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CASE REPORT

Common Variable Immunodeficiency (CVID): A Case Report

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ABSTRACT

Common variable immune deficiency disease (CVID) is the most prevalent acquired immune deficiency in a human being after selective immunoglobulin A deficiency. It causes reduction of immunoglobulin levels and specific antibodies production and enhancement of recurrent and chronic infections risk, especially respiratory infections. The CVID patients face increased the risk of granulomatous disease, autoimmune and phenomenon, and malignancy. The disease involves males and females equally. Some studies showed that early diagnosis of CVID disease and conventional treatment of patients with intravenous immunoglobin might have an effective role in decreasing pneumonia and frequency of hospitalization due to infections and its complications. In this study, we report a 16 years old girl with CVID, without a clinical history of determined infection with recurrent sinusitis.

INTRODUCTION

Common variable immune deficiency (CVID) is a primary immunodeficiency disease which is associated with decreasing of antibody response and defect in T cell proliferation and cytokines secretion especially interlukin-2. Therefore, these patients are at high risk of granulomatous disease, autoimmune phenomenon, and malignancies (1). Prevalence of this disease is 1 per 50,000 persons.

The disease involves males and females equally. The probability of hypo-gamma globulinemia in a family of CVID patients is estimated to be 20%. The onset of disease in early age and the close consanguineous relationship of parents of patients indicate an autosomal recessive etiology for this disease. Overall, the disease characterized by hypo-globulinemia and recurrent infections. The respiratory system is one of the common organs that involved in CVID patients and pneumonia is its most common manifestation (2). Utilization of immunoglobulin products (e.g. intravenous immunoglobulin (IVIG)) is an appropriate and selective treatment method in CVID patients, which cause reduction of infections in CVID patients in two recent decades (3,4).

Some studies showed that early diagnosis of CVID disease and conventional treatment of patients with IVIG might have an effective role in decreasing pneumonia and frequency of hospitalization due to infections and its complications. These patients should be followed up more regular and exact to increase life expectancy with high quality with accurate and complete consumption of IVIG (5). In this study, we report a 16 years old girl with CVID.

CASE PRESENTATION

A 16-years old female referred to our center with fever and sputum cough signs/symptoms. Symptoms of the disease had started with sputum coughs in last week despite antibiotic therapy. There was no cure observed, and the patient suffers fever and worsening of symptoms.

The patient was the first child of her parents whom had a close consanguineous relationship and there was a history of recurrent respiratory infection in her family. History of recurrent infection (at least 6 times a year) was expressed during childhood which lasted more than peers and failed to respond to treatment well.

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The child had growth disorder, and her growth characteristic (height, weight, and body mass index (BMI) was comparable with a 10 years old child. There was no sign of puberty in her. There was tenderness of maxillary sinuses in both sides and frontal sinus at face touch. In an examination of the pharynx, there was green postnasal discharge (PND). Lung examination revealed tachypnea with crackles on auscultation. Other examinations findings were normal, and there was no lymphadenopathy. In paraclinical surveys, sinus radiography showed thickening of maxillary and frontal sinuses. Patient treated with broad-spectrum antibiotics as a resistant sinusitis. After three days her fever was disrupted, and after one week she permitted in a good general condition with a prescription. After two months patient presented again with a recurrence of symptoms, which was treated and permitted through a respective recovery (cure). After one month, the patient has admitted to hospital with a recurrence of symptoms again. Due to recurring of symptoms, she was put under further investigations.

Computed tomography (CT) scan of sinuses showed an increase in mucosal thickness and presence of fluid in paranasal sinuses and unilateral tonsillitis. High resonance CT (HRCT) scan of lung confirmed bronchiectasis at inferior lobes of both sides, with the partial collapse of the right medial lobe and mediastinal lymphadenopathy, that was suggestive for cystic fibrosis or allergic reaction. In laboratory tests, high ESR levels and leukocytosis with a shift to the left was reported. Patient's tests results are summarized in Table 1.

According to decrease levels of immunoglobulins and recurring infections, diagnosis of CVID raised for this case and treated by IVIG. After a 10-day period with twice injection per day, the patient was permitted in good general condition and monthly IVIG prescription. During 6-month follow-up recurrence of symptoms or opportunistic infections were not observed.

DISCUSSION

The CVID incidence is 1 case per 25-66 thousand people in different populations. It usually becomes symptomatic in 2nd and 3rd decades of life and after puberty, and 20% of patients are in childhood (6). Our patient was a 16-years old child that had become mature. Based on a study in Iran, the average age onset of symptoms was 11 and 12 years for girls and boys, respectively (7).

Table 1. Summary of patient's test in 3 hospitalization period

Variables	Amount index (%)		Amount
Alkaline phosphatase (Alp)	1731	(CD3%)	91.09
C-reactive protein (CRP)	3+	(CD4%)	33.36
WBC	8041	(CD%)	2.99
PMN	51%	(CD16%)	3.06
C3	154	(CD19%)	56.05
C4	78	IgA (mg/dl)	Undetectable
Ch50	118	IgG (mg/dl)	90
ESR	31	IgM (mg/dl)	216

IgG (n.l: 7–17g/L), IgM (n.l: 0.4–2.8 g/L), IgA (n.l: 0.7–4 g/L)

In family and relatives of presented child, there wasn't any similar case of the disease reported. In a study in Germany between 32 children with CVID, only one patient had a positive family history (8). In a study which published in Iran in 2007 among 69 CVID patients, 10 of them had a positive family history (9). The reason for this difference may be a high prevalence of consanguineous marriage in Iran.

In Iran, most common symptoms are infections. In some cases, the disease indicated with autoimmune disorders or malignancy. In a study in Tehran most common infection in CVID patients was pneumonia and after that sinusitis (9). Agha Mohammadi et al. reported most common infection in these patients were median otitis, respiratory and gastrointestinal systems infections (7). In general, these people are susceptible to Streptococcus Pneumoniae and Hemophilus Influenza. Also, mycoplasma infections have seen in these patients (10).

Pulmonary infection and bronchiectasis were observed in our patient. In various studies delayed diagnosis, recurrent infections and inadequate treatment were known as lung damage cause. However, in some people despite the appropriate and timely diagnosis and treatment, pulmonary disease progress bronchiectasis occurs (7). Respiratory failure due to chronic pulmonary infection is one of the common reason of death in CVID patients. In a study in Iran, morbidity and mortality rate of these patients reported 20% in a 25 years' period (11).

The CVID diagnosis usually delayed due to nonspecific symptoms. Therefore, this diagnosis should consider in any patient with recurrent infection and low serum immunoglobulin levels. Our patient was diagnosed after 6 months since visiting the doctor. In a study in Tehran, the average time between outbreaks of symptoms and the CVID diagnosis was about 4 years (9). In a study in the United Kingdom, the disease was diagnosed in an average two years after the onset of symptoms (12). Studies showed that delayed diagnosis cause incidence of lasting effects (permanent complications) in these patients. Education of physician about primary immune deficient diseases would be effective in reduction of burden (disease load) and delayed diagnosis.

CONCLUSION

Our patient was treated with IVIG and had been asymptomatic during the follow-up period. In some studies, IVIG prescription causes improvement of patient's quality of life. Delayed diagnosis and treatment of CVID patients can lead them to death.

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AUTHORS CONTRIBUTION

All the authors contributed equally to this manuscript.

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