Reviewer's report

Title: Erythromelalgia, a rare condition: a case report and review of the literature.

Version: 1 Date: 6 January 2009

Reviewer: Joost P Drenth

Which of the following following best describes what type of case report this is?: None

Has the case been reported coherently?: No

Is the case report authentic?: Yes

Is the case report ethical?: Yes

Is there any missing information that you think must be added before publication?: Yes

Is this case worth reporting?: Yes

Is the case report persuasive?: No

Does the case report have explanatory value?: No

Does the case report have diagnostic value?: No

Will the case report make a difference to clinical practice?: No

Is the anonymity of the patient protected?: Yes

Comments to authors:

Erythromelalgia, a rare condition: a case report and review of the literature.

Dr. Gaur et al present a 33-yr old female who presented with complaints of episodic burning pain and flushing restricted to face, ears, upper chest and rarely, the upper extremities. Her symptoms were triggered by lying down or warm exposure and relieved by cooling measures. Thermography demonstrated elevated temperature in affected body parts. Family history is negative, and an extensive search excluded an apparent underlying disorder.

The case history contains typical and atypical aspects of erythermalgia. While burning pain and flushing are typical for erythermalgia, as is precipitation by warmth, lying down most often leads to amelioration of symptoms. Further, patients most commonly present with warm, symmetric painful feet, which is not
present in this case. True, in advanced cases it is possible to note flushing in
face and ears, but that is a rare exception.

This is an interesting case observation but apart from the observation perse there
is not much to be learned. Browsing through the paper I asked myself 2
questions (1) Is the material new and will it have any impact on clinical practice
or add substantially to current knowledge? and (2) Is the message appropriate
for the practicing clinician? At this point I do not see what this paper/case report
adds. The authors cite premature ovarian failure, but how should this fit with the
pathogenesis of erythermalgia? If there is a connection how to follow that up?
Any clinical implications?

I am still puzzled by the low core body temperature of 94 degrees Fahrenheit. Is
this secondary to the low ambient living temperature, or are there alternative
explanations. Was poikilothermia excluded?

The discussion is difficult to follow and the authors compound erythromelalgia,
with primary and secondary erythermalgia and hypertension associated
erthermalgia in childhood. I agree, the literature on this syndrome is difficult to
understand, but as it stands, the discussion is rather free floating citing some but
not other important material.

To be clear, primary or idiopathic erythermalgia (OMIM 133020) is an autosomal
dominant, inherited disorder. Clinically, PE is characterized by attacks or
episodes of symmetrical burning pain of the feet, lower legs, and sometimes
hands, elevated skin temperature of affected areas, and reddened extremities.
PE is sometimes termed erythromelalgia, although some authorities reserve the
latter term for a condition that is caused by arteriolar inflammation as a result of
platelet-rich thrombi in the end-arterial microvasculature, in which the platelet
count is invariably elevated (> 400 × 10^9 cells/l) and a short course of aspirin
brings swift relief. Platelet counts in PE are invariably normal, and aspirin is
ineffective, as in this patient. This patient probably suffers from a non-inherited
form of erythermalgia, although the clinical presentation is somewhat atypical.

The authors indicate that genetic testing of SCN9A is feasible. Did they perform
that for this patient? A novel mutation would greatly enhance the value of this
manuscript.

The authors cite the effect of mexiletine, and indeed several reports suggest a
positive effect. Was it tried in their patient? (Pediatrics. 2005 Apr;115(4):e504-7)

Minor typos
be excluded.

Quality of written English: Acceptable

Declaration of competing interests:

I have written several papers on this topic