

Erdheim-Chester Disease: A Case Report Exploring the Crucial Role of Multimodality Imaging and Targeted Specimen Re-review

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Abstract

Erdheim-Chester disease (ECD) is a rare multisystemic, non-Langerhans cell histiocytic neoplasm that is becoming increasingly recognized. The discovery of somatic mutations in the mitogen-activated protein kinase signaling pathway, most commonly *BRAF*^{V600E}, has led to the disease being reclassified from an inflammatory process to a neoplastic one. The disease often presents with non-specific symptoms, most commonly bone pain, often leading to a delay in diagnosis, with the diagnosis often first being suggested by imaging findings. We present the case of a 56-year-old gentleman who presented with an atypical, five-year prodrome of progressive proximal muscle weakness, fatigue, hyperreflexia, and focal fasciculations, initially mimicking a primary neuromuscular or motor neuron disease. Subsequent multimodality imaging revealed classic features of systemic histiocytosis, including symmetric appendicular osteosclerosis on bone scintigraphy, retro-orbital masses with exophthalmos on brain magnetic resonance imaging, and perinephric soft tissue infiltration ("hairy kidney") and periaortic encasement ("coated aorta") on abdominal computed tomography, alongside primary testicular involvement on positron emission tomography. Although an initial retroperitoneal tissue biopsy yielded non-diagnostic fibroadipose tissue, specimen re-review histopathologically confirmed the *BRAF*^{V600E} mutation. The patient was initiated on targeted kinase inhibitor therapy with vemurafenib. This report highlights the vital role of multimodality imaging in suggesting the diagnosis and the necessity of expert tissue re-review or molecular sequencing when initial biopsy results appear non-specific.

Categories: Radiology, Pathology, Rheumatology

Keywords: braf v600e mutation, cd68-positive, diabetes insipidus, ecd, hairy kidney, histiocytic neoplasm, histiocytosis, mapk pathway, non-langerhans cell histiocytosis, osteosclerosis

Introduction

Erdheim-Chester disease (ECD) is a rare form of non-Langerhans cell histiocytosis, which involves xanthogranulomatous infiltration of tissues by foamy histiocytes, lipid-laden macrophages, or histiocytes, surrounded by fibrosis. In ECD, the characteristics of histiocytes are positive for CD68, negative for CD1a, and negative for the S-100 protein in 80% of cases [1].

ECD most commonly affects adults with a mean age at diagnosis of 55 years, with a predominance in males, with men being three times more likely to be affected than women. The disease affects a variety of systems, and manifestations differ greatly between patients. Any of the clinical signs, such as bone pain, ataxia, xanthelasma, exophthalmos, or cardiac tamponade, may lead to a diagnosis of the disease. The most common first presentation of ECD is an unknown appearance of diabetes insipidus, which may occur several years before the diagnosis, followed by neurological manifestations. Due to the variability and rare presentation, misdiagnosis is often inevitable. ECD is currently diagnosed by histology and characteristic radiological findings, excluding alternative possibilities such as malignancy or an autoimmune disease [2].

Furthermore, the detection of a *BRAF*^{V600E} mutation or of another mitogen-activated protein kinase pathway alteration has become an important component of diagnostic confirmation of ECD in the absence of typical presentation.

Here, we present a case of ECD in a 56-year-old gentleman whose initial presentation mimicked a rheumatological or neuromuscular disorder. Rather than the onset of signature symptoms like diabetes insipidus, exophthalmos, and bone pain, the patient presented with progressive proximal muscle weakness, generalized fatigue, and combined upper and lower motor neuron signs leading to a differential diagnosis of statin-induced myopathy, motor neuron disease, or inflammatory myelitis. Furthermore, while ECD is most often diagnosed with characteristic histopathology on tissue biopsy, the initial biopsy in this case yielded inadequate samples for assessment, revealing only small fragments of foamy scattered histiocytes.

Molecular testing confirmed the presence of the *BRAF*^{V600E} mutation. This case emphasizes the necessity of considering ECD in non-typical neurological and rheumatological presentations and demonstrates the potential for molecular testing in a definitive diagnosis.

