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Case Report

Dorsal Localization of "Cutis Gyrata" in a Patient with Noonan Syndrome

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Abstract

Background: Among the genetic disorders associated with cutis gyrata the most frequent ones are Noonan and Turner syndromes. Lymphedema has been proposed as a pathogenically cause of cutis gyrata in both syndromes.

Case Report: A girl, with a clinical phenotype of Noonan syndrome, who has also Parry Romberg syndrome. She showed a dorsal plaque with clinical and histological features of cutis gyrata but in a non-verticis localization.

Conclusion: The same physiopathological mechanism of lymphatic anomalies can be assumed as a possible cause of the cutis gyrata, in this case, in a dorsal localization.

Keywords: Cutis Gyrata; Cutis Verticis Gyrata; Lymphedema; Noonan Syndrome; Parry-Romberg Syndrome; Turner Syndrome.

Introduction

Cutis Verticis Gyrata (CVG), first reported by Alibert in 1937, is a descriptive name meaning the clinical features of thick skin furrows and thick folds localized usually on the scalp. It has been classified in primary and secondary forms. The primary ones are further divided into essential and nonessential CVG. The latter has been associated with mental deficiency, neurologic and ophthalmologic abnormalities. It has been reported a strong association between nonessential primary forms and chromosomal and genetic abnormalities, such as Turner and Noonan syndromes [1-3]. In Turner and Noonan's syndromes congenital lymphedema has been postulated as its possible etiopathogenetical cause [4].

The aim of this report is to describe a clinical case with an unusual localization of CVG in a girl affected with Noonan's and Parry-Romberg syndrome.

Case Report

This female patient was born from non-consanguineous parents, by C-section of a full-term pregnancy, complicated by maternal hypertension. Her birth weight was 3500 g. At birth, she was noted to have edema of the hands and feet and redundant nuchal folds. Chromosome studies revealed a normal 46 XX karyotype.

She was derived to our dermatology department for evaluation of progressive facial hemiatrophy.

On dermatological exam at 10 years of age, she was a well-developed girl with normal weight, height and mental developmental for her age. She has left facial atrophy; a linear atrophic lesion was more evident on her frontal skin. She also presented an inexpressive, inverted triangle face, small, low setting, posteriorly rotated ears with thick lobules, and micrognatia. A curly hair with low hairline on the nuchal region and webbed short neck was also appreciated (Figure 1). Other clinical features were wide spaced nipples, and short 4th and 5ft metacarpal bones.

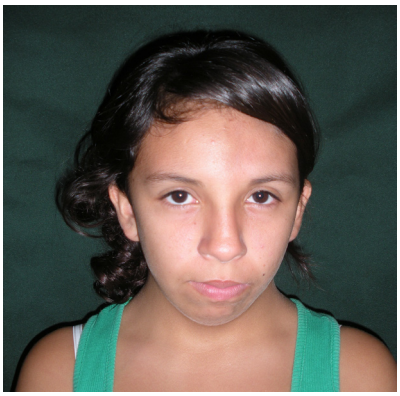

Figure 1: Note left facial atrophy, linear atrophic lesion Inexpressive, inverted triangle face, small, low setting ears, micrognatia, and curly hair.

On her right dorsal region, a 6 x 8 cm. skin colored, asymptomatic plaque with thick folds and deep furrows was noted (Figure 2). The girl also has 3 café-au-lait spots and one hypopigmented macule. Histopathologic evaluation of a 3-mm punch biopsy specimen of the dorsal plaque showed a normal epidermis with disorganization of the dermal collagen.

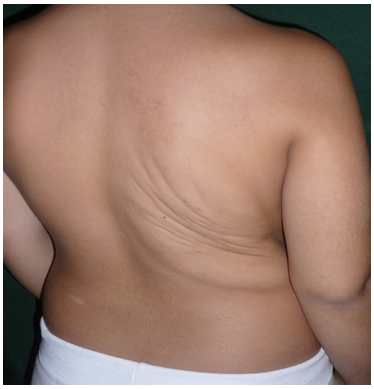

Figure 2: Skin colored plaque with thick folds and deep furrows. Note hypopigmented macule.

