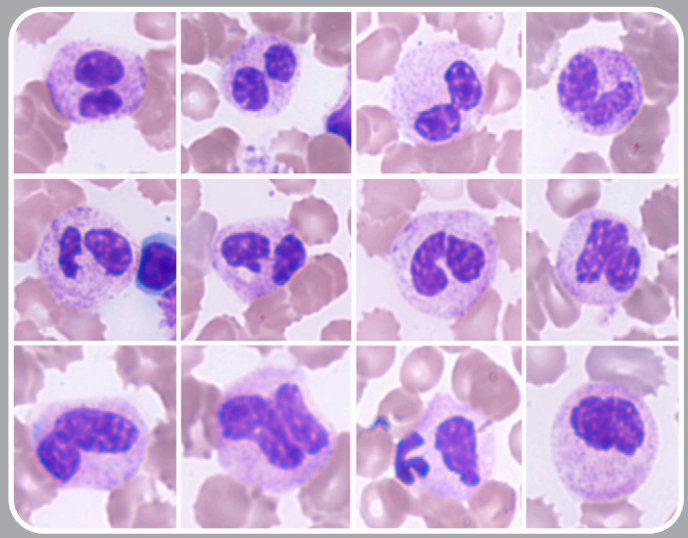
bloodwork
IMAGES IN HEMATOLOGY

# Dysplastic neutrophils in the bone marrow of a Shwachman-Diamond syndrome patient



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A 5-year-old girl presented with growth failure, feeding difficulties, and behavioral problems. She had low levels of serum amylase, lipase, and fat-soluble vitamins (A, D, E, and K) and elevated transaminases, favoring pancreatic exocrine dysfunction. On review of complete blood counts, intermittent neutropenia of varying severity (ranging from 0.200 to 1.400 $\times$ $10^9$/L) was evident since the neonatal period. Bone marrow examination showed moderate hypoplasia. Interestingly, the granulocytic lineage alone was dysplastic, with half the neutrophils harboring bilobulated "pseudo-Pelger" nuclei and abnormally clumped chromatin (composite panel; original magnification $\times$1000, May-Grünwald-Giemsa stain). The bone marrow karyotype did not reveal any clonal abnormalities, but mutations of the *SBDS* gene were observed in one exon (#2) and both introns (#2 [183-184TA>CT;258+2T>C]), resulting in a composite heterozygote. No signs of transformation appeared at 2-year follow-up.

Shwachman-Diamond syndrome is characterized by exocrine pancreatic insufficiency, bone marrow dysfunction, and skeletal abnormalities, and is typically diagnosed in early childhood. Ninety percent of patients have a biallelic mutation in the *SBDS* gene (chromosome 7(q11)). Neutropenia is the most common hematologic abnormality. The bone marrow is often hypocellular and usually contains mild dysplastic changes in the erythroid, myeloid, and megakaryocytic series, even in the absence of clonal cytogenetic anomalies. Prominent multilineage dysplasia, which may correlate with malignant transformation, is less commonly observed.



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