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Case Presentation

A 56-year-old gentleman was referred to the rheumatology clinic in August 2023 presenting with progressive muscle weakness and an elevation in C-reactive protein (CRP). He had a past medical history of hypertension, dyslipidemia, hemorrhoids, prediabetes, a prior lower leg benign bone growth resection (1986), a right eye pterygium repair (1999), and a colonoscopy showing active ileitis alongside mild iron-deficiency anemia (2022).

The patient's symptoms began in March 2020, with subtle imbalance and exertional fatigue during his walks to work. By early 2021, the patient noted upper extremity muscle achiness and fatiguability while performing heavy manual labor. Increasing muscle fatigue was also noted in the quadriceps and hamstrings while walking. An intentional 40-pound weight loss in April 2022 failed to improve his fatigue with an onset of cold peripheries, reduced grip strength, and brittle fingernails. By May of 2023, he noted markedly decreased finger dexterity and a burning sensation in his thighs proximally and symmetrically bilaterally. He reported difficulty walking characterized by often deviating either to the right or to the left.

Physical examination was largely unremarkable, revealing normal motor strength with no evidence of inflammation, myelitis, or connective tissue disease.

Laboratory investigations demonstrated a discordance between systemic inflammatory markers and primary neuromuscular testing (Table 1). Inflammatory myositis was deemed highly unlikely given normal creatine kinase levels in June 2023, antinuclear antibodies and rheumatoid factor in March 2023, and an absence of other connective tissue disease features. Recommendations were made to hold rosuvastatin to rule out statin-induced myopathy. However, a marked elevation in CRP (13.6 mg/L) in September 2023 confirmed an underlying active inflammatory process, prompting urgent referral to neurology and central neuroimaging.

Date	Laboratory test	Patient result	Reference range	Units
March 2023	C-reactive protein (CRP)	3.7	0.0-5.0	mg/L
	Rheumatoid factor (RF)	<10	<4	IU/mL
	Antinuclear antibodies (ANA)	Negative	Negative	-
	Thyroid-stimulating hormone (TSH)	1.93	0.35-5.00	mIU/L
June 2023	Hemoglobin	136	120-160	g/L
	Leukocytes (WBC)	7.1	4.0-11.0	×10 ⁹ /L
	Platelets	376	150-400	×10 ⁹ /L
	Creatinine	75	45-90	μmol/L
	Alanine aminotransferase (ALT)	21	<35	U/L
	Creatine kinase (CK)	55	30-170	U/L
September 2023	C-reactive protein (CRP)	13.6	0.0-5.0	mg/L
	Platelets	450	150-400	×10 ⁹ /L
	Alkaline phosphatase (ALP)	116	35-120	U/L
	Serum ferritin	221	15-240	μg/L
	Calcium	2.39	2.15-2.55	mmol/L
	Creatine kinase (CK)	44	30-170	U/L

TABLE 1: Initial diagnostic laboratory evaluation (March 2023 to September 2023)

The bold values are the values outside of the reference range.

Seven weeks following his initial visit to the rheumatology clinic, in September 2023, the patient was evaluated in an urgent neurology clinic for persistent proximal weakness in the bilateral thighs and arms, left hand tremor, and visuospatial changes. Despite withholding rosuvastatin, the patient reported no improvement. He reported a new burning sensation in his left arm, though primary sensory loss was absent during testing.

On physical examination, the patient demonstrated subtle bilateral wrist flexion weakness (4+/4) and subtle external shoulder rotation weakness (4+/4+). He also demonstrated fasciculations at the right biceps and right quadriceps and hyperreflexia at the brachioradialis as well as the knees and ankles bilaterally (3+ reflexes at the brachioradialis, knees, and ankles) with spread into the fingers and bilateral positive Trömner reflexes.

Given the combined upper and lower motor neuron findings, the differential diagnosis expanded to include a possible motor neuron disease. The patient was referred for an outpatient electromyogram and a nerve conduction study. Additionally, an urgent outpatient magnetic resonance imaging (MRI) of the brain and cervical spine was ordered to rule out a structural lesion.

Initial MRI of the head revealed bilateral, avidly enhancing intraconal masses that partially encased the optic nerves. There was mild displacement of the optic nerves without proptosis, and the nerves maintained a normal caliber. Intracranially, there were patchy, non-enhancing T2 hyperintensities within the deep cerebellar white matter, bilateral middle cerebellar peduncles, and pons, accompanied by moderate cerebral and mild cerebellar atrophy (Figure 1).

