

Neutropenia in Infants: A Comprehensive Narrative Review

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Abstract

Neutropenia in infants represents a heterogeneous group of disorders characterized by a reduced absolute neutrophil count (ANC), predisposing affected individuals to recurrent and potentially life-threatening bacterial and fungal infections. This narrative review provides a comprehensive synthesis of the current evidence regarding the epidemiology, etiology, classification, pathophysiology, clinical manifestations, diagnostic approach, differential diagnosis, management, prognosis, and future perspectives of neutropenia in the neonatal and infant population. Neutropenia may arise from transient physiological processes, immune-mediated mechanisms (alloimmune and autoimmune), congenital bone marrow failure syndromes (including severe congenital neutropenia, cyclic neutropenia, and Shwachman-Diamond syndrome), infections, drug-induced causes, or nutritional deficiencies. The diagnostic approach requires systematic evaluation integrating clinical history, serial complete blood counts, peripheral blood smear, antineutrophil antibody testing, bone marrow examination, and increasingly, next-generation sequencing for germline and somatic genetic variants. Management is tailored to the underlying etiology and severity, with recombinant human granulocyte colony-stimulating factor (rhG-CSF) serving as the cornerstone of therapy for severe and symptomatic forms. Prognosis varies markedly by etiology; while autoimmune neutropenia of infancy typically resolves spontaneously within 2–4 years, severe congenital neutropenia

carries a substantial risk of leukemic transformation. Emerging therapies, including targeted gene editing approaches, hold promise for definitive treatment of monogenic forms. This review consolidates contemporary knowledge to guide clinicians in the recognition, evaluation, and evidence-based management of neutropenia in infants.

Keywords: Neutropenia; Infant; Newborn; Severe Congenital Neutropenia; Autoimmune Neutropenia; Alloimmune Neutropenia; G-CSF; ELANE; Bone Marrow Failure