SOHO ANNUAL MEETING 2024

# An Unusual Presentation of Erdheim-Chester Disease: A Case Report

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## INTRODUCTION

Erdheim-Chester Disease (ECD) is a rare form of non-Langerhans cell histiocytosis with only a few hundred cases reported worldwide. It was previously thought to be a non-neoplastic inflammatory disorder, but as a result of increasing number of cases harboring the BRAFV600E mutations, it is now better described as a clonal neoplastic disorder with a characteristic inflammatory cytokine profile.

## CONTEXT

This case study documents a rare challenging presentation of ECD

## PATIENT

A 29-year-old female patient presented with left orbital pain and swelling, with no previous orbital or ocular complaints. She was otherwise asymptomatic. General examination was unremarkable

## RESULTS

MRI Orbit was ordered which showed a medial orbital mass measuring 3 x 2 cm with periosteal attachment. Surgical excision of the mass was performed and pathological examination revealed surface stratified squamous epithelium. The subepithelial tissue showed proliferation of oval mononuclear cells with pale cytoplasm with interspersed inflammatory cell infiltrate and multinucleated giant cells, plasma cells & lymphocytes.

Immunohistochemistry using CD68, CD1a , CD207 (Langerin), S100 and BRAF revealed
CD68: Diffuse positive staining.
CD1a: Negative
S100: Negative
CD207: Negative
BRAF: Negative

BRAF-V600E mutational analysis was positive. Subsequently, the patient was diagnosed with ECD. Further investigations including post contrast CT scan of the chest, abdomen and pelvis, and MRI brain with gadolinium, excluded further masses and any systemic involvement. A bone marrow examination revealed a normocellular marrow with no abnormal deposits. Blood tests including a complete blood count, comprehensive metabolic panel, ESR and CRP were unremarkable.

As our patient presented with a single mass that was surgically excised, no further treatment was initiated.

## OUTCOMES

Our patient was advised to follow up every three months for the first year during which no further masses or symptoms developed. PET CT scans at the six- and twelve-month were free.

## CONCLUSION

This is a rare case report of ECD presenting with a solitary retro-orbital mass. Reporting such cases may help to delineate their actual incidence and prognosis.

## ACKNOWLEDGEMENTS

Gaurav Goyal, Mark L. Heaney et al; Erdheim-Chester disease: consensus recommendations for evaluation, diagnosis, and treatment in the molecular era. *Blood* 2020

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