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# Erdheim-Chester disease: a case report

## *Doença de Erdheim-Chester: relato de caso*

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**ABSTRACT:** Erdheim-Chester disease is a rare non-Langerhans cell histiocytosis, first described by Jakob Erdheim and William Chester in 1930 of unknown etiology. Clinically, it can reach all organs and systems and the diagnosis is based on radiological and anatomopathological findings. It was reported a case of the disease where the clinical, radiological and anatomopathological findings.

**Keywords**: Erdheim-Chester disease; Immunohistochemistry; Xanthogranuloma

**RESUMO:** A doença de Erdheim-Chester é uma rara histiocitose de células não-Langerhans, primeiramente descrita por Jakob Erdheim e William Chester em 1930 de etiologia desconhecida. Clinicamente, pode atingir todos os órgãos e sistemas e o diagnóstico baseia-se em achados radiológicos e anatomopatológicos. Foi relatado um caso da doença onde se estudaram os achados clínicos, radiológicos e anatomopatológicos.

**Palavras-chaves**: Doença de Erdheim-Chester; Imunohistoquímica; Xantogranuloma

## CASE REPORT

A 40-year-old female patient was admitted with neurological symptoms and elevated skin lesions to be investigated. Physical examination revealed multiple nodular and hyperchromic lesions on the scalp, face and breast. In addition, they observed other lesions with characteristics similar to those previously mentioned on large lips and Genital-femoral grooves to the vulvoscopy and massive lesion in the cervix of the uterus, proceeding close to the pelvic wall to colposcopy. Magnetic resonance imaging of the total abdomen showed expansive and circumferential nodular formation in the uterine cervix, with irregularity of the right lateral wall of the cervical canal, presenting an approximate extension of 5.7 cm from the plane of the hysterorrhaphy in the anterior isthmic region to the external orifice of the bulging neck and with apparent discontinuity of the cervical fibrous stroma in both lateral walls, inferring impairment of both proximal paramétriums. Magnetic Resonance of the skull showed nodular lesions with isosignal at T1, discrete hypersignal at T2, and intense contrast enhancement, without restriction to diffusion, sparse both infra and supratentorial, the largest thus located: right upper front swivel measuring 5 mm; left mid-front swivel measuring 5 mm; left hippocampal gyrus measuring 17 mm, upper temporal gyrus left measuring 5 mm; right cerebellar hemisphere measuring 7 mm. Brain nodular lesions, more compatible with secondary dissemination. Nuclear Magnetic Resonance showed a circumscribed nodule in the middle / posterior third of the super-lateral quadrant (10 hours) of the right breast. Nodules with bilateral annular enhancement and circumscribed nodule with heterogeneous enhancement on the periphery of the inferolateral quadrant (8 hours) of the right breast. Upper gastrointestinal endoscopy demonstrated esophageal subepithelial lesions. A biopsy of such lesions was performed and the anatomopathological result was xanthogranuloma. Subsequent immunohistochemical

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examination evidenced cytoplasmic labeling for CD68 and absence of labeling for S-100, consistent with the diagnosis of Erdheim-Chester disease. The patient is well and in outpatient follow-up.

## DISCUSSION

Erdheim-Chester disease is a rare form of non-Langerhans cell histiocytosis that occurs most frequently after 40 years of age, with a slight predominance in men[1]. Recent studies have shown that these patients have mutations in the proto-oncogene BRAF (and exceptionally in some genes related to activation of MAPK pathway), implying a crucial part on this pathway in the pathogenesis and besides a potential clonal origin of the illness[2]. Clinically all organs and systems can be affected. The most common clinical features include skeletal involvement with bilateral sclerotic lesions of long bones of the lower limbs, diabetes insipidus, cardiovascular involvement and retroperitoneal fibrosis. The most frequent cutaneous manifestations of the disease are papillomatous lesions with smooth surface and regular xanthelasma-like contours[3]. Involvement of the central and cardiovascular nervous system are indicators of worse prognosis[4,5]. Diagnosis is based on radiological and pathological findings[6,7]. Histologically there appears to be a tropism of histiocytes by perivascular connective tissue and adipose, with infiltrative pattern and local fibrosis that manifests microscopically as a dense histiocytic infiltrate, sometimes containing giant cells multinucleated and without epidermotropism. The immunohistochemical examination should present CD68 positive and S-100 negative.

