Case 14869

Eurorad

Exophytic tumour in the nipple-areola complex of the breast in a female patient with neurofibromatosis

Published on 31.07.2017

DOI: 10.1594/EURORAD/CASE.14869
ISSN: 1563-4086
Section: Breast imaging
Area of Interest: Breast
Procedure: Diagnostic procedure
Imaging Technique: Mammography
Imaging Technique: Ultrasound
Imaging Technique: Ultrasound-Colour Doppler
Imaging Technique: CT
Special Focus: Neoplasia Case Type: Clinical Cases
Authors: Yazmín Aseret Ramírez Galván MD, Miguel Antonio Sada Treviño MD, Néstor Alejandro Rangel Ovalle MD
Patient: 21 years, female

Clinical History:

21-year-old female patient with exacerbated chronic abdominal pain in right hypochondrium and lower back. She presented multiple cutaneous “cafe au lait” spots (>5) and axillary freckling (Fig. 1). In abdomen a stony mass and a Giordano sign were present.
She referred that two brothers died due to complications of schwannomas.
Imaging Findings:

Abdominal IV contrast CT showed an oval retroperitoneal mass, well delimited, of 16 cm, predominantly hypodense with heterogeneous enhancement and hypodense necrotic areas, that displaced the ascending colon and right kidney without infiltrating them (Fig. 2). Given that it may be difficult to differentiate MPNST from plexiform neurofibromas based on image, and as part of treatment, surgical resection was performed. The histopathological diagnosis of a low-grade malignant peripheral nerve sheath tumour (MPNST) was made.
Incidentally an exophytic tumour in the nipple-areola complex of the left breast was seen on CT, that was dark brown at inspection; at ultrasound it appeared as an oval dermal lesion with lobed borders, hypoechoic and no flow (Fig. 3 and Fig. 4). Complementary mammography projection of left breast showed a multilobulated and isodense mass in the topography of the nipple, the mammary parenchyma had no lesions. (Fig. 5)
Discussion:

Background
Neurofibromatosis is a phakomatosis (neurocutaneous syndrome) that involves tissues originated from embryonic ectoderm (central nervous system, skin and eyes) [1, 2]; in neurofibromatosis type 1 (NF1, von Recklinghausen disease), the alteration lies in the long arm of chromosome 17 where neurofibromin (a tumour-suppressing protein) is coded, with its function being diminished or absent [1, 5].

Clinical perspective
NF1 has an inherited autosomal dominant pattern, and its diagnosis is based on clinical criteria established by the

National Institutes of Health Consensus Development Conference, with at least two of them required to make the diagnosis [2, 5, 7]; features are summarised in Fig. 6. The patient met several criteria: “cafe au lait” spots, axillary freckling, retroperitoneal MPNST, breast lesions and family history.

NF1 is the most frequent phakomatosis, affecting multiple systems with masses in subcutaneous tissue (including breast), that can become expansive and infiltrative (plexiform type) [2, 5]. Skin lesions may grow to become nodular, hyperpigmented and pedunculated [1, 7]. The risk of malignant transformation varies, being low for neurofibroma and greater for the plexiform type [1, 3]. Because of its high penetrance, all relatives of patients with NF1 should undergo screening tests [7].

Once diagnosis is considered, referral should be made to clinicians experienced in NF1, the basis of management being monitoring of manifestations and patient education [6, 7].

Imaging perspective
Breast lesions mammographically appear as well-defined peri-areolar masses, often multiple, borders may be surrounded by air density reflecting their superficial nature; the skin lesions can mimic and partially obscure parenchymal lesions [8, 9]. At ultrasound they appear as well-defined hypoechoic mass with posterior acoustic enhancement, located in the subcutaneous tissue similar to fibroadenoma [4, 9].

Neurofibromas of breast are rare manifestations of NF1 and occur on the nipple-areolar complex. [9]. The literature suggests that women with NF1 are at a higher risk of developing breast cancer compared to general population; however, there are not specific considerations for screening in these patients [10]. Surgical removal of cutaneous neurofibromas is indicated if they cause discomfort or are visible and stigmatising [11].

Outcome
There is an 8–13% lifetime risk of developing MPNST in NF1, principally in individuals aged 20–35 years, and are hard to detect, metastasise widely and often have a poor prognosis [7]. Purpose of treatment is removal of lesions with tumour free margins; adjuvant radiotherapy has a role in treating MPNST larger than 5 cm, high grade lesions and incompletely excised tumours [7].

