

Caring for children with hyper-IgE syndrome

One of my pediatric patients was just diagnosed with hyperimmunoglobulin E syndrome (HIES). What nursing considerations are associated with this diagnosis?—K.D., N.H.

Kathleen Peterson, PhD, RN, PCPNP-BC, and Paula Barbel, PhD, RN, PNP, reply: HIES is a rare primary immunodeficiency disorder, affecting an estimated 1:100,000 people with an equal distribution between males and females.¹ HIES is a chronic syndrome characterized by recurrent primarily bacterial skin and pulmonary infections, eczematous dermatitis, and elevated serum immunoglobulin E (IgE) levels.²

Genetic studies and molecular testing have shown two main types of HIES, autosomal dominant (AD-HIES) and autosomal recessive (AR-HIES), among several other mutations.³ Both main forms of HIES are characterized by extremely elevated IgE serum levels. Normal serum IgE levels range from 3 to 423 IU/mL.⁴ Serum IgE levels may range from 1,000 to over 50,000 IU/mL in patients with HIES, though a lower limit of 2,000 IU/mL is generally used for diagnosis.²

Skin manifestations are the most obvious signs of HIES. Patients may have dermatitis and folliculitis, abscesses, and chronic skin infections caused by *Candida albicans* and/or *Staphylococcus aureus*.² Parenchymal lung disease, pneumatoceles, recurrent pneumonias, and pulmonary abscesses are possible pulmonary manifestations of HIES.²

AD-HIES is characterized by eczema, recurrent staphylococcal skin and lung infections, pneumatocele

formation, mucocutaneous candidiasis, eosinophilia, and various nonimmune manifestations.⁵ More than half of patients with AD-HIES are born with or develop a rash in the newborn period (the first 28 days). This newborn rash consists of eosinophilic pustules.⁵

Abnormal craniofacial features (facial asymmetry, prominent forehead, broad nasal bridge and base, and deep-set eyes, wide outer canthal distances), craniosynostosis, retained childhood dentition as well as noneruption of permanent teeth, and a high arched palate are characteristic.^{1,2} Musculoskeletal abnormalities, including hyperextensible joints, scoliosis, and osteoporosis leading to minimal trauma fractures may be seen.¹ Vascular abnormalities include aneurysms, hypertension, and coronary artery tortuosity/dilatation.¹

Although patients with AR-HIES don't have a newborn rash, eczematous dermatitis develops in infancy in nearly all patients.⁵ Most patients with AR-HIES develop severe allergies, including anaphylaxis, to food and environmental antigens as well as asthma.⁵ They're also highly prone to malignancy, with 10% to 36% of patients developing cancers in late childhood to late adulthood.⁵ Vascular structural anomalies may be seen in both types of HIES but are more common in AR-HIES. These vascular anomalies may be venous or arterial, congenital or acquired, and include aneurysms and pseudoaneurysms.²

Various treatments for HIES are emerging. Recombinant human interferon-gamma, omalizumab (anti-IgE), cyclosporine, high-dose I.V. immune globulin, and hematopoietic cell transplantation have all

been used as therapies with varying results.^{1,2,5-7} However, no large-scale clinical trials have been conducted to establish an evidence-based treatment protocol.

The first line of defense for patients with HIES is good general hygiene, including good dental hygiene. For children with HIES, regular visits to the dentist are imperative to help prevent infections and malocclusion.

Treatment also includes the prevention of skin and pulmonary infections with prophylactic antimicrobials.^{2,7} Allergies and asthma, more common with AR-HIES, require conventional treatment according to evidence-based guidelines.⁷ Overall, data on the systemic treatment of HIES are quite limited, making the prevention of skin and pulmonary infections very important. Nagaraj et al. also report prescribing emollients for dry skin, bleach baths to prevent staphylococcal infection, and antihistamines to control pruritus.³

Parents of children with this disorder need to understand that it's a chronic illness and they can never let down their guard. Teach parents to be vigilant to recognize and manage skin infections because these need prompt treatment. Pulmonary complications must be identified early to treat them effectively.

Antimicrobials are prescribed as needed for infections. Teach patients and family members about using antibiotics properly. Assisting with the diagnosis of HIES is an important nursing consideration. Because this disease is so rare, it may not be considered as a differential diagnosis for children with recurrent skin and pulmonary infections. Nurses can suggest that

babies who have severe eczema and pruritus be referred for a dermatology consult earlier rather than later. Suggesting IgE serum evaluation and molecular testing with such children may assist in diagnosis.

Nurses must be active case managers for these children in their medical home. Parents need a good working relationship with the healthcare team; nurses can facilitate the relationship. Nurses should contact the healthcare provider with abnormal bloodwork results or physical assessment findings that suggest an infection. Nurses are critical to ensuring that children

with HIES receive safe, effective, patient-centered, and equitable healthcare. ■

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