

Cutaneous Manifestations of Hyper-IgE Syndrome in Twins: a case report from Saudi Arabia

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The hyper-IgE syndrome with recurrent infections is a rare immunodeficiency disorder characterized by very high levels of IgE in serum. It is inherited in most of the cases in a dominant fashion meaning that it can run in families with a high frequency. About 250 cases have been published in literature so far.

Case Report

Twin brothers aged 9-years were seen over a period of 5-years and were diagnosed and treated as atopic dermatitis. They came to our attention due to repeated exacerbations of dermatitis and multiple skin abscesses. Physical examination showed that both the brothers had coarse facial appearance with prominent brow and deep-set eyes (Figure 1). There were papulopustular lesions all over the body especially on the face and scalp (Figure 2). These bouts of infections had no seasonal variation and was present to some degree at all times. The elder twin always had more extensive eczema with secondary infections, as such his physical growth was less as compared to his twin brother as shown in the (Figure 1). Recurrent small and large skin abscesses were also observed during bouts of exacerbation (Figure 2). Both the brothers had repeated attacks of conjunctivitis with purulent discharge resulting in dry scabs along the eyelid margins which is clearly visible in (Figures 1 and 2). In addition to that they suffered from recurrent attacks of uni or bilateral ear infections, cultures taken from the ear were positive for *Staphylococcus aureus*, *Streptococcus pneumoniae* and on one occasion for *Pseudomonas aeruginosa*. They also suffered from frequent attacks of candidiasis mostly in the groin responding dramatically to local anti fungal ointments.

The mother gave the history that her sons were suffering from repeated episodes of skin infections since

infanthood, there was also history of cough, fever and breath-

lessness due to chest infections and one of them was admit-

ted in the past in a children's hospital for pneumonia .

The recovery was however uneventful and the patient left hospital after 10 days. Father of the twins was atopic in his childhood, while the rest of the family were normal.

Routine blood counts revealed that both the brothers had normal white cell counts while the differential counts showed persistent eosinophilia with eosinophil count of $1.17 \times 10^9/L$ and $1.09 \times 10^9/L$ respectively (Lab normal $0.04 \times 10^9/L - 0.4 \times 10^9/L$).

Blood chemistry was essentially normal. The serum IgE level was more than 2500IU/ml and 2751IU/ml respectively (Lab normal 0.3- 215 IU/ml) with normal levels of other immunoglobulin classes.

Radiological findings revealed no abnormality in the chest and spine. The culture of the pus from the gluteal abscess revealed staphylococcus aureus. H&E stained histological sections of 4mm punch biopsy from both the cases revealed spongiosis and perivascular dermatitis with a preponderance of eosinophils (Figure 3).

Discussion

Hyper IgE syndrome also known as Jobs syndrome is a rare genetic disease, characterized with frequent skin and chest infections, bone abnormalities and markedly elevated levels of immunoglobulin E (IgE). No specific race is affected, it occurs in diverse ethnic backgrounds and is usually seen in infancy but the diagnosis is often missed until childhood or even adulthood. The Waorani Indians of Eastern Equador have the highest blood levels of immunoglobulin E that have been reported in a human population so far.¹ These twins had the classical triad of hyper IgE syndrome, i.e., recurrent skin infections and abscesses, pneumonia, and elevated IgE levels, similar clinical findings with high levels of immunoglobulin IgE causing recurrent pyogenic infections, chronic dermatitis and osteopenia have been reported in the past.²⁻⁴

Notably the boys had coarse facial appearance with prominent brow and deep-set eyes, similar clinical observations of facial and skeletal features have been recognized and reported.⁴ The elder twin had limitation of extension of the right elbow following supracondylar fracture of the right

humerus two years back; multiple fractures of the long bones, ribs or pelvis are common in this disorder^{2,4}, however both the brothers did not have scoliosis which is also a common feature in most teenaged patients.

The histologic examination of the twins in this study revealed some areas of eosinophilic infiltration in addition to dermatitis which is one of the characteristic feature of this condition, similar histological features have been reported in the past.²

The diagnosis of hyper- IgE syndrome is usually made on an average of 18 months after the onset of initial papulopustular eruption. It is important to recognize this disease early so that appropriate treatment is started which is critical for the optimum outcome of the disease.² There is no definitive therapy for this condition and the aim of the treatment is to control the infection, however several regimes have been tried and one of them is life long administration of therapeutic doses of penicillinase-resistant penicillin with the addition of other antibiotics or anti fungal agents as required for specific infections. Anti viral and IV gamma globulin may be given when required. The response to monthly intravenous human immunoglobulin has been reported to be good.^{5,6}

This case of twins serves as a reminder that hyper-IgE is a multisystem disorder that affects the skeleton, connective tissue and the immune system and this syndrome must be therefore borne in mind whenever children with repeated skin and chest infections are encountered.

References

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