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Neutrophils and monocytes with increased azurophilic granules resembling toxic changes in mucopolysaccharidosis type VI

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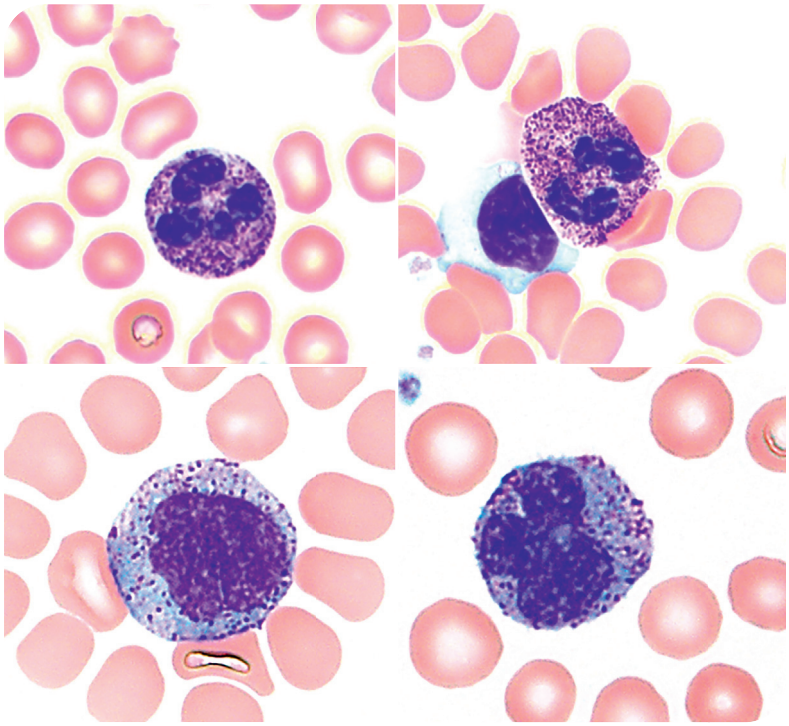

A 2-year-old female patient presented with a history of macrocephaly and short stature. On examination, it was found that she had camptodactyly and stiffness in the joints of her elbows and knees. A skeletal survey showed a J-shaped sella region and rings of dysostosis multiplex, suggestive of a metabolic genetic disorder. Peripheral blood smear review showed normal numbers of granulocytes (top panel, ×100 objective, and total magnification ×1000) and monocytes (bottom panel, ×100 objective, and total magnification ×1000) with increased coarse, azurophilic granules resembling toxic granulation. Additional testing showed low arylsulfatase B (ARSB) activity in white cells and elevated glucosaminoglycans (GAGs) in urine, suggestive of mucopolysaccharidosis type VI (MPS VI). Genetic testing revealed 2 pathogenic variants, c.1449A>T (p.Glu483Asp) and c.1208C>G (p.Ser403*) in the *ARSB* gene on opposite chromosomes, confirming the diagnosis of mucopolysaccharidosis type VI (MPS VI), also known as Maroteaux-Lamy syndrome.

MPS VI is a rare, autosomal recessive metabolic disease caused by mutations in the *ARSB* gene, resulting in the deficiency of the lysosomal enzyme ARSB, which is part of the breakdown process of GAGs in lysosomes. Onset and clinical manifestations can vary greatly. Symptoms include decreased growth velocity, coarse facial features, skeletal deformities, frequent upper airway infections, hearing loss, joint stiffness, cardiac valve abnormalities, spinal cord compression, and enlarged liver and spleen. This case highlights the association of toxic granulation-like changes in MPS VI, affecting all granulocytes and monocytes.

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