Take home message

Close collaboration between clinicians and radiologist will facilitate diagnosis and management of NF1 and its complications.
Differential Diagnosis List: NF1 with abdominal low-grade MPNST and breast lesion., Other forms of neurofibromatosis (segmental/mosaic NF1, Watson syndrome)., Other conditions with “cafe au lait spots” or with pigment changes confused with “cafe ?au lait” (LEOPARD syndrome, Peutz-Jeghers syndrome, McCune-Albright syndrome).

Final Diagnosis: NF1 with abdominal low-grade MPNST and breast lesion.

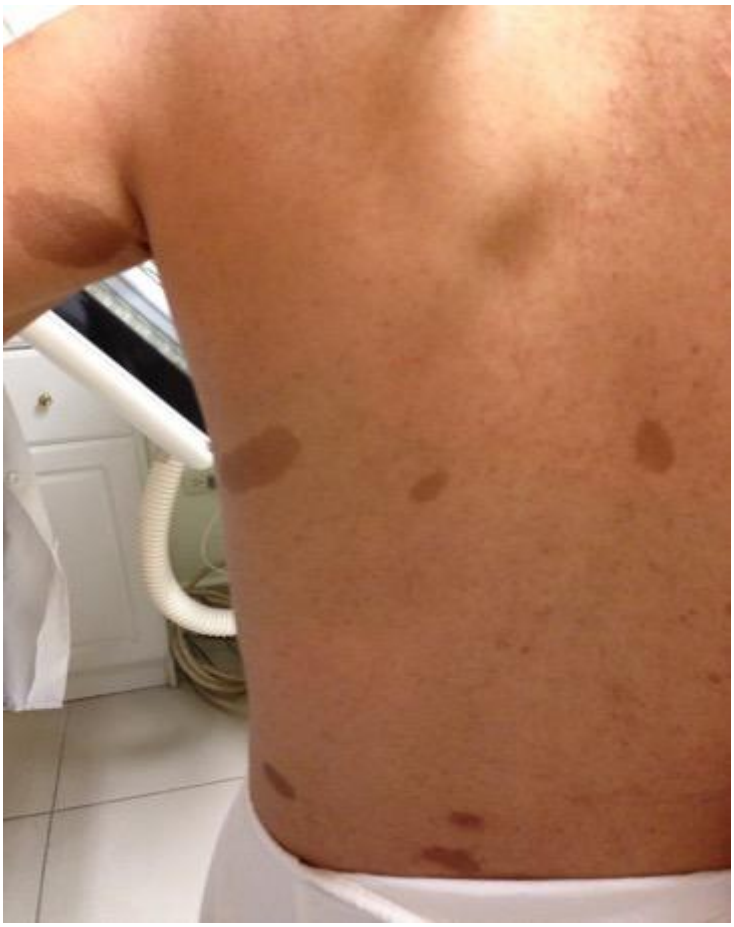
References:

Levy A, Patel N, Dow N, Abbott R, Miettinen M, Sobin L (2005) Abdominal Neoplasms in Patients with Neurofibromatosis Type 1. RadioGraphics 1. 25(2), pp.455-480. (PMID: 15798063)
Gokalp G, Hakyemez B, Kizilkaya E, Haholu A (2007) Myxoid neurofibromas of the breast: mammographical, sonographical and MRI appearances. The British Journal of Radiology 80, e234–e237 (PMID: 17959912)

Levy A, Manning M, Miettinen, M (2017) Soft-Tissue Sarcomas of the Abdomen and Pelvis: Radiologic-Pathologic Features, Part 2—Uncommon Sarcomas. RadioGraphics 1. 37(3), pp.797-812. (PMID: 28493803)
McDonnell, O. (2008) Mammographic Appearances of Neurofibromatosis,. http://www.eurorad.org/case.php?id=6369 CASE.6369
Hirbe, A. and Gutmann, D. (2014) Neurofibromatosis type 1: a multidisciplinary approach to care. The Lancet Neurology 13(8), pp.834-843 (PMID: 25030515)
Friedman JM. (1998) Neurofibromatosis 1. GeneReviews Seattle (WA): University of Washington, Seattle; 1993-2017. (PMID: 20301288)
Ferner R. E, Huson S. M, Thomas N, Moss C, Willshaw H, Evans D. G, Kirby A. (2007) Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. Journal of Medical Genetics 44(2), 81–88. (PMID: 2598063)
Porter GJ, Evans AJ, Lee AH, Hamilton LJ, James JJ. (2006) Unusual benign breast lesions. Clinical Radiology 61, 562–569 (PMID: 16784941)
Bongiorno M, R Doukaki S, Aricó M. (2010) Neurofibromatosis of the nipple-areolar area: a case series. Journal of Medical Case Reports 4, 22 (PMID: 2823760)
Khalil J, Afif M, Elkacemi H, Benoulaid M, Kebdani T, Benjaafar N. (2015) Breast cancer associated with neurofibromatosis type 1: a case series and review of the literature. Journal of Medical Case Reports 9,61 (PMID: 4372231)
Tonsgard, J (2006) Clinical Manifestations and Management of Neurofibromatosis Type 1. Seminars in Pediatric Neurology 13(1), pp.2-7 (PMID: 16818170)