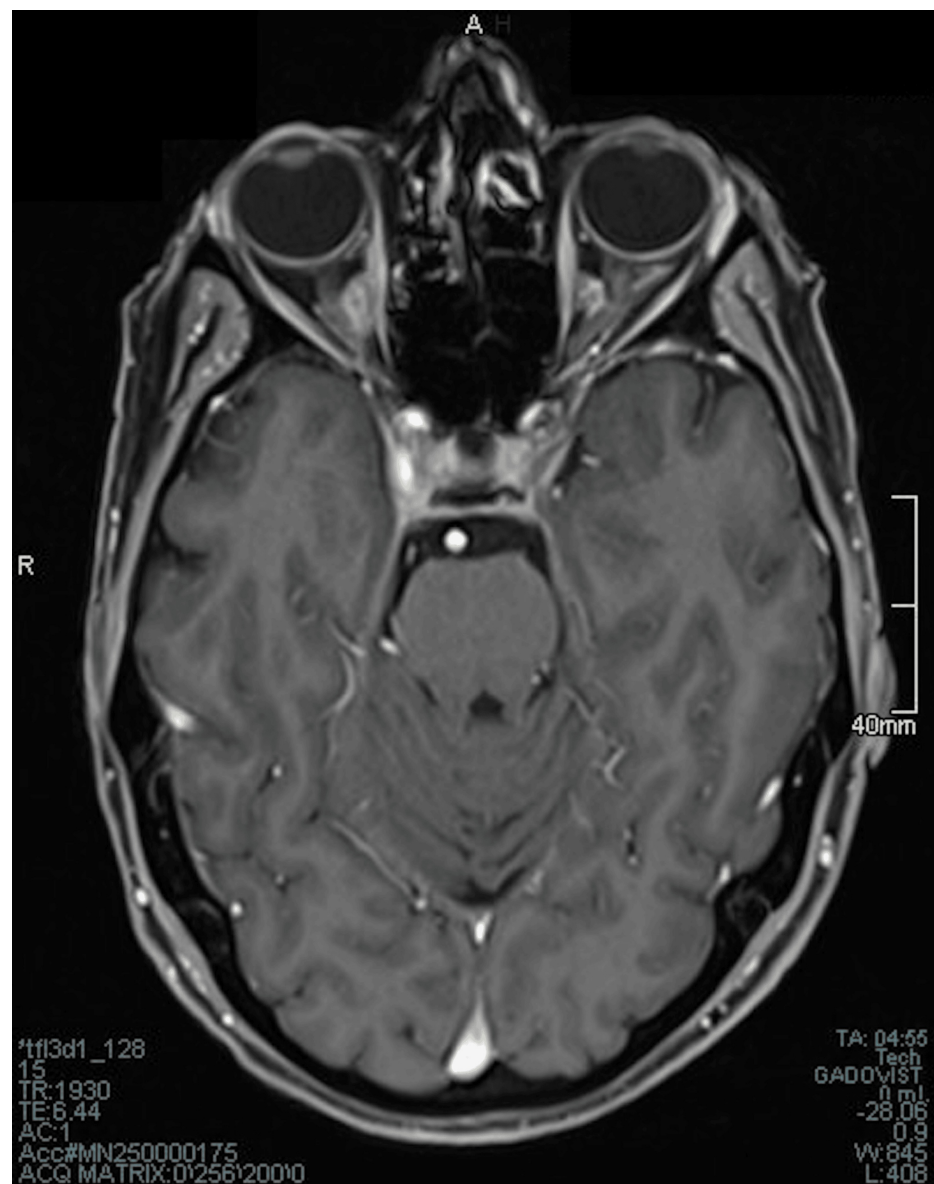


FIGURE 1: Axial magnetic resonance imaging scan of the head demonstrating bilateral avidly enhancing intraconal masses that partially encase the optic nerves, with mild displacement of the optic nerves, while maintaining a normal nerve caliber

A subsequent computed tomography (CT) performed in October 2023, as a broader workup, identified abnormal sclerotic foci within the L3 vertebral body, bilateral femurs, and sacrum, prompting targeted skeletal and retroperitoneal imaging.

