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Radiological Case Report / Caso Clínico

Plexiform Neurofibroma of the Breast: A Rare Presentation

Neurofibroma Plexiforme da Mama: Uma Apresentação Rara

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Abstract

Plexiform neurofibromas are benign peripheral nerve sheath tumors that commonly occur in individuals with Neurofibromatosis Type 1 and are considered pathognomonic for the disorder. We describe the case of a 21-year-old woman who presented with a painful palpable mass on her left breast and multiple congenital pigmented skin lesions. Imaging revealed replacement of the normal breast parenchyma by tortuous tubular structures extending into the chest wall, demonstrating T2 hyperintensity with a central hypointense focus. Histopathologic examination of the cutaneous lesions confirmed plexiform neurofibromas, establishing the diagnosis. This case represents an uncommon presentation of breast involvement and emphasizes the key role of Magnetic Resonance Imaging in recognizing characteristic imaging features and delineating lesion extent. Early diagnosis of Neurofibromatosis Type 1 and ongoing radiologic surveillance are crucial for detecting potential malignant transformation and ensuring appropriate multidisciplinary follow-up.

Keywords

Neurofibromatosis 1; Neurofibroma plexiform; Breast neoplasms; Magnetic resonance imaging; Peripheral nerve sheath neoplasms.

Resumo

Os Neurofibromas Plexiformes são tumores benignos com origem na bainha dos nervos periféricos e são considerados patognomónicos da Neurofibromatose Tipo 1. Descreve-se o caso de uma mulher de 21 anos com uma massa palpável e dolorosa na mama esquerda, associada a múltiplas lesões cutâneas pigmentadas congénitas. A avaliação imagiológica revelou uma substituição completa do normal parênquima mamário por estruturas tubulares tortuosas, com extensão e infiltração pela parede torácica, demonstrando hipersinal T2 com foco central hipointenso. A análise histopatológica das lesões cutâneas confirmou tratar-se de neurofibromas plexiformes, estabelecendo o diagnóstico. Este caso representa uma apresentação incomum deste tipo de lesão, com envolvimento mamário, destacando o papel fundamental da Ressonância Magnética na identificação das características típicas e na delimitação da extensão das lesões. O diagnóstico precoce da Neurofibromatose Tipo 1 e a vigilância clínica e imagiológica são essenciais para a deteção de possível transformação maligna e para assegurar um acompanhamento multidisciplinar adequado.

Palavras-chave

Neurofibromatose tipo 1; Neurofibroma plexiforme; Neoplasias da mama; Ressonância magnética; Neoplasias da bainha do nervo periférico.

Case Presentation

A 21-year-old female patient, originally from Angola and currently residing in Portugal, with no known prior medical conditions, sought medical observation due to persistent, long-lasting pain in the left breast. During a surgical consultation, physical examination revealed a large, irregular and painful palpable mass, which prompted the request for further diagnostic investigation.

Concurrently, the patient had been referred for a Dermatology consultation due to multiple scattered skin lesions, all of which she reported having been present since birth. Two large skin lesions were noted on her left breast and described as oval/circular plaques, measuring 5 cm and 8 cm in their largest diameter, darkly pigmented with an irregular surface and velvety texture (Figure 1). When asked about her family's medical history, the patient mentioned a sister with similar lesions and birthmarks. A large, exophytic, pedunculated lesion in the sacral region was excised.



Figure 1 – left breast cutaneous plaques, darkly pigmented with an irregular surface and velvety texture.

Breast ultrasound (US) of the right breast revealed normal fibroglandular tissue with dense distribution across all quadrants, while the left breast showed multiple tubular-looking structures containing heterogeneous hypoechogenic material and exhibiting mostly peripheral Doppler flow (Figure 2). These findings led to further assessment with Magnetic Resonance Imaging (MRI).

MRI showed the normal left breast tissue completely replaced by tortuous tubular structures, infiltrating the chest wall extending to the mediastinum, with hyperintensity on fat-suppressed T2-weighted images and a central hypointense "target sign", as well as very slight central delayed contrast enhancement. The skin surface lesions could also be seen on axial MR-imaging as T2 hyperintense and contrast-enhancing structures (Figure 3).

