

## Case Report

# An Unusual Case of Bilateral Orbital Infiltrative Disease and Papillary Thyroid Cancer

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## Abstract

Erdheim-Chester disease (ECD) is a non-Langerhans cell histiocytic disorder characterized by infiltration of foamy histiocytes in multiple organs. Herein, we describe an unusual presentation of ECD with concomitant diagnosis of papillary thyroid carcinoma. The majority of the cases of ECD are BRAF V600E mutation positive. Whether there is an increased risk of BRAF mutant papillary thyroid carcinoma in patients with ECD needs to be further investigated. ECD is undoubtedly a rare disease, however it is an overlooked diagnosis and raising awareness of ECD in the medical community is highly warranted.

## INTRODUCTION

A 38-year-old woman presented with swelling of bilateral eye lids and yellowish discoloration of the overlying skin (Figure 1).

On evaluation, magnetic resonance imaging (MRI) of the head revealed symmetrical orbital infiltrative processes of unknown origin (Figure 2). Work up for Graves' disease with thyroid-stimulating immunoglobulin testing was negative. Given prior history of follicular hyperplasia of a right inguinal lymph node, a PET/CT scan was performed to rule out lymphoproliferative disorder. Marked FDG uptake (SUV max 7.8) of the extra ocular musculature was noted (Figure 3) together with an intensely FDG avid (SUV max 10.9) nodule within the posterior left thyroid lobe (Figure 4). No bony involvement was identified.

Patient underwent biopsy of the left orbital mass and a xanthogranulomatous inflammation was noted on pathology. Immunoperoxidase studies revealed IgG4 positive and CD1a negative cells, supporting the diagnosis of Erdheim-Chester disease (ECD). BRAF V600E mutation was negative. Subsequently, fine needle aspiration of the 1.6 cm thyroid nodule was consistent with papillary thyroid carcinoma (PTC).



Figure 1 Cutaneous involvement of the skin (xanthelasma).

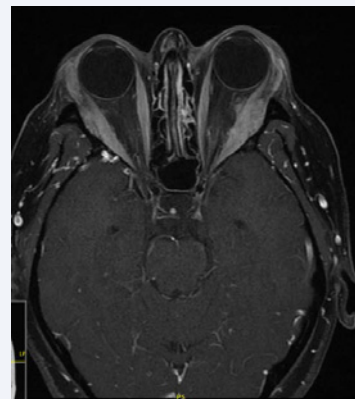


Figure 2 <sup>18</sup>FDG PET/CT scan. High FDG uptake of bilateral orbital muscles.

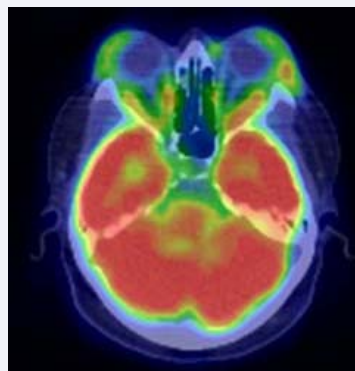
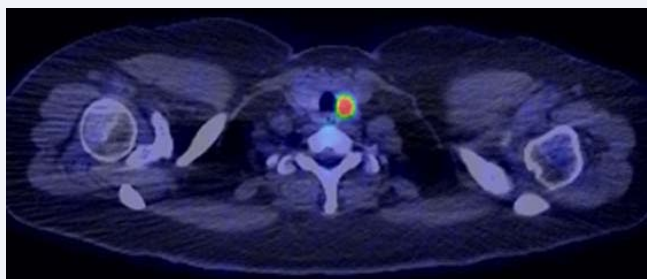


Figure 3 MRI of the head-bilateral symmetrical infiltration of the orbital musculature.



**Figure 4**  $^{18}\text{F}$ FDG PET/CT scan. Focal FDG uptake of the left thyroid gland.

ECD is a rare non-Langerhans histiocytic disorder, characterized by infiltration of tissues by foamy lipid-laden macrophages [1]. Clinical presentation is highly variable with most patients presenting with osseous involvement and orbital disease is seen in only 22% of the cases [2]. The pathogenesis is poorly understood, however an activating point mutation of the proto-oncogene BRAF V600E is identified in 38-100% [3,4] of cases of ECD and an NRAS mutation in some BRAF V600E-negative ECD patients. Two case reports of concomitant ECD and papillary thyroid carcinoma have been reported in the literature [5,6]; whether there is a true association between ECD and BRAF mediated tumors, such as papillary thyroid cancer remains unknown. ECD is undoubtedly a rare disease; however it is an overlooked diagnosis and raising awareness of ECD and its potential association with PTC in the medical community, is warranted.

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