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CASE REPORT

Monocytosis and Clonal Hematopoiesis in Peripheral Blood as Indicators of Myeloid Neoplasm in Erdheim-Chester Disease

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ABSTRACT

Background: **Erdheim-Chester disease (ECD) is a rare non-Langerhans histiocytosis, with ~10% of cases showing myeloid neoplasms (MNs).**
Methods: **A 70-year-old woman with cardiac arrhythmia and right atrial mass was diagnosed with ECD by biopsy. Complete blood count showed persistent monocytosis. Targeted next-generation sequencing (NGS) of peripheral blood (PB) for evaluating clonal hematopoiesis (CH) was performed.**
Results: **The NGS revealed *ASXL1* Asn893Cysfs*12 (variant allele frequency [VAF]: 37.2%), *TET2* His1031 Glnfs*11 (VAF: 49.0%) and *TET2* Tyr1255* (VAF: 47.5%). Bone marrow examination confirmed concomitant chronic myelomonocytic leukemia.**
Conclusions: **NGS using PB needs to be considered in ECD for the detection of CH and for the diagnosis of coexisting MNs.**
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Supplementary Data

Supplementary Table. Variants detected by targeted next-generation sequencing of peripheral blood in a patient with ECD.

Gene	Locus	Transcript	Exon	Variant ID	Coding	Amino acid change	VAF	Variant effect
ASXL1	chr20:31,023,192-31,023,201	NM_015338.5	12	COSM131571	c.2677_2686delAACAGTTCTT	p.Asn893Cysfs*12	37.2%	Frame-shift
TET2	chr4:106,158,192-106,158,194	NM_001127208.2	3	-	c.3093_3095delTCTinsA	p.His1031Glnfs*11	49.0%	Frame-shift
TET2	chr4:106,164,896	NM_001127208.2	6	COSM211719	c.3764dupA	p.Tyr1255*	47.5%	Non-sense

VAF - variant allele frequency, ECD – Erdheim-Chester disease.