In December 2023, total body delayed images and single-photon emission computed tomography/computed tomography (SPECT/CT) images of the lower arms/abdomen/pelvis/femurs were obtained following the intravenous administration of technetium-99m methylene diphosphonate (99mTc-MDP). The images revealed pronounced, symmetrical radiotracer uptake throughout the appendicular skeleton, specifically involving the distal femurs, proximal and distal tibias, ankles, bilateral radii, and humeri. Additional uptake was noted in the L3 vertebral body and the skull. This pattern of prominent increased MDP uptake symmetrically in the long bones was flagged as highly suspicious for ECD (Figure 2).

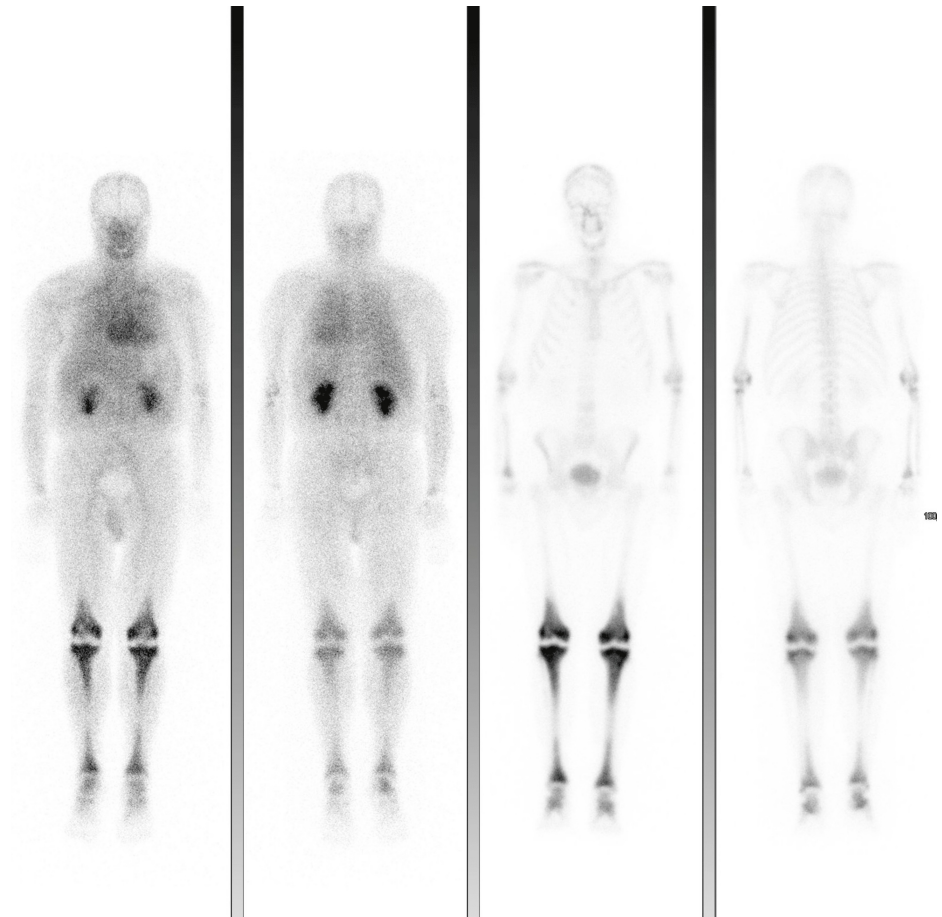


FIGURE 2: Total body delayed images and single-photon emission computed tomography/computed tomography images of the lower arms, abdomen, pelvis, and femurs following the administration of technetium-99m methylene diphosphonate

A contrast-enhanced CT of the thorax, abdomen, and pelvis in September 2024 revealed widespread soft tissue infiltrative changes classic for a multisystem histiocytosis. In the retroperitoneum, imaging depicted bilateral renal enlargement with prominent perinephric stranding and a thin rim of fluid, characteristic of the "hairy kidney" sign. This was accompanied by bilateral moderate hydronephrosis, thickened and enhancing urothelium, and soft tissue encasement of the proximal ureters (Figure 3). The adrenal glands were bilaterally bulky with surrounding inflammatory changes. Cardiovascular evaluation demonstrated aortic wall thickening and mild inflammatory changes extending into the iliac arteries, consistent with periaortitis or a "coated aorta" appearance (Figure 4).



