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Case Report
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A case of severe combined immunodeficiency presenting with CMV pneumonia

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Abstract

A five month-old girl was admitted in Ali Asghar Children's Hospital with a history of three months of fever, cough and dyspnea that her symptoms have exacerbated since two weeks before admission.

She was the first child of the family born to consanguineous parents. She was clinically healthy in the past and had gained weight normally and undergone vaccination program with no complication until the age of 5 months. Her CMV infection rapidly progressed and she was admitted in PICU for severe respiratory distress.

Keywords: Combined immunodeficiency, Consanguineous parents, Ali Asghar Children's Hospital

Background

Severe Combined Immunodeficiency (SCID) is a group of combined immunodeficiencies described by profound defect in T Cell differentiation and development which affects both cellular and humoral arms of adaptive immunity, thus, resulting in complete lack of adaptive immunity (1-3).

SCID usually presents in early infancy (4) and is considered a medical emergency (5) due to its high mortality rate in the first one or 2 years of life, so early diagnosis of the disease is of critical importance to provide hematopoietic cell transplantation in the means of successful treatment and cure (6).

One should notice that SCID patients appear clinically healthy at birth and are affected by different bacterial, fungal and viral infections in early infancy (7-8).

Cell mediated immunity is the major defense against many viral infections including CMV infection (9). Inadequate immune response to CMV (acquired in uretro, during birth, or via breast milk), can cause syndromes of CMV infection including pneumonitis, colitis, encephalitis and retinitis (4) with high morbidity and mortality rates (9).

Case Report

A 5-month-old girl was admitted in our hospital with a history of three months of fever, cough and dyspnea that her symptoms have exacerbated since two weeks before admission.

She was the first child of the family born to consanguineous parents. She was born term with a birth weight of 3200grs. She gained weight normally and undergone routine immunization program at birth and her second month of life. She was exclusively breast feed till date.

There was no history of abortion or death in early infancy in the family history. Her umbilical cord was separated at the right time. She had normal neck holding but no social smile.

She had been prescribed cephalexin and azithromycin before being admitted, with no significant response. At the time of admission there was no sign of thymic shadow on CXR and patchy infiltrations and atelectasis were reported

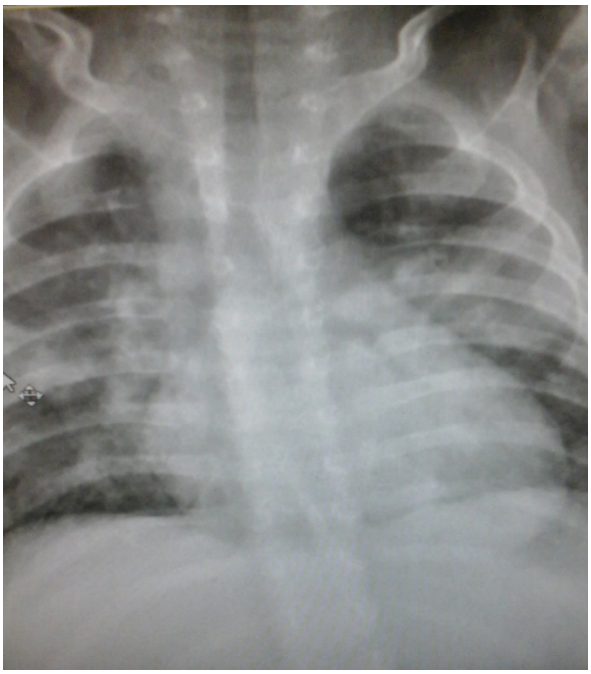

Fig. 2. At the time of admission

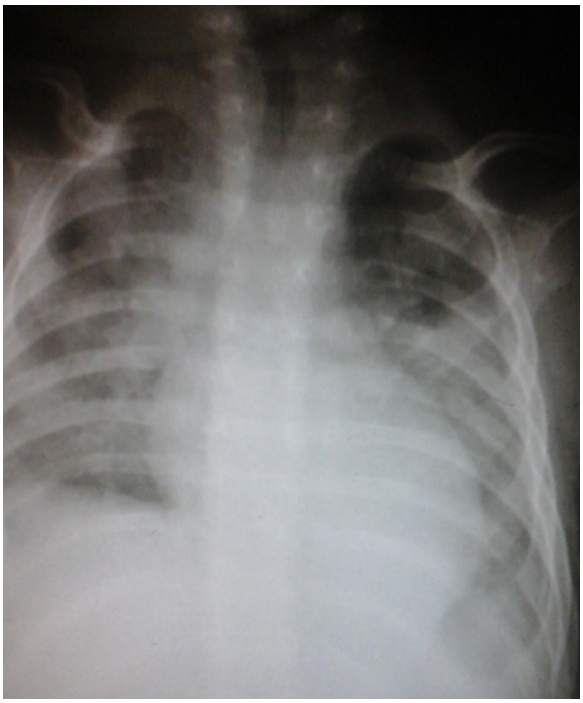

Fig. 3. After one week

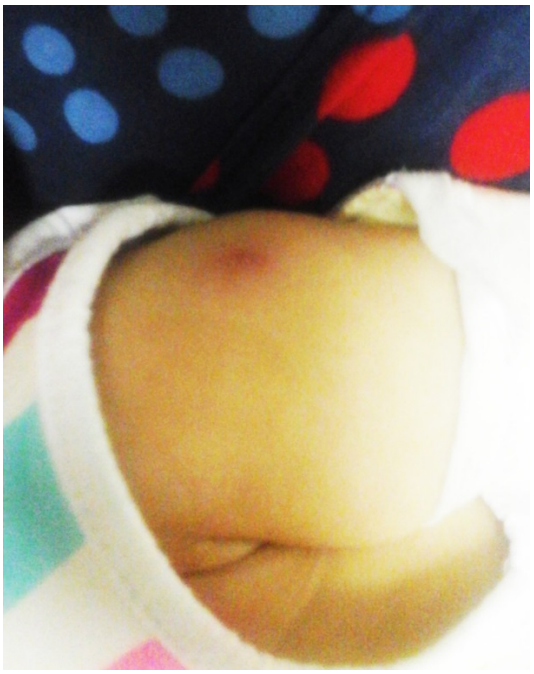

Fig. 1. Progressive erythema at the site of BCG vaccination on the arm of the case at the time of admission

in the hilar regions of the lungs (Fig. 1), which progressed to white lung appearance in one week (Fig. 2) and at the time, she was admitted in PICU due to exacerbation of respiratory distress and hypoxia.

She received broad-spectrum antibiotics, and was evaluated for different types of organisms. Due to high levels of ferritin in serum she undergone bone marrow aspiration which revealed no pathologic change, and diseases like hemophagositic lymphohystiocytosis were ruled out.

Laboratory investigations revealed positive IgM for CMV, so her serum undergone PCR and showed more than one million copies of the virus. She was started on Gancyclovir and her condition improved in time. After three weeks of treatment, another PCR was performed this showed less than 2 thousand copies of the virus.

Due to the diagnosis of sever life threatening CMV pneumonia and absence of the thymus on CXR, she was suspected to have immunodeficiency and undergone immunological panel tests which revealed: WBC=6.3 $\times$ 10^3 counts with 32% lymphocyte and 64% segment, Hemoglobolin= 8.9 gr/dl, plt= 285 $\times$ 10^3. NBT>90%, and normal DHR. IgG=0.89 mg/dl (4.1-10.81), IgM=0.2 mg/dl (0.48-2.28), IgA=0.04 mg/dl (0.13-0.82). Flowcytometry revealed: CD8=5% (19-37), CD4=0.1% (35-55), CD3=0.4% (68-82), CD2=9% (78-85), CD19=27% (5-15), CD20=20% (5-15), CD16/56=24%, CD11a=88% (85-106), CD11b=99% (25-35), CD11c=77% (25-35), CD18=95%, CD45+99%. HIV Eliza was negative. Laboratory tests were redone in another laboratory setting which revealed the same findings and she was diagnosed with T negative B^