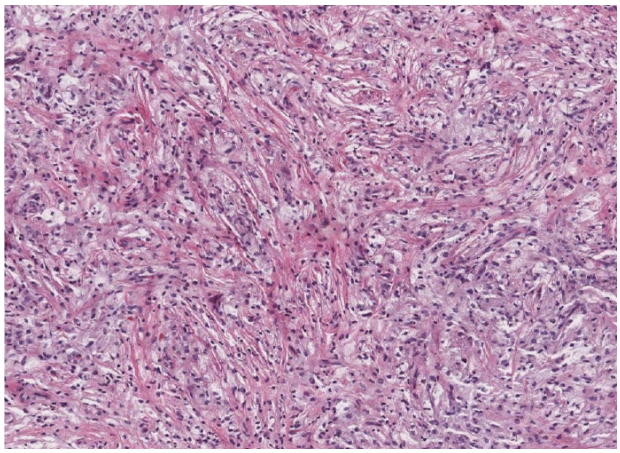

**Figure 1**: Histological examination of breast injury with dense histiocytic infiltrate

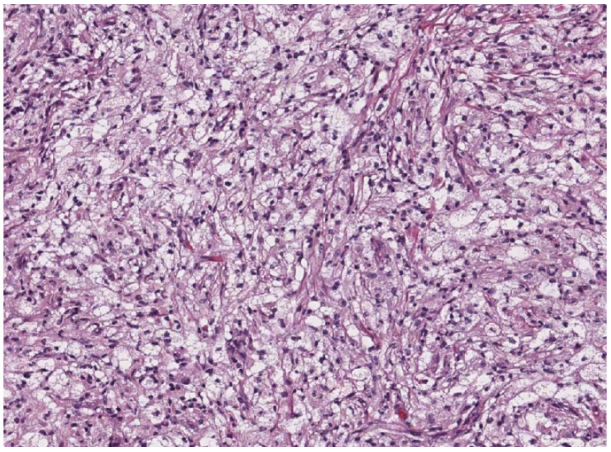

**Figure 2:** Histological examination of cervical lesion with dense histiocytic infiltrate

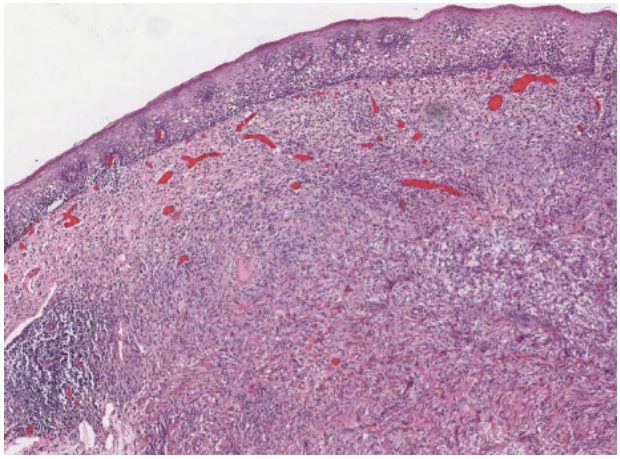

**Figure 3:** Histological examination of esophageal lesion with dense histiocytic infiltrate

## CONCLUSION

The diagnosis of Erdheim-Chester disease can be challenging and requires a multidisciplinary approach. The recognition of radiological and histological characteristics is essential. There is a new consensus on the individualized treatment of Erdheim-Chester disease, showing promising resultus with the use of corticosteroids, immunosuppressive drugs such as cyclophosphamide, chemotherapy, radiation therapy and establish mutational status to potentially guide therapy. The prognosis depends on the extent of visceral impairment at the time of diagnosis.

**Authors' participation:** *Aline Alencar Giongo*: wrote the manuscript and provide clinical and pathological information. *Bruno Santos Leal Campos*: wrote the manuscript and provide clinical information. *Tercia Jovino Neves Santos*: wrote the manuscript and provide clinical and pathological information. *Felipe D'Almeida Costa*: diagnosed the case and read and revised the manuscript.

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