FIGURE 3: Axial contrast-enhanced computed tomography image of the abdomen demonstrating bilateral renal enlargement with surrounding perinephric inflammatory stranding and fluid, consistent with the "hairy kidney" appearance. Bilateral hydronephrosis and urothelial thickening/enhancement are also demonstrated



FIGURE 4: Axial computed tomography demonstrating cardiomegaly and a small pericardial effusion

With imaging highly suggestive of a systemic histiocytosis, a targeted tissue biopsy was required for definitive diagnosis and molecular profiling. A left renal and perinephric biopsy performed in December 2024 initially showed only chronic inflammation; however, a histopathologic re-evaluation of the specimen

revealed a small fragment of soft tissue containing scattered histiocytes, some of which exhibited foamy cytoplasm.

Subsequently, in February 2025, molecular genetic testing documented the presence of a *BRAF*^{V600E} mutation. In conjunction with multimodal imaging, these results supported the diagnosis of ECD.

Following the confirmation of the *BRAF*^{V600E} mutation, the patient was referred to a medical oncologist for targeted systemic therapy with vemurafenib, a *BRAF* kinase inhibitor.

Discussion

ECD is a rare, multisystemic histiocytic neoplasm driven primarily by somatic mutations in the mitogen-activated protein kinase pathway, most notably *BRAF*^{V600E}. Because of its broad clinical presentation, ECD frequently poses a significant diagnostic challenge, often masquerading as rheumatologic, neurological, or primary renal disorders prior to definitive diagnosis. While classical presentations involve symmetrical long-bone osteosclerosis and diabetes insipidus, our patient's initial presentation with progressive proximal weakness, hyperreflexia, and fasciculations closely mimicked a motor neuron disease or inflammatory myopathy. Establishing the diagnosis required multimodality imaging, careful histological re-evaluation, and molecular testing. Below, we analyze the patient's multisystem organ involvement in the context of established ECD literature.

Bone and skeletal involvement

Skeletal involvement, predominantly presenting as osteosclerosis, occurs in greater than 90% of patients with ECD. Sclerotic lesions most commonly impact long bones of the lower extremities bilaterally, particularly the distal femurs and proximal tibias around the knees. Although the presentation of osteosclerosis is frequently asymptomatic, patients may present with persistent dull leg pain [3]. In an observational study with 60 ECD patients, bilateral involvement was identified in the femur (88%), tibia (88%), humeri (42%), and radius (42%) [4]. ECD characteristically occurs in the diaphyseal regions of the long bones within the appendicular skeleton, while the epiphyses and axial skeleton are unaffected. Though rare, cases with lytic and sclerotic lesions have been described. While radiographs, CT, and MRI can detect cortical thickening and sclerosis, 99mTc-MDP bone scintigraphy and fluorodeoxyglucose positron emission tomography-computed tomography (FDG PET-CT) remain the most sensitive modalities demonstrating intense, symmetrical, and bilateral radiotracer uptake. It is important to not only rely on osseous involvement as ECD can mimic several conditions such as osteomyelitis, Paget's disease, lymphoma, sarcoidosis, bony metastases, and lipid storage diseases such as Gaucher's disease and Niemann-Pick disease [5]. Clinicians must look beyond skeletal imaging alone and correlate these findings with systemic features and histopathology.

Our patient presented with classic radiotracer accumulation on bone scintigraphy with symmetrical, increased MDP uptake within the distal femurs, proximal tibias, distal tibias, and ankles. There were also lower-grade, symmetrical increased MDP uptake within the bilateral radii and distal humeri and mild MDP uptake in the skull. Although Langerhans cell histiocytosis was initially considered in the differential diagnosis, the presentation of symmetric, diffuse appendicular osteosclerosis was most concerning for ECD.

Central nervous system involvement

Central nervous system involvement occurs in 25-50% of patients with ECD and acts as an independent predictor of poor prognosis, accounting for nearly one-third of disease-related deaths. Clinical manifestations vary depending on lesion location and may include exophthalmos, gaze disturbances, diabetes insipidus, cerebellar ataxia, seizures, peripheral neuropathy, and focal mass lesion-related radiculopathy. In a systematic review of 448 patients, diabetes insipidus was the most common neurological/endocrinological manifestation, associated with the thickening of the pituitary stalk [6]. Patients do not always display clinical neurological symptoms despite radiological findings. In a cohort study of 62 patients, 11 patients remained asymptomatic despite radiological evidence of central nervous system involvement. The most frequent neuroradiologic abnormalities included atrophy, cerebral masses, retro-orbital masses, and dural thickening [4].

