

Acquired Rectovaginal Fistula in Job's Syndrome

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A 6 month old female patient admitted to King Hussein Medical Center with 2 weeks history of fecal discharge from lower labia, found to be a result of acquired rectovaginal fistula. Patient had a previous history of reluctant eczematous dermatitis and recurrent skin abscesses. The diagnosis of hyper IgE syndrome was made on basis of recurrent infections and a very high level of IgE. To the best of our knowledge this is the first case of proved hyper IgE syndrome presenting with rectovaginal fistula.

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Hyper IgE syndrome (Job's syndrome) is a rare disorder of unknown etiology characterised by recurrent (usually staphylococcal) abscess formation that are often 'cold', lung abscess which results in pneumatoceles as well as skeletal anomalies, coarse facies, eosinophilia and very high serum level of IgE¹. The β -lymphocytes from these patients spontaneously produce large amounts of IgE in vitro. Several kindreds involving both males and females, and affected mothers and fathers with affected children have been reported, suggesting that in some instances the disease is autosomal dominant. Sporadic cases also occur. The immunological defect is not yet fully understood²⁻⁴. We report a 6 month old female infant who presented with acquired rectovaginal fistula proved later to be Job's syndrome.

THE CASE

Patient was born at term after an uneventful pregnancy and weighed 3.2 kg. As early as the third week of age, she developed eczematous dermatitis over her scalp, face and neck and recurrent subcutaneous abscesses of the face, buttocks and posterior aspects of thighs. She was admitted twice to the local hospital for abscesses drainage and parenteral antibiotics. At the age of 2 months, she was admitted to King Hussein Medical Centre with high fever and severe eczematous dermatitis (Fig 1). In addition she had fecal discharge through a fistula in lower labia (Fig 2). Her parents are not consanguineous, and her 4 brothers and one sister are healthy. Laboratory investigations are presented in Table 1.

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Figure 1.

Figure 2.

Table 1. **Results of laboratory investigations**

<i>Investigation</i>	<i>Results</i>
PCV	27 mg%
WBC	22000/cubic mm
Neutrophils	11000/cubic mm
Eosinophiles	800/cubic mm
IgG	Normal for age
IgM	Normal for age
IgA	Normal for age
IgE	2500 Iu/L
CH50	Normal for age
B cells number	Normal for age
T cell number	Normal for age
NBT	Normal for age
Burst Test	Normal for age
Phago Test	Normal for age
HIV (ELISA)	Negative

A diagnosis of hyperimmunoglobulinemia E-recurrent infection syndrome was made based on a history of recurrent Staph.aureus infection and extremely elevated serum IgE.

DISCUSSION

The hyper-IgE syndrome with recurrent infection is a rare immunodeficiency disorder characterized by recurrent staphylococcal skin and pulmonary abscess formation and extremely elevated levels of IgE in serum. Associated facial and skeletal features have been recognized, but their frequency is unknown and the genetic basis of the hyper IgE syndrome is poorly understood²⁻⁵. Abnormalities in dentition, bone and connective tissues were found to be as common as immunological abnormalities in Job's syndrome. These abnormalities include failure to shed primary teeth, recurrent fractures, hyper extensible joints and scoliosis. Such abnormalities were more

common in older age groups^{6,7}. None of these abnormalities were documented in our case.

Even though some complications reported in Job's syndrome like liver tumors and cerebral vascular accident were not related to immunological abnormalities but the majority of other complications are due to compromised immune status⁸.

Pasic et al⁶ reported disseminated tuberculous infection in a girl with Job's syndrome. Misago et al and Yates et al^{3,7} reported separately necrotizing fasciitis and candida endocarditis in two different patients with Job's syndrome. Santambrogio et al⁹ and Sehrt et al⁴ reported other complications like fungal lung abscesses, multiple papillomas and ulcers of the mucus membranes. Chen et al² and Hwang et al¹⁰ reported on two different occasions colonic perforation in an 18 year and 8 year female patients with Job's syndrome respectively. Rectovaginal fistula has not been reported yet in this syndrome, but has been reported in other immunodeficiency diseases like AIDS¹¹.

CONCLUSION

To the best of our knowledge this is the first case of proved hyper IgE syndrome presented with rectovaginal fistula in which history of perianal abscess was never documented. An awareness of the diverse features of the hyper IgE syndrome will facilitate the diagnosis of sporadic cases, make genetic counseling possible and improve patient care.

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