Histologic analysis of the removed sacral lesion revealed a neurofibroma with plexiform growth pattern. Subsequent biopsies of the two skin lesions on the patient´s left breast confirmed the same. The presence of one or more plexiform neurofibromas, as well as numerous café-au-lait spots (flat, light to dark brown pigmented birthmarks), establish the diagnosis of Type 1 Neurofibromatosis (NF1). The breast lesions were not biopsied, as they presented typical features in this clinical context – only required if presenting suspicion for malignancy. The patient was referred to a specialized center for surveillance and evaluation of potential malignant change in her skin lesions.

Discussion

Plexiform neurofibromas (PN) are benign tumors that arise from peripheral nerve sheaths. They occur in up to 40-50% of patients with NF1, for which they are regarded as pathognomonic. These lesions are typically congenital and identified in early childhood, appearing as superficial/subcutaneous masses anywhere on the body, often enlarging during childhood and puberty.[1,2] The most common site of plexiform neurofibromatosis is along the trigeminal nerve distribution (head and neck), but it can also occur on the trunk and extremities.[3]

The term "plexiform" refers to the histopathologic pattern which resembles a plexus or network, which may be grossly apparent or otherwise visible only on microscopic examination.[4,5]

PN can exhibit diffuse or nodular growth along long nerve segments and their branches, sometimes extending beyond

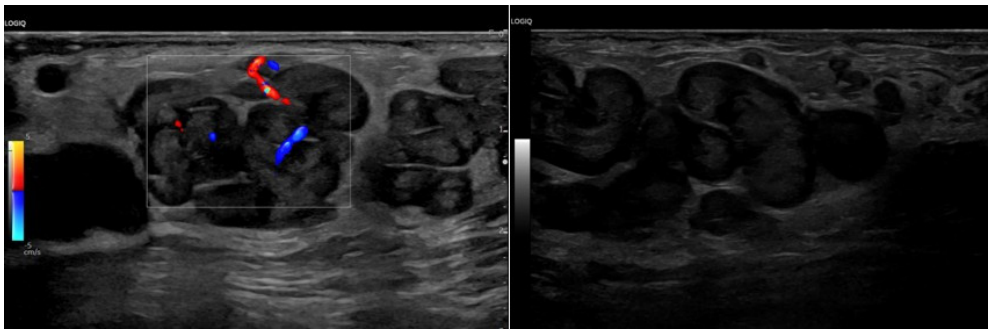


Figure 2 – Left breast ultrasound revealed multiple tubular-appearing structures containing heterogeneous hypoechogenic material with mostly peripheral Doppler flow.

