ID Design Press, Skopje, Republic of Macedonia
Open Access Macedonian Journal of Medical Sciences. 2018 Jan 25; 6(1):74-75.
Special Issue: Global Dermatology-2
https://doi.org/10.3889/oamjms.2018.024
eISSN: 1857-9655
Case Report

Van Lohuizen Syndrome – A Case Report with a Diagnostic Delay of Four Years

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Citation: Wollina U, França K, Lotti T, Tchernev G. Van Lohuizen Syndrome – A Case Report with a Diagnostic Delay of Four Years. Open Access Maced J Med Sci. 2018 Jan 25; 6(1):74-75. https://doi.org/10.3889/oamjms.2018.024

Keywords: Cutis marmorata telangiectatica congenita; Van Lohuizen syndrome; vascular-cutaneous disorders

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Received: 20-Aug-2017; **Revised:** 16-Nov-2017; **Accepted:** 29-Oct-2017; **Online first:** 02-Jan-2018

Funding: This research did not receive any financial support

Competing Interests: The authors have declared that no competing interests exist

Abstract

BACKGROUND: Cutis marmorata telangiectatic congenital or Van Lohuizen syndrome is a rare vascular disorder that may be associated with other congenital malformations. Around 300 cases have been reported so far.

CASE REPORT: We present a 4-year-old girl with Van Lohuizen syndrome of the leg, but without any other malformations.

CONCLUSION: Neonatal lupus erythematosus may resemble congenital vasculopathy, but histopathology and immune-serology are characteristic.

Introduction

Van Lohuizen – a female paediatrician from the Netherlands - first described a congenital vascular-cutaneous disorder in 1922 - cutis marmorata telangiectatica congenita [1]. Its aetiology is unknown, and the occurrence is spontaneous. The clinical presentation is a combination of cutis marmorata with telangiectasia. Skin atrophy or ulcerations are sometimes present. The cutaneous features show a tendency of improvement over time [2]. The most common associated cutaneous malformations include port-wine stains and hemangiomas [3].

Other associated anomalies may affect any organ of the body, such as the eye, skeleton, brain, kidneys, etc. [4].

We report the case of a female patient with van Lohuizen syndrome not associated with other anomalies.

Case report

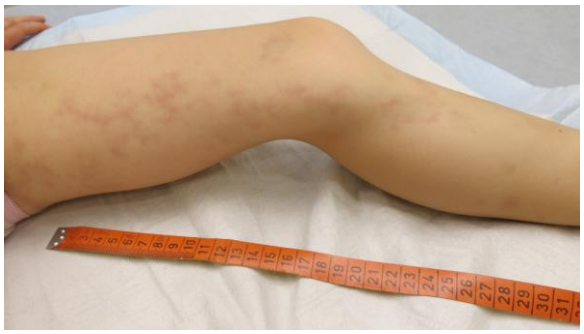
A 4-year-old girl was referred with her parents to our department for consultation. She had segmental skin disorders in her left leg that showed an enlargement during the last three months.

The girl was born from a non-consanguineous marriage. It is the first child of a healthy mother. Her delivery was uncomplicated on time. Skin changes were observed at birth demonstrating hyperpigmented macules of a reticular pattern and telangiectasia. The changes were unresponsive to temperature.

Her further development was unremarkable.

There was no evidence of skeletal, motoric, neuronal or ophthalmologic malformations. She was monitored on a regular basis by her paediatrician and neither laboratory, nor functional abnormalities were noted. Hyperpigmentation partially faded away by time. There was neither atrophy nor ulceration at any time.

a)



b)

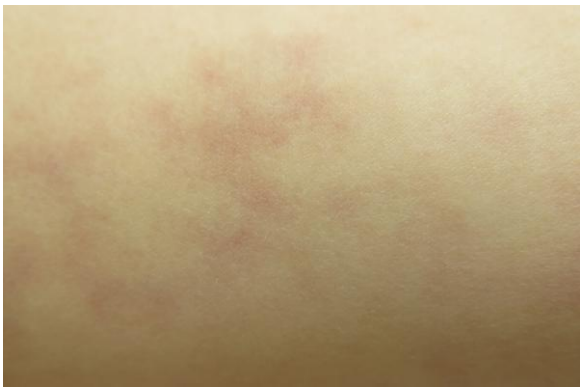


Figure 1: Van Lohuizen syndrome on the leg in a 5-year-old girl. (a) Overview, (b) detail of the hyperpigmented reticular patches

We observed a reticular slightly hyperpigmented pattern on the inner side of the thigh and lower leg. This feature was associated with telangiectasias. On palpation, the whole lesion remained painless. The parents were informed about the benign nature of the vascular-cutaneous disorder. Based on medical history and clinical presentation the diagnosis of Van Lohuizen syndrome was confirmed. The treatment is impossible and in the present case also unnecessary.

Discussion

Van Lohuizen syndrome (OMIM 219250) is a rare congenital disorder with no clear gender prevalence caused by genetic mosaicism. No specific mutations have been discovered until the present moment [5]. Around 300 cases have been reported so far [6]. The diagnostic criteria include three major criteria such as congenital reticular (marmorated) erythema, the absence of venectasia, and unresponsiveness to local warming. Furthermore, two or more minor criteria should be present such as fading of erythema within two years, telangiectasia within the affected area, port-wine stain, ulceration within the affected area, and atrophy within the affected area [6]. Our patient fulfilled the three major and two of the minor criteria.

There are many differential diagnoses. The closest disease to Van Lohouizen syndrome is congenital livedo reticularis, where ulceration and phlebectasia do not occur. It is part of the congenital livedo reticularis-megalencephaly syndrome (OMIM 602501) being caused by mosaic *PIK3CA* gene mutations [7]. Other differential diagnoses include Klippel-Trenaunay syndrome and Sturge-Weber syndrome. The latter is caused by mosaic *GNAQ* gene mutation *c.548G>A* [5]. Neonatal lupus erythematosus may resemble congenital vasculopathy, but histopathology and immune-serology are characteristic [8].

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