Hyper IgE Syndrome

Hyper IgE syndrome, or Job syndrome, is an autosomal dominant disease caused by a mutation in the STAT3 gene. Patients have increased levels of IgE and abnormal neutrophil chemotaxis. On physical exam, patients may present with leonine facies, cold abscesses, retained primary teeth, and an eczematous rash. Patients often require lifelong prophylactic antibiotic treatment.



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Job Syndrome (Autosomal Dominant Hyper IgE Syndrome)
Job-briefcase
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Pathophysiology

Autosomal Dominant
Domino
Hyper IgE syndrome, or Job syndrome, is most often an autosomal dominant disease associated with STAT3 gene mutations. As it is an autosomal dominant disease, all carriers of the disease will present with symptoms and affected parents will pass on the disease to 50% of their children.

STAT3 Mutation
STATue with (3) Tree Mutant
Mutations in the STAT3 gene are responsible for the pathogenesis of Job syndrome. This mutation leads to increased IgE production by B lymphocytes as well as resistance to anti-inflammatory mediators. Additionally, patients have a relative deficiency of Th17 cells.

Increased IgE
Up-arrow (IgE) Electric-goblin
Patients with Job syndrome will have increased levels of IgE on laboratory tests. The underlying STAT3 mutation in these patients leads to unregulated IgE production by B lymphocytes.

Abnormal Chemotaxis
Chemical-taxi
The defect in STAT3 seen in these patients also results in impaired chemotaxis of neutrophils to sites of infection. As a result, these patients have difficulties with staving off infection.

Signs and Symptoms

Leonine Facies
Lion Face
Leonine facies is a descriptive term used for a deeply furrowed ‘lumpy' face with prominent superciliary arches that is seen in Hyper IgE syndrome.

Cold (Non-Inflamed) Staph Aureus Abscesses
Cold Abscess-guy with Staff-of-Oreos
Patients with Job syndrome have difficulty staving off infection. As a result, they often develop staph aureus skin abscesses. These abscesses are typically non-inflamed, or cold, due to impaired neutrophil recruitment.

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Retained Primary Teeth

Retainer Teeth

Patients with Job syndrome typically fail to lose baby teeth. This is due to failed resorption of the roots of primary teeth.

Eczema

Ax-zebra

Patients with Job syndrome typically present with an eczematous rash. Patients are additionally prone to staphylococcal superinfections.

Management

**Prophylactic Antibiotics
**

Purple-axes ABX-guy

Patients with Hyper IgE syndrome typically require lifelong prophylactic antibiotics. Preferred agents include cephalosporins, antifungal agents, and penicillinase-resistant penicillin.

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