E-Poster

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# Thevenard's Disease: A Case Report

**Topic:** Health Conditions (Nervous System, Musculoskeletal System, Cardiopulmonary System)

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## BACKGROUND

Thevenard's disease is a rare hereditary sensory and autonomic neuropathy (HSAN type 1) characterized by progressive sensory loss predominantly affecting the distal extremities. It typically manifests in adolescence or adulthood, leading to severe complications such as painful fractures, neuropathic ulcers, osteomyelitis, and Charcot arthropathy, which can result in amputations. Diagnosis is based on clinical presentation and family history, with genetic confirmation via specific gene sequencing.

## CASE DESCRIPTION

We report a case of a 23-year-old male diagnosed with Thevenard's disease, presenting with multiple orthopaedic complications. The patient had a strong family history of the disease, and symptoms began at the age of one with a spontaneous, painless right wrist fracture, identified by his mother. The diagnosis was established based on clinical findings without genetic testing.

Over the years, the patient experienced recurrent dislocations, neuropathic ulcers with secondary infections, and progressive joint degeneration, requiring multiple hospitalizations and surgical interventions, including amputations of the left fifth toe and distal phalanges of the second fingers on both hands.

On the most recent clinical examination, the patient exhibited painless shoulder movement limitation (Flexion/Abduction: 100°/0°/90°), glenoid cavity emptiness, and an epaulet sign along with hypoesthesia of the left upper limb. Additional findings included bilateral knee deformities, malformation of the left first toe, and moderate cognitive impairment. Radiographic imaging revealed left shoulder dislocation, bone lysis, and bilateral knee joint degeneration consistent with Charcot arthropathy.

## DISCUSSION

Thevenard's disease is a dominantly inherited neuropathy that significantly impacts joint stability and function. The absence of pain perception predisposes patients to repeated trauma, joint destruction, and severe deformities. In this case, progressive left shoulder dislocation and bilateral knee degeneration indicate the progressive nature of the disease. Furthermore, cognitive impairment may pose challenges to self-care and adherence to rehabilitation protocols, highlighting the need for tailored management strategies.

## CONCLUSION

This case underscores the severe orthopaedic and neurological consequences of Thevenard's disease at a young age. A multidisciplinary approach is essential for stabilizing function, preventing further damage, and improving mobility. Comprehensive management, including rehabilitation, orthotic support, and patient education, is crucial to improve independence and quality of life.

**Statement of Disclosures**

None

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