheral tactile, thermal and pain sensitivity. Later neurological findings are hyporeflexia and weakness. The most common skin manifestations are xerosis, onychomycosis, repeated burns and traumatic lesions, leading to poor healing ulcers and skin infections. Renal and cardiac failures contribute for poor outcomes. The diagnosis is based on peripheral nerve biopsy and genotyping. If left untreated, the mean survival rate is 10 years.

Observation: 37-year-old female patient with 1st and 2nd degrees familial history of TTR-FAP presenting with dysautonomia and unexplainable weight loss prior to the polyneuropathy with neuropathic pain, paresthesia, reduction of proprioception, thermal and pain sensitivity in extremities. Peripheral nerve biopsy and genotyping were performed, diagnosing TTR-FAP. The patient remained untreated for 6 years after the onset of the first symptoms, developing heart failure, chronic kidney disease and more severe polyneuropathy. The skin assessment showed multiple burns and traumas, lower limbs lymphedema with hyperkeratosic, verrucous and lichenified plaques on the pre-tibial region, plantar keratosis and perforating ulcer on the right calcaneus. On the following three years the patient presented recurring pyodermas, sepsis and chronic osteomyelitis, resulting in below-knee amputation of the right leg and sacral pressure ulcers due to the loss of mobility.

Key message: TTR-FAP is a rare multisystemic disease with progressive and severe course. Even though the amyloid deposit and tissular damage are irreversible, early suspicion and diagnosis may reduce morbidity and improve quality of life and survival rates.