Our patient experienced a five-year neurological prodrome characterized by progressive proximal lower extremity weakness, muscle fatigability, hyperreflexia, and focal fasciculations. Clinical concern was raised for a primary neuromuscular disorder or motor neuron disease. Over time, our patient's motor deficit progressed to a loss of fine motor dexterity, mild dysidiadochokinesia, and an intention tremor in the left hand. Neuroimaging revealed bilateral intraconal retro-orbital masses partially encasing and displacing the optic nerves. Despite retro-orbital and infundibular involvement, our patient did not present with endocrinopathies such as diabetes insipidus, demonstrating the clinical heterogeneity of central nervous system manifestations in ECD.

Retroperitoneal involvement

Although the majority of ECD cases with retroperitoneal involvement are asymptomatic, clinical manifestations can include symptoms such as dysuria, abdominal pain, and large palpable kidneys which may be detected during physical examination [5]. Retroperitoneal involvement in ECD involves perinephric infiltration, resulting in the characteristic "hairy kidney" appearance visible on contrast-enhanced CT or MRI scans. Hairy kidneys are more frequently found in patients with the *BRAF*^{V600E} mutation occurring in roughly 65-75% of overall patients [7].

The infiltration of histiocytes extends past the perirenal space to involve the renal pelvis and upper ureteral segments. In ECD, fibrosis encircles the aorta, whereas in idiopathic retroperitoneal fibrosis, the posterior wall of the aorta is rarely affected, and instead, the distal ureters are involved [2,5]. Perirenal infiltration may progress to the renal sinuses potentially causing a post-renal obstruction. Continued progression may result in fibrosis and bilateral ureteric obstruction leading to hydronephrosis and compromised renal function. Fibrosis most commonly affects the middle and distal ureteral segments. The involvement of the adrenal gland is frequently noted resulting in adrenal insufficiency. Although unilateral adrenal involvement has been documented, the condition typically affects both sides [5].

In the presented case, the patient displayed classic, extensive retroperitoneal pathology. Contrast-enhanced CT revealed bilateral renal enlargement with prominent perinephric stranding ("hairy kidneys"), perirenal fascial thickening, and bilateral adrenal gland enlargement with surrounding inflammatory changes. Severe encasement of the proximal ureters resulted in bilateral hydronephrosis. Furthermore, the patient exhibited circumferential thickening and periaortic inflammatory changes extending into the iliac arteries, reproducing the characteristic periaortic encasement of ECD.

Endocrine involvement

The main endocrine involvement detected in individuals with ECD is diabetes insipidus, found in 25% of patients. Although effective therapies can reduce radiological pituitary infiltration, diabetes insipidus is typically permanent once it develops. Furthermore, comprehensive anterior pituitary testing reveals functional abnormalities in the vast majority of cases (91% of evaluated patients). These most frequently include growth hormone deficiency (79%) and hyperprolactinemia (44%), alongside less common deficits involving follicle-stimulating and luteinizing hormones (22%), thyrotropin (10%), and adrenocorticotrophic hormone (3%) [2].

The presented patient's evaluation mirrored these established systemic risks. Clinically, the patient reported significant polydipsia and polyuria (consuming up to 15 liters of fluid daily and voiding 10-15 times per day), prompting confirmation via an arginine copeptin stimulation test. Biochemical testing revealed elevated gonadotropins (luteinizing hormone 20.8 IU/L and follicle-stimulating hormone 25 IU/L), which correlated with intense bilateral testicular uptake on FDG PET-CT (maximum standardized uptake value 16.2 on the right and 11.9 on the left), confirming primary hypogonadism secondary to direct histiocytic gonadal infiltration rather than isolated central axis failure.

