Plexiform neurofibroma of the penis as an infrequent manifestation debut in a pediatric patient

M. PEÑA [1], A. MARTÍNEZ [1], T. DOSSI [1], M. KOLBACH [1], C. GARCÍA [1], C. MELLADO [1].
1. Pontificia Universidad Católica de Chile, Santiago, Chile.

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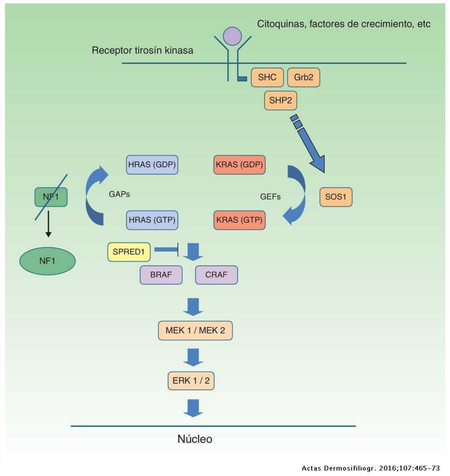
INTRODUCTION

Neurofibromatosis type 1 (NF1) caused by loss of function mutation in the NF1 gene; leads to the hyperactivation of RAS and its downstream mediators and contributes to tumour formation. The main manifestations of NF1 are café au lait macule, axillary and/or inguinal freckling. Neurofibroma plexiform is specific for NF1 and identified on the face and trunk. Urogenital presentation is infrequent in the penis. Identification of this lesion is essential because it carries a risk of malignancy in about 5%–10% of patients.

NF1

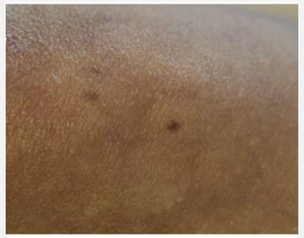


Actas Dermosifiliogr. 2016;107:465–73

CASE REPORT

We present a five-year-old boy who presented to us with progressive swelling of the penis since he was a newborn. He had no swelling in other parts of the body, without urinary dysfunction and no history of neurological symptoms. Genitourinary examination revealed a penis length of 9 cm (greater than 2 SD for age) and a girth of 10 cm at the base level. Small lentigines at the tip and base of the penis. Unpigmented scrotum and testicular volume less than 3 mL. After a dermatological evaluation, small café-au-lait spots observed on the right hip and posterior side of the left leg. The sister presented a hypopigmented spot on the hip skin and a café-au-lait spot on the right leg, and the father with reddish spots in the popliteal with a lymphatic appearance. A genital ultrasound showed lesions compatible with plexiform neurofibroma, and his hormonal profile, such as androstenedione, total testosterone, DHEA-S, 17-OHP and LH were within normal ranges. INVITAE study for RASopathies Comprehensive Panel and EPHB4 gene reported negative.

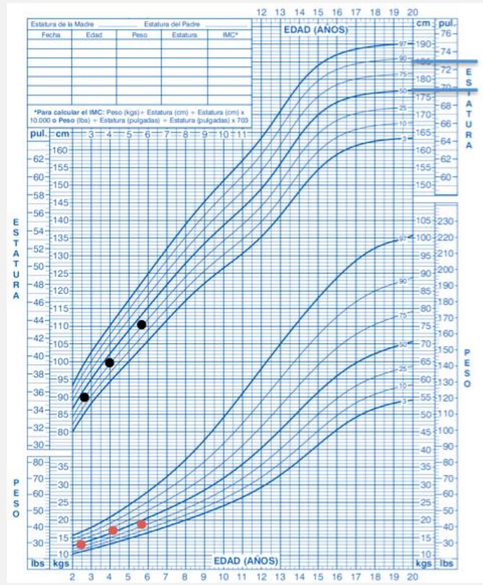
Physical exam



Café-au-laits spots

Base of the penis

Growth Chart



CONCLUSIONS

The genetic testing of NF1 would help to clarify the diagnostic process; however, this can be difficult because phenotypes in NF1 are caused by mosaicism; and are classified as segmental, generalized, or gonadal. The segmental type causes regions with pigmentary alterations, tumour growths, or both, limited to one or more regions of the body, precisely the diagnosis that we consider corresponds to our patient, even though genetic study (INVITAE) is reported as negative. It is essential to identify the diagnosis due to the increased risk of malignancy.

REFERENCES

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Division of Genetics, Pontificia Univerdad Católlica de Chile.
Division of Radiology, Pontificia Universidad Católica de Chile
Division of Dermatology, Pontificia Universidad Católica de Chile.

CONTACT INFORMATION

Fer.pm83@gmail.com

mfpena3@uc.cl