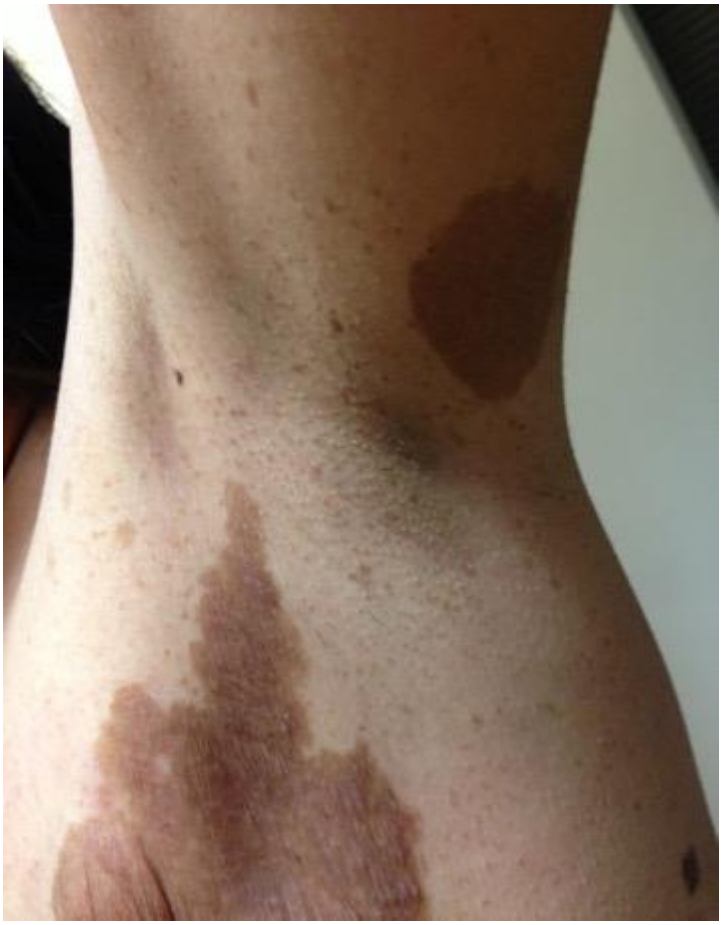
Figure 1

a



Description: Multiple back cutaneous lesions, “cafe au lait” spot. **Origin:** Departamento de Radiología, Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