Role of multimodality imaging

Imaging is critical in the diagnosis of ECD for detecting systemic organ involvement, identifying accessible targets for biopsy, and assessing the extent of the disease. The typical image findings in ECD include bilateral, symmetric osteosclerosis of the long bones, retroperitoneal hairy kidneys, and cardiac infiltration. Bone scintigraphy reveals uptake of radiotracers in the diaphyseal regions of the long bones, while MRI shows hypointense marrow signal on both T1- and T2-weighted images [8]. Ultrasound and CT of the abdomen in over 50% of cases reveal infiltrative soft tissue masses in bilateral perirenal spaces encasing the kidneys, resulting in the "hairy kidney". ECD also manifests in up to 40% of patients as a focal mural mass of the right atrium, coronary sulcus, or periaortic wall. On CT imaging, the masses appear as soft tissue masses which are isoattenuating in comparison to the myocardium. On MRI, the mass appears hypointense and shows post-contrast enhancement in T1- and T2-weighted imaging. Intracranial lesions present as extra-axial meningeal thickening, intra-axial parenchymal lesions, perivascular infiltration, or infundibular lesions. A thickened pituitary stalk and loss of T1 hyperintense signal in the posterior pituitary gland is a common manifestation when presented with lesions [9]. While the patient's initial clinical presentation was non-specific and uncharacteristic of ECD, multimodality imaging findings ultimately established a differential diagnosis for ECD and prompted tissue biopsy.

Role of histopathological and molecular testing

While multimodal imaging can point strongly towards ECD, histological confirmation remains essential particularly in patients with atypical or isolated symptoms. Tissue biopsies of an affected mass or lesion will be performed as part of the diagnostic workup for ECD. Affected tissue typically contains foamy lipid-laden histiocytes, Touton-type giant cells, and a background of dense fibrosis with signs of chronic inflammation [10] (Figure 5). The *BRAF*^{V600E} mutation is present in 50-60% of cases [11]. ECD histiocytes also express CD68, CD163, and factor XIIIa while testing negative for CD1a and Langerin.

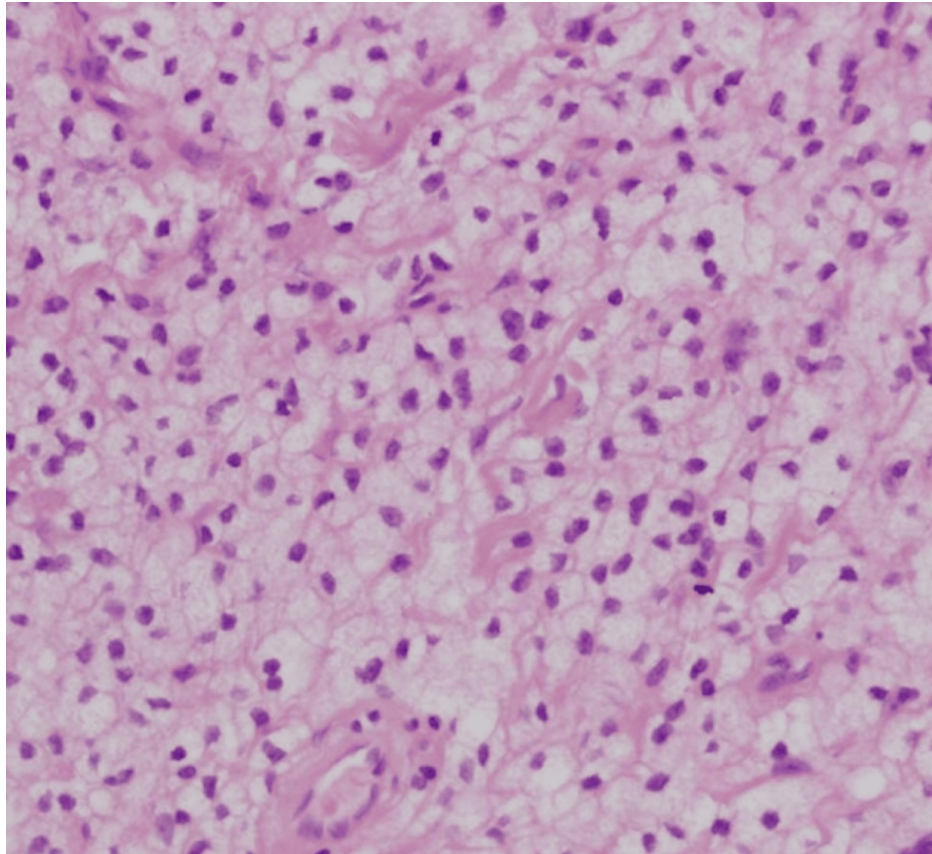


FIGURE 5: Representative histopathology demonstrating multiple characteristic foamy histiocytes infiltrating the tissue

