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HEMATOLOGY: BRIEF REPORT

Acquisition of monosomy 7 and a *RUNX1* mutation in Pearson syndrome

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Abstract

Pearson syndrome (PS) is a very rare and often fatal multisystem disease caused by deletions in mitochondrial DNA that result in sideroblastic anemia, vacuolization of marrow precursors, and pancreatic dysfunction. Spontaneous recovery from anemia is often observed within several years of diagnosis. We present the case of a 4-month-old male diagnosed with PS who experienced prolonged severe pancytopenia preceding the emergence of monosomy 7. Whole-exome sequencing identified two somatic mutations, including *RUNX1* p.S100F that was previously reported as associated with myeloid malignancies. The molecular defects associated with PS may have the potential to progress to advanced myelodysplastic syndrome .

KEYWORDS
monosomy 7, pearson syndrome, *RUNX1*

1 | INTRODUCTION

Pearson syndrome (PS) is a multiorgan system disorder characterized by refractory sideroblastic anemia with vacuolization of bone marrow (BM) precursors, lactic acidosis, and exocrine pancreatic dysfunction that result from the deletion of mitochondrial DNA (mtDNA) sequences. Pancreatic dysfunction frequently accompanies PS but it is not critical for the diagnosis.[1,2] The incidence of PS is very low, at approximately one case per million individuals.[3] PS is one of the disorders to be considered in the differential diagnosis of hypocellular BM in young children.[4,5] It is not clear whether PS is associated with malignant transformation; the long-term prognosis of PS is generally poor, as children often succumb to fatal lactic acidosis.[6]

Monosomy 7 is a common cytogenetic abnormality identified in inherited BM failure syndromes (IBMFSs) and pediatric

Abbreviations: BM, bone marrow; HSCT, hematopoietic stem cell transplantation; IBMFS, inherited bone marrow failure syndrome; MDS, myelodysplastic syndrome; mtDNA, mitochondrial DNA; PS, Pearson syndrome; WES, whole-exome sequencing