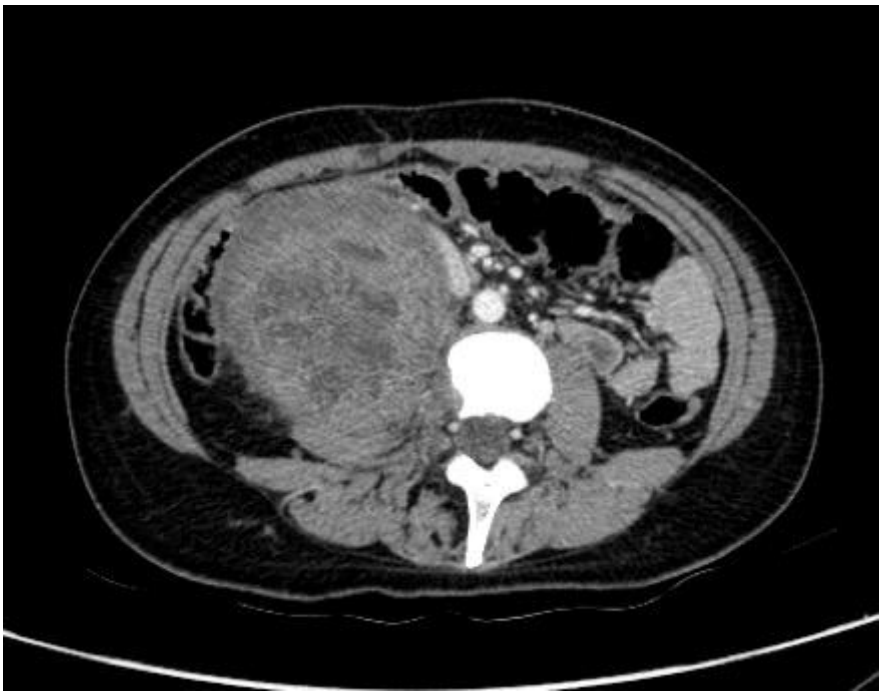
b



Description: Axillary freckling **Origin:** Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

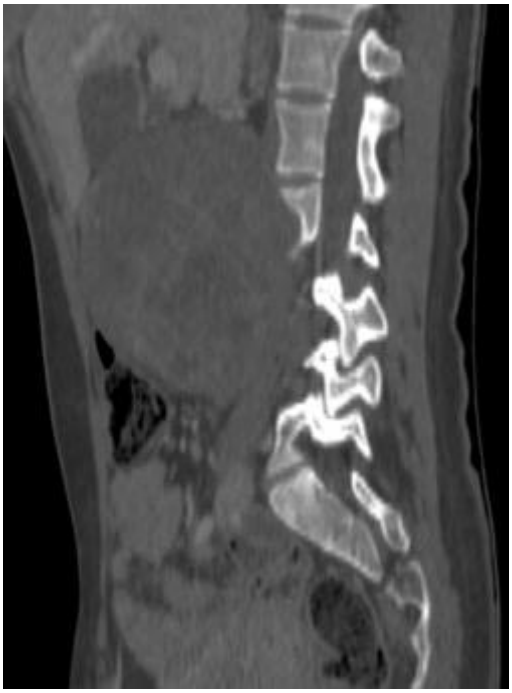
Figure 2

a



Description: Axial CT showing a retroperitoneal mass, hypodense, with heterogeneous enhancement and central necrosis, which displaces the ascending colon. **Origin:** Departamento de Radiología, Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

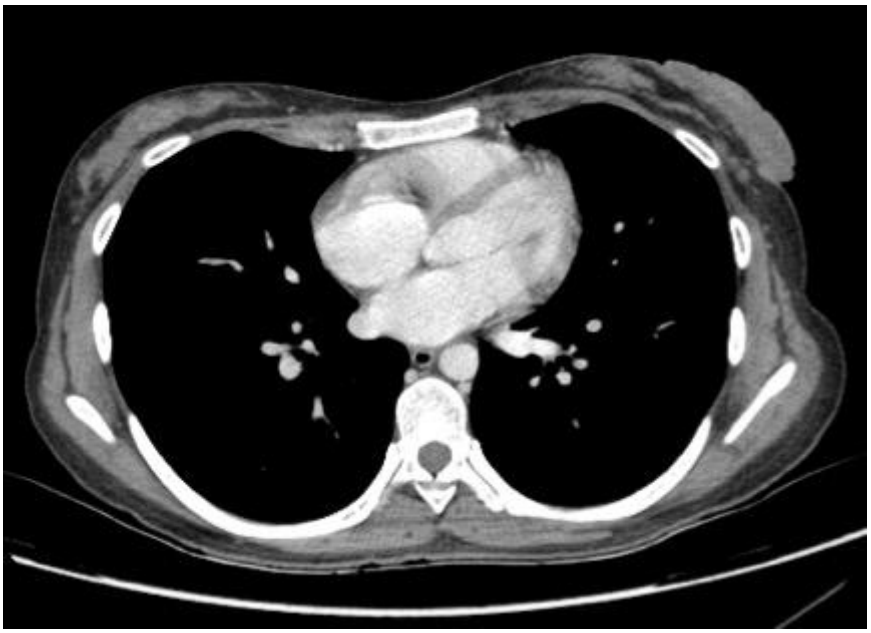
b



Description: Sagittal CT reconstruction at the level of the retroperitoneal mass. **Origin:** Departamento de Radiología, Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