Figure Source: Alexiou and Klastersky [12]; reproduced under the terms of the Creative Commons Attribution-NonCommercial-NoDerivs 3.0 Unported License

Our patient's initial retroperitoneal biopsy demonstrated fibroadipose tissue with mild chronic inflammation due to inadequate sampling. However, given the pathognomonic multimodal imaging findings, expert re-review of the specimen identified sparse foamy histiocytes and the successful confirmation of the *BRAF*^{V600E} mutation. As illustrated by this case, when classic clinical and radiological features are present, a specimen re-review or molecular sequencing is warranted even when the initial biopsy appears non-specific.

Treatment options and current treatment plan

Currently, various therapies are utilized to induce remission or stabilize the progression of ECD. Interferon- α (IFN- α) has historically been shown to improve survival and is widely considered a standard initial treatment, administered at doses from 3 million units three times weekly up to 9 million units three times weekly. Peginterferon alfa-2a serves as a viable, equivalent alternative, given weekly at doses between 135 and 200 μ g. While IFN- α therapy is intended for long-term use, it frequently causes side effects like depression and fatigue in roughly half of patients, whereas polyethylene glycol-conjugated forms tend to offer better tolerability [5].

More recently, the *BRAF* inhibitor vemurafenib has emerged as a key treatment option for ECD, building on its success in improving survival for *BRAF*^{V600E}-mutated metastatic melanoma. The most common adverse effects of vemurafenib involve cutaneous complications, ranging from photosensitivity and keratosis pilaris to squamous cell carcinoma, new primary melanomas, and QT prolongation [2].

Prognosis and management

Following the molecular confirmation of a *BRAF*^{V600E} mutation in the soft tissue specimen, targeted therapy with vemurafenib was selected as the optimal precision treatment plan for this patient. Given his presentation with significant polyuria and polydipsia, an arginine copeptin stimulation test and a full anterior pituitary panel were scheduled through an outpatient endocrinology clinic to confirm arginine vasopressin (AVP) deficiency (diabetes insipidus) and screen for associated endocrinopathies, such as

primary hypogonadism supported by his PET-avid testicular findings. Additionally, his ongoing management includes annual screening for adrenocortical insufficiency and other pituitary hormone deficiencies, alongside a non-urgent bone mineral density assessment to monitor for osteoporosis secondary to his extensive appendicular and axial skeletal involvement.

Conclusions

ECD is a disease that frequently mimics and is misdiagnosed as other conditions, leading to significant delays in diagnosis. As demonstrated by this patient's five-year symptom progression and two-year diagnostic delay, ECD can present atypically long before the appearance of classic symptoms. Our patient's initial presentation with progressive proximal weakness, hyperreflexia, and fasciculations closely mimicked a motor neuron disease or inflammatory myopathy. This case highlights the importance of an expert specimen re-review or molecular sequencing, even when the initial biopsy is non-specific, whenever classic clinical and radiological features are present. Ultimately, combining pathognomonic imaging with persistent molecular evaluation is critical to establishing a diagnosis despite atypical symptom onset, allowing the early initiation of targeted therapy.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Isabelle Ho, Oluwaseun Olabiyi, Keng Yeow Tay

Acquisition, analysis, or interpretation of data: Isabelle Ho, Oluwaseun Olabiyi

Drafting of the manuscript: Isabelle Ho, Oluwaseun Olabiyi

Critical review of the manuscript for important intellectual content: Isabelle Ho, Oluwaseun Olabiyi, Keng Yeow Tay

Supervision: Keng Yeow Tay

Disclosures

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Isabelle Ho and Oluwaseun Olabiyi contributed equally to the work and should be considered co-first authors. Dr. Keng Yeow Tay (MD, FRCPC), Associate Professor of Medical Imaging at Western University and Diagnostic Radiology at London Health Sciences Centre, serves as the senior author supervising this case report.

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