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Primary erythromelalgia: a clinical diagnosis

Eritromelalgia primária: um diagnóstico clínico

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Keypoints

What is known
- Erythromelalgia is a rare chronic disorder responsible for neuropathic pain in pediatric age.
- Its etiology can be genetic or secondary.
- There is no cure or universally effective therapies.

What is added
- Awareness about erythromelalgia as a possible cause of neuropathic pain in children is important for its prompt diagnosis.
- Due to the impact on quality of life, its early identification is essential.

Erythromelalgia is a rare chronic disorder characterized by a triad of intermittently red, hot and painful extremities. The syndrome usually affects the lower extremities with symmetrical cutaneous findings, but may also involve the upper extremities and, rarely, the face and present with unilateral involvement only. These episodes can last from minutes to days and are characteristically aggravated by warming and relieved by cooling measures. The condition can be classified as primary or secondary, depending on the detection of an underlying cause. Multiple disorders can be linked to erythromelalgia, but special attention should be given to myeloproliferative illnesses, responsible for about 10 percent of secondary cases. Up to the present time, there is no cure or universally effective therapies[1,2].

We report a case of a previously healthy 19-month-old male child referred to a pediatric appointment for recurrent episodes of erythema, heat, edema and pain in the lower limbs, which endured for one to two hours, triggered by defecation or local trauma and relieved by immersion of the extremities in cold water. These episodes had been occurring in a monthly matter for seven months. His mother brought photographs that evidenced the complaints (Figs. 1 and 2). He had no other associated symptoms or signs, his physical examination was normal and laboratorial evaluation excluded secondary causes. There was family history of similar episodes in the maternal branch, without an established diagnosis. In view of the suspicion of primary familial erythromelalgia, a directed genetic study was performed, which showed a mutation in the *SCN9A* gene, confirming the diagnosis. Regarding the association of the child's symptoms to defecation effort, a laxative therapeutic was started and topical lidocaine was prescribed for acute relief. To date, more sparse intermittent episodes keep occurring, without significant daily functioning compromise.

Primary erythromelalgia is associated with a mutation in the *SCN9A* gene, which encodes an aberrant ion channel responsible for the hyperexcitability of peripheral nociceptors. This mutation can be sporadic or familial, being transmitted as an autosomal dominant trait.

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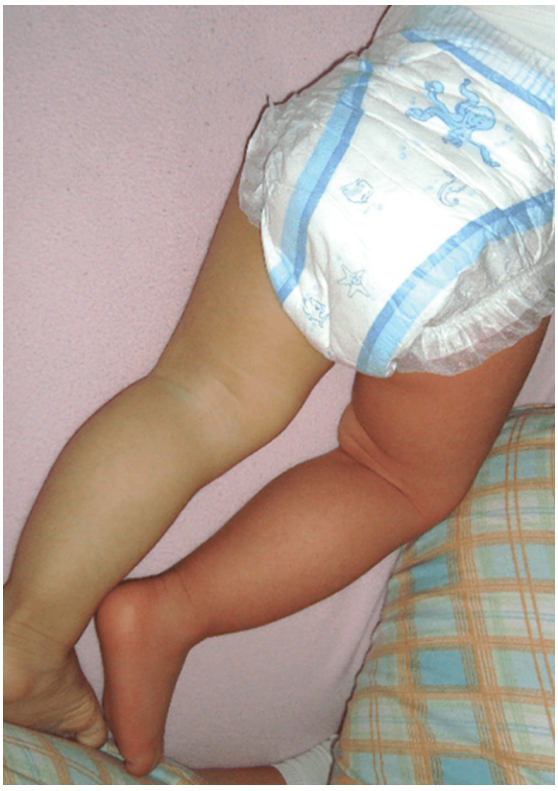

Figure 1. Child with erythematous right lower limb.

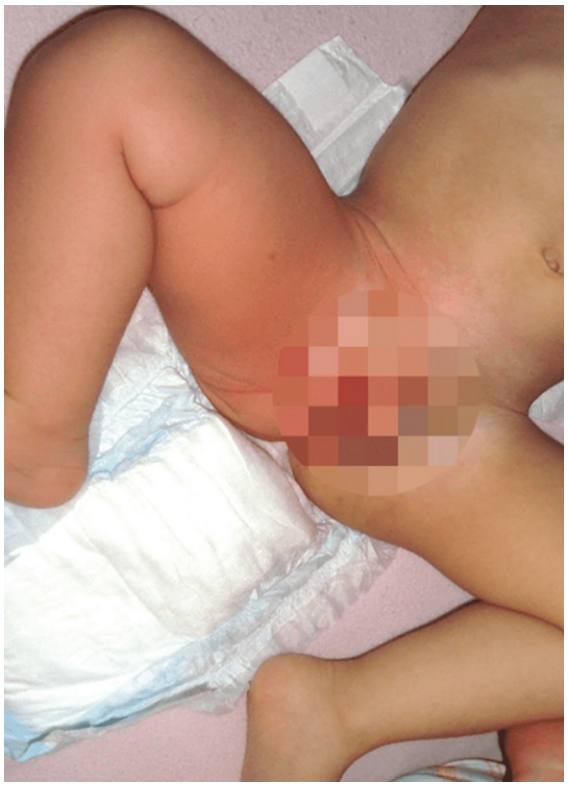

Figure 2. Child with erythematous and swollen right lower limb.

Diagnosis is clinical, genetically confirmed[3]. Treatment is based on behavioral measures, such as avoiding precipitating factors and body cooling measures, and pain-modulation pharmacological therapies. Ice-cold water immersion can attain instant relief, but it also can lead to severe complications, such as skin ulcers and necrosis. Pharmacological intervention with topical anesthetics or oral agents for neuropathic pain may also be used, but pediatric data is still limited. Due to the impact on quality of life, its early identification is essential[1,2,4].

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Conflicts of interest

None.

Ethical disclosures

Protection of human and animal subjects. The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data.

Right to privacy and informed consent. The authors have obtained the written informed consent of the patients or subjects mentioned in the article. The corresponding author is in possession of this document.

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