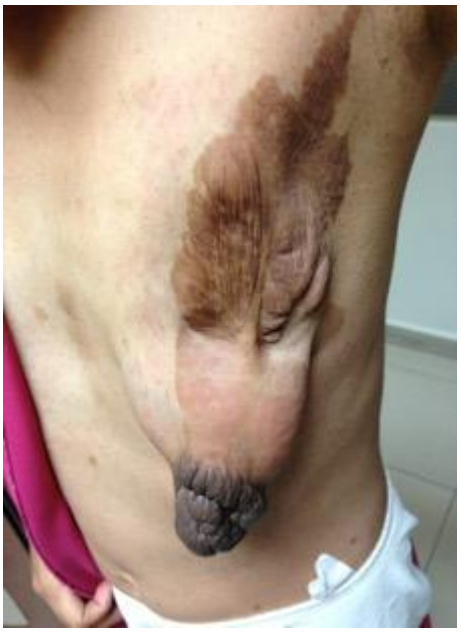
Figure 3

a



Description: Axial CT of the chest shows an exophytic tumour found in the complex nipple-areola of left breast. **Origin:** Departamento de Radiología, Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

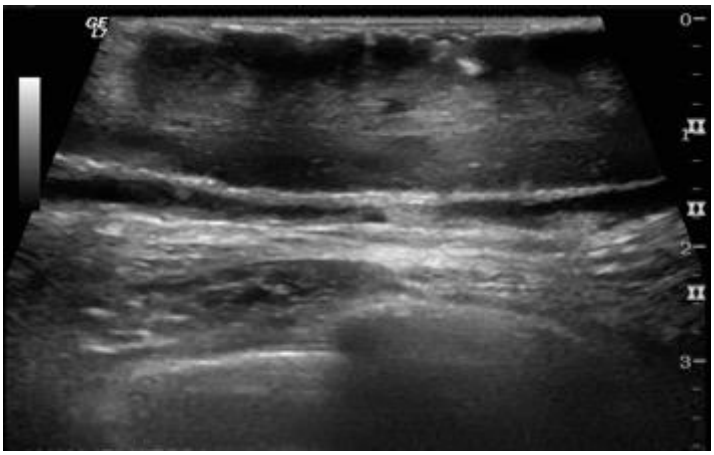
b



Description: Aspect of the exophytic tumour in the physical examination, presenting a dark brown colour. **Origin:** Departamento de Radiología, Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

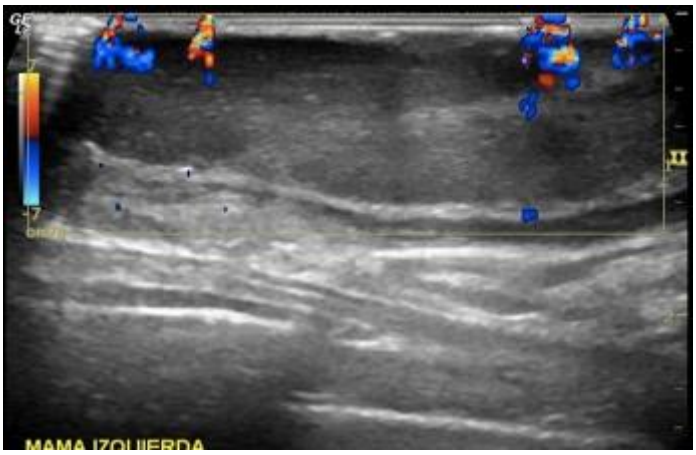
Figure 4

a



Description: Shows an oval, hypoechoic dermal lesion with lobed borders. **Origin:** Departamento de Radiología, Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

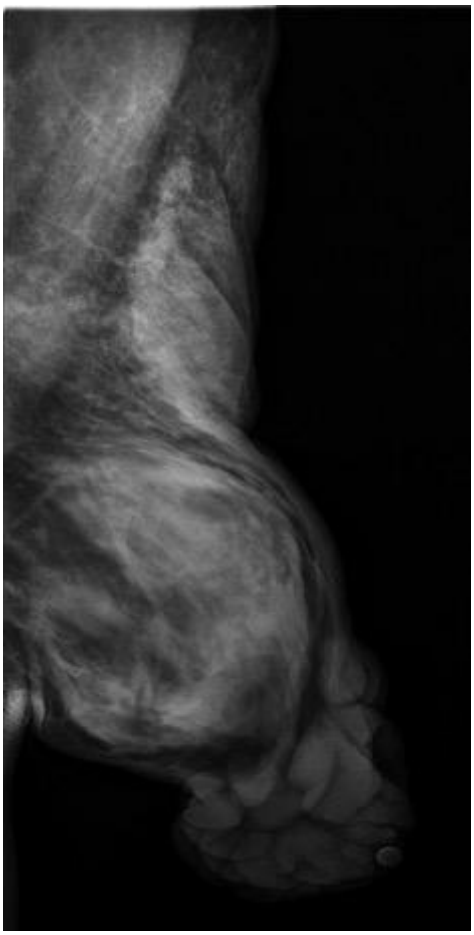
b



Description: No vascular flow was demonstrated in the examination with Doppler. **Origin:** Departamento de Radiología, Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

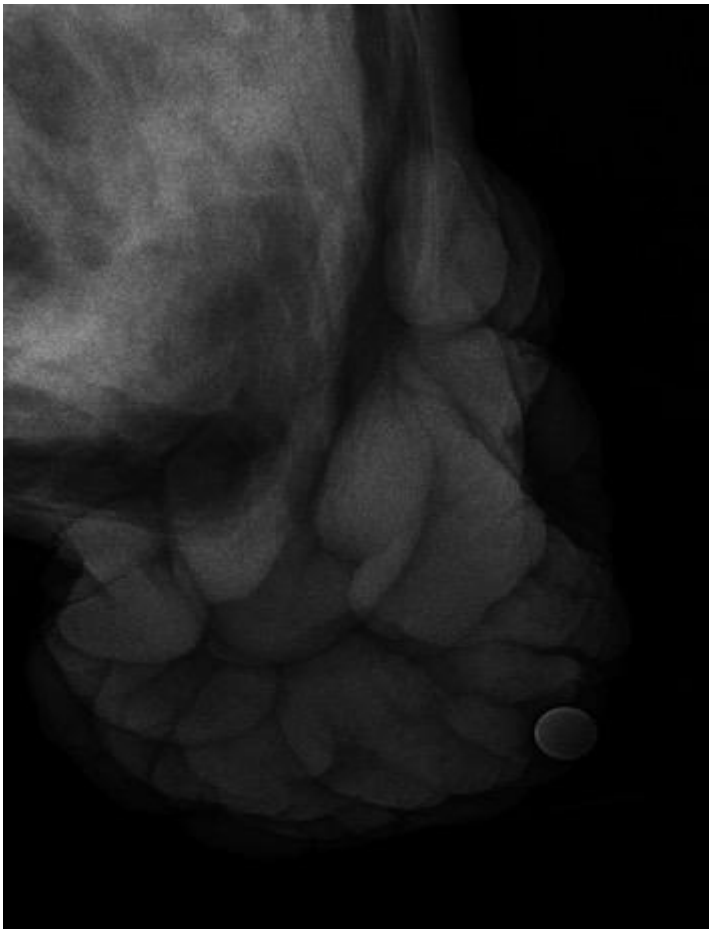
Figure 5

a



Description: Left MLO view of mammography presents an exophytic tumour in the topography of the nipple. **Origin:** Departamento de Radiología, Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

b



Description: Zoom of the left MLO view shows a multilobulated and isodense mass. **Origin:** Departamento de Radiología, Facultad de Medicina y Hospital Universitario “José Eleuterio González”, Universidad Autónoma de Nuevo León. Monterrey, Nuevo León, México

Figure 6

a

Diagnostic criteria for Neurofibromatosis type 1 (clinical criteria established by the National Institutes of Health Consensus Development Conference 1988); at least two of them are required to perform the diagnosis:
1. More than 6 “cafe au lait” spots evident during one year (5 mm in greatest diameter in prepubertal individual or 15 mm in postpubertal individuals).
2. Two or more neurofibromas or one plexiform neurofibroma.
3. Optic nerve glioma.
4. Distinctive osseous lesion (sphenoid wing dysplasia or tibial pseudarthrosis).
5. Two or more iris hamartomas (Lisch nodules).
6. Axillary or inguinal freckling.
7. Primary relative with NF1 with above criteria.

Description: Diagnostic criteria for Neurofibromatosis type 1 (NIH consensus development conference 1988). **Origin:** Taken from Ferner, R. E., Huson, S. M., Thomas, N., Moss, C., Willshaw, H., Evans, D. G., Kirby, A. (2007) Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. Journal of Medical Genetics 44(2), 81–88.