The cardiovascular, neurologic, ophthalmologic and neurologic evaluations gave normal results. Abdominal ultrasound showed bilateral dilatation of the renal pelvis; both ovaries were present and showed normal structure for her age. Echocardiogram was normal. The brain TC showed normal results.

Based on the clinical history and the physical examination we consider the patient diagnosis was consistent with cutis gyrata in a dorsal localization in a patient with Noonan syndrome and Parry Romberg syndrome.

Discussion

The clinical descriptive term "cutis verticis gyrata" is used to refer to the clinical features of thick folds and deep furrows usually localized on the scalp [1]. Polan and Butterworth classified CVG into primary and secondary forms [2]. Between the primary forms, he recognizes essential and nonessential forms. The former is those were no detectable cause can be found, and the latter when an association with other abnormalities such as mental deficiency neurologic or ophthalmologic abnormalities is identified. In such cases, a strong association with chromosomal and genetic disorders, such as Turner and Noonan syndrome has been reported. Most the primary cases of CVG occur before the age of 30. In the secondary forms, those cases in which an underlying cause such as nevi, inflammatory dermatosis, acanthosis nigricans, hamartomas, dermatofibromas, between others, has been included [1,2].

Turner and Noonan syndromes share many clinical features, i.e. low posterior hairline, webbed neck, low set posteriorly rotated ears, wide-spaced nipples, and puffy hands, all probably are caused by the resolution of intrauterine lymphedema. Our case has many clinical features characteristic of both syndromes, but Turner syndrome was ruled out by 46, XX on cytogenetic analysis. Both Turner and Noonan syndromes has been reported as primary cause of cutis gyrata. In most of the cases it is localized on the head and neck area, as the name "cutis verticis gyrata" implies [1]. The precise mechanism by which CVG occurs remains to be determined [1]. Larralde et al. [3]. proposed lymphedema as a possible cause of CVG in Turner syndrome, where the resolution of lymphedema leaves redundant skin. In uterus compression may fix lymphedematous skin into the folds that manifest clinically as cutis gyrata [1,4]. Histopathologic findings in CVG range from essentially normal skin structure to thickened connective tissue [3]. Both histological findings could be considered as resolving stages of lymphedema. Lymphedema is relevant to the development of not only soft tissue and visceral anomalies but also skeletal anomalies in Noonan syndrome. The association between NS and lymphedema has been well established, although its significance is not yet clear. It may affect localized areas of the body or be more generalized; several forms of presentation have been reported: generalized lymphedema, peripheral lymphedema, pulmonary lymphangiectasia, intestinal lymphangiectasia, cystic hygroma, and hydrops fetalis [5,6]. Chevernak et al [7]. consider that a cystic hygroma occurs due to failure of the lymphatic vessels to mature during the intrauterine period of life. The pterygium coli may be explained by the regression of a cystic hygroma following correction of the lymphatic obstruction, or the formation of collateral lymphatic channels. The edema may also affect the migration of tissues during development of the embryo and explain the anomalous location of some structures (e.g., cryptorchidism, separated nipples, hypertelorism) and, may also explain the development of pulmonary stenosis [5-10].

Although the CVG has been reported in Noonan syndrome,

and patients with this genetic disorder presents lymphedema at birth, the relationship between congenital lymphedema, CVG and Noonan syndrome has been only considered in the case published by Fox et al. [1]. Taken together these data suggest that in our case the dorsal plaque, that fulfils the clinical and histological features of cutis gyrata, present on the dorsal thoracic skin, could also be considered a sign of the lymphatic alteration within this syndrome.

To conclude, even though in our patient the skin lesion, described as a cutis gyrata was located outside the head and neck area we consider that abnormalities in the lymphatic vessels should be consider in the development of this lesion in our patient. Meanwhile, as other aetiologies of CVG in Turner and Noonan have not been proposed, we believe this is the most feasible cause of cutis gyrata in these patients. We consider the genetic evaluation for both syndromes are necessary in case of CVG and that is also important to search for an extra-cranial localization of this type of lesion in future cases.

We consider that, in this patient, the association of Parry-Romberg syndrome was a fortuitous coincidence.

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