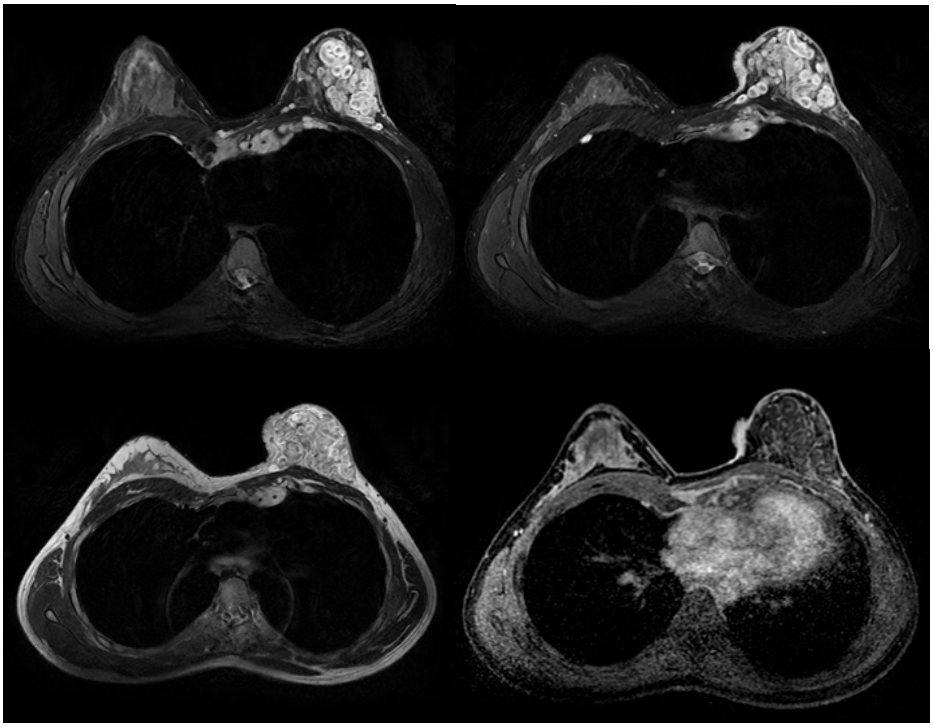


Figure 3 – MRI - left breast tissue completely replaced by tortuous tubular structures, deeply infiltrating into the chest wall, with T2 hyperintensity and a central hypointense focus ("target sign"), as well as very slight central delayed contrast enhancement. Skin surface lesions appear as T2 hyperintense and contrast-enhancing structures.

the epineurium into surrounding tissues such as skin, subcutaneous tissue and muscle. They may also deeply infiltrate organs or bones, causing disfigurement and/or dysfunction.[1] Patients can be asymptomatic or notice pain, change in texture or sudden increase in growth, potentially causing symptoms related to compression and local mass effect, depending on their location. For example, patients may experience bowel obstruction, bladder dysfunction, breathing difficulty or neurologic deficits secondary to spinal cord compression.

The MRI appearance of PN may vary depending on their location and depth of infiltration. Superficial PN are more common and they typically show a diffuse or infiltrating morphology, often with poorly defined margins, extending towards the skin surface and involving the subcutaneous tissues in a reticular and branching configuration. These lesions are predominantly hyperintense on T2-weighted images and may be mistaken for venous malformations. Deep PN manifest as masses composed of multiple nodules or varicose tubular structures, often described as resembling a "bag-of-worms". They show T2-weighted hyperintensity and exhibit a characteristic "target" appearance, with a central zone of low signal intensity surrounded by a peripheral zone of higher intensity6 (Figure 3).

Surveillance recommendations for individuals with NF1 were established by the European Reference Network on Genetic Tumor Risk Syndromes. Although the PN are generally benign, there is a risk of malignant transformation, which increases with age. Clinical assessment for PN should begin at the time of NF1 diagnosis or at birth. Whole-body MRI is recommended at least once, during the transition from childhood or adolescence to adulthood, to assess the internal tumor burden, which serves as a predictor for the development of malignant peripheral nerve sheath tumors. When a PN is identified, the lesion should be clinically monitored and evaluated for possible malignant transformation. Assessment should include tumor growth (increase or change in growth rate), change in consistency or texture, new or persistent pain, recent motor or sensory deficits. 18F–fluorodeoxyglucose (FDG) PET/MRI may be useful as there is higher uptake in malignant peripheral nerve sheath tumors than in benign PN. For symptomatic plexiform neurofibroma, surgery should be considered, as it is the only potentially curative treatment. NF1 is associated with an increased risk of breast malignancy in patients younger than 50, with screening recommended to begin at age 30 with yearly MR.[2]

This case demonstrates a rare presentation of plexiform neurofibromas involving the breast in a young woman with previously undiagnosed Neurofibromatosis Type 1. MRI played a key role in characterizing the extent and nature of the lesions, revealing features typical of deep plexiform neurofibromas, such as the "target sign" and diffuse infiltration of subcutaneous tissues. Early recognition of NF1 is essential for appropriate multidisciplinary management and surveillance, given the lifelong risk of complications, including malignant peripheral nerve sheath tumor transformation. This case also highlights the Radiologists' role as a key member of the multidisciplinary team in the detection of these lesions, ensuring comprehensive care and timely intervention for patients with NF1.

Ethical Disclosures / Divulgações Éticas

Conflicts of interest: The authors have no conflicts of interest to declare.

Conflitos de interesse: Os autores declaram não possuir conflitos de interesse.

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Confidentiality of data: The authors declare that they have followed the protocols of their work center on the publication of data from patients.

Confidencialidade dos dados: Os autores declaram ter seguido os protocolos do seu centro de trabalho acerca da publicação dos dados de doentes.

Protection of human and animal subjects: The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Protecção de pessoas e animais: Os autores declaram que os procedimentos seguidos estavam de acordo com os regulamentos estabelecidos pelos responsáveis da Comissão de Investigação Clínica e Ética e de acordo com a Declaração de Helsínquia da Associação Médica Mundial.

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