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Familial Primary Erythromelalgia: Diagnostic and Therapeutic Challenges

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Introduction & Objectives: Erythromelalgia is a rare acrosyndrome characterized by a triad of redness, localized heat, and burning pain, typically affecting the lower limbs. It exists in primary (familial or sporadic) and secondary forms. Familial primary erythromelalgia, a rarer form with autosomal dominant inheritance, presents significant therapeutic challenges.

Materials & Methods: An 18-year-old male, born from a first-degree consanguineous marriage, presented with paroxysmal episodes of pain, heat, and erythema in the lower limbs since the age of 12. These episodes (10–12/day) were triggered by heat and relieved by cold immersion. Multiple symptomatic treatments yielded no relief, severely impacting quality of life and leading to psychiatric complications. Three family members had similar symptoms; two had died from related complications.

Clinical examination showed bilateral violaceous erythema of the feet extending to the lower legs, plantar keratoderma with fissures, and diffuse intertrigo. Routine blood tests revealed thrombocytosis, but thrombocythemia was excluded. Immunologic tests were negative. Based on clinical and familial data, a diagnosis of familial primary erythromelalgia was made. Treatment with gabapentin, SSRIs, and aspirin led to slight improvement. Adding a beta-blocker significantly reduced episodes and erythema. Psychiatric symptoms remained difficult to manage.

Results: First described in 1878, erythromelalgia manifests as episodic burning pain and erythema, worsened by heat and relieved by cold. The primary form may be familial, linked to mutations in SCN9A, encoding the NaV1.7 sodium channel involved in nociceptive signaling and vasodilation. Diagnosis is clinical, supported by exclusion of secondary causes such as myeloproliferative disorders or autoimmune conditions.

Management is complex. Secondary forms improve with treatment of the underlying disease; aspirin is often effective in such cases. For primary forms, no standardized treatment exists. Reported options include beta-blockers, tricyclic antidepressants, gabapentin, calcium channel blockers, opioids, lidocaine, mexiletine, and novel NaV1.7 blockers (e.g., funapide, vixotrigine). Surgical approaches (e.g., sympathectomy) and neuromodulation techniques are considered in refractory cases. Psychological support remains essential.

Conclusion: Familial primary erythromelalgia is a rare and debilitating condition with limited effective treatments. A multidisciplinary and individualized approach is vital. Psychological support is crucial to improve quality of life and treatment adherence.