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Patterns of Immunodeficiency in a Tertiary Care Hospital: The Experience at King Faisal Specialist Hospital from 1981-1988

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ABSTRACT. Primary immunodeficiency disorders are rather common in the Kingdom. One hundred and eight cases in patients and their close relatives have been diagnosed at KFSH & RC between 1981-1988. It is believed that this number represents but a fraction of the total cases, most of whom die undiagnosed. Consanguinity may be a major factor in the high incidence of primary immunodeficiency. A high index of suspicion by physicians should result in early diagnosis and referral to a center where advanced methods for diagnosis and treatment in the Kingdom are available.

KEY WORDS : Immunologic deficiency, Antibody deficiency, B lymphocytes, T lymphocytes, Hereditary diseases.

Introduction

The immunodeficiency disorders are a diverse group of diseases, which result from defective functions of one or more elements of the immune system. As a result, the host suffers from recurrent, unusual and very often, severe infections. The primary immunodeficiency syndromes are, generally, inherited and congenital, hence the majority of affected patients are infants and young children.

The incidence of primary immunodeficiency disorders is difficult to estimate and may vary from country to country and from one ethnic group to another[1-5]. However, in the general population, one may expect one affected individual, mostly male, per ten thousand (1:10,000), excluding asymptomatic IgA deficiency[1-3]. In hospitalized patients, the prevalence of immune deficiency is around 1%. Selective IgA deficiency varies from 0.03 to 0.97% in general population. Fifty-seven (57%) to seventy

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(70%) percent of all primary immunodeficiency cases occur in male infants[5]. Sixty percent or more of the cases are diagnosed below the age of three years.

There are no clinical studies to assess the true incidence of primary immunodeficiency diseases in Saudi Arabia. However, cases referred to a major Tertiary Care Hospital over the years may reflect a pattern, and, hence, the frequency of occurrence of these rare diseases.

This paper represents the author's experience at King Faisal Specialist Hospital and Research Centre, a Tertiary Care Hospital, from 1981 to 1988.

Results and Discussion

A total of 108 cases of primary immunodeficiency have been diagnosed in the last 7 years. Table 1 summarises these cases in the order of frequency. Eighty patients of the total were males representing 74% of all the patients, and the rest were females (28) representing 26%. Their ages ranged from one day to 19 years. Consanguinity among families of patients was 65% compared to about 20% in Saudi population.

Of particular interest is the rarity of selective IgA deficiency among our population compared to figures quoted in the literature[6-8]. Among all immune deficient patients, only one case of IgA deficiency was diagnosed. This rarity of IgA deficiency can be related to the fact that most patients with selective IgA deficiency are asymptomatic. However, since the author screened a large number of healthy children and patients with respiratory allergy and found no case of selective IgA deficiency, it is believed that the rarity of IgA deficiency in Saudi patients is related to genetic and ethnic factors rather than lack of diagnosis. The other observation, is the relative large number of patients diagnosed with hyper IgE syndrome, 17 patients in 7 years. Four cases of a newly recognized immunodeficiency have been seen. These patients have many features of Chediak-Higashi Syndrome except that they lack the giant granules in their leukocytes. Most patients are males, and the majority die before they reach their third birthday, unless treatment is started early.

The clinical presentation of these patients included a variety of symptoms, depending on the underlying immune defect. However, the hallmark of this was recurrent and chronic deep seated serious infections, which were at least 2-3 times more than their siblings or peers for the same age. These infections included recurrent pneumonias, chronic otitis media, oral thrush, cellulitis and skin abscesses, meningitis and sinusitis. Most fatalities were related to severe sepsis. Chronic diarrhoea and failure to thrive were common features.

The family history of these patients was very impressive with frequent consanguinity and early infant deaths with infections.

Although the number of patients diagnosed in our institution in the last 7 years is considered large compared to centers in USA and Europe, it is believed that the true number with primary immunodeficiency in Saudi Arabia is much more. It is believed that for each patient diagnosed at least three cases die undiagnosed. This is due to the

fact that many physicians are unfamiliar with the different presentations of the various immunodeficiency syndromes.

Since many of these patients can be salvaged if diagnosed early and properly treated, awareness among physicians and education of the public is needed if mortality is to be reduced.

TABLE 1. Primary immunodeficiency disorders diagnosed at KFSH & RC, 1981-1988.

Diagnosis	No. of Cases	% of Total
Antibody deficiency	23	21.3
Hyper IgE (Job's) syndrome	17	15.7
Combined immunodeficiency	14	13.0
Polymorphnuclear defects including chronic granulomatous disease	11	10.0
Chediak-Higashi syndrome	10	9.3
Ataxia telangiectasia	7	6.5
Mucocutaneous candidiasis and endocrinopathy	6	5.6
Complement deficiency	5	4.6
Thymic hypoplasia including Di-George syndrome	5	4.6
Wiscott-Aldrich syndrome	4	3.7
Partial albinism and immunodeficiency (a new syndrome)	4	3.7
Hyperkeratosis and recurrent infection (Papillon-Le Fevre syndrome)	2	2.0
Total	108	100%

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أنماط قصور المناعـــة : عرض لخبرة مستشفى الملك فيصل التخصصي

حـــرب هـــرفي
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المستخلص : تعتبر علل قصور المناعة الأولية على قدر من الشيوع في المملكة ، فقد تم تشخيص ١٠٨ حالات من هذه العلة لدى المـرضى وأقـربـائهم بين عامي ١٩٨١ و ١٩٨٨م . ويعتقد أن هذا العدد لا يمثل إلاّ جزءًا من مجموع الحالات إذ يقضي معظم الآخرين نحبهم دون تشخيص المرض . وربما كان زواج الأقارب عاملاً مهمًّا لذيوع مرض قصور المناعة الأولى .

وللتشخيص المبكر لهذه الحالات ، ينبغي تقديمها في التشخيص التفريقي عند معاينة المـرضى ومن ثم تحويل الحالات المشتبه فيها إلى المراكز المتقدمة في المملكة لإجراء سبل التشخيص المتطورة وبدء العلاج .