

AUTOINFLAMMATORY DISEASES IN CHILDREN

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Background. Systemic autoinflammatory diseases are a heterogeneous group of disorders characterized by recurrent or persistent sterile inflammation caused mainly by dysregulation of the innate immune system. In children, symptoms may include fever, rash, serositis, arthritis, abdominal pain, mucosal lesions, ocular inflammation, bone pain, or systemic inflammatory complications. Because their manifestations often overlap with infection, malignancy, immunodeficiency, and autoimmune disease, diagnostic delay remains a major clinical problem.

Aim. To summarize the key clinical features, diagnostic principles, and therapeutic approaches to pediatric autoinflammatory diseases, with emphasis on their relevance in Ukraine.

Main theses. Autoinflammatory diseases should be suspected in children with recurrent, stereotyped inflammatory episodes, elevated inflammatory markers during flares, poor response to antibiotics, and absence of confirmed infectious cause. The diagnostic process should be phenotype-driven and include assessment of age at onset, attack duration, periodicity, triggers, family history, ethnicity, organ involvement, and laboratory dynamics between attacks. The pediatric spectrum includes PFAPA, familial Mediterranean fever, CAPS, TRAPS, mevalonate kinase deficiency/HIDS, chronic nonbacterial osteomyelitis, interferonopathies, and other rare monogenic or multifactorial conditions. Genetic testing is important, especially in early-onset, severe or atypical disease, but results must be interpreted in the context of the clinical phenotype. Treatment depends on the underlying inflammatory pathway. Colchicine, IL-1 blockade, JAK-inhibitors, anti-TNF, others biological drugs and steroids had significantly improved outcomes in several severe systemic autoinflammatory diseases.

Ukrainian context. According to the presented Ukrainian data, 38 patients with serious systemic autoinflammatory diseases and more than 150 patients with PFAPA have been identified. Diagnostic delay is usually 3-7 years and may reach 20-30 years. Ukraine has state-funded access to IL-1 inhibitors, national guidelines, international cooperation, and active patient advocacy. However, low awareness, limited local genetic and functional testing, absence of a dedicated national center and the impact of the Russian invasion remain important barriers.

Conclusions. Early recognition of autoinflammatory diseases in children requires clinical awareness, structured diagnostic reasoning, and access to targeted therapy. In Ukraine, priority should be given to education, referral pathways, diagnostic capacity, registry development, and uninterrupted biologic treatment.

Keywords: *monogenic autoinflammatory diseases, children, recurrent fever, IL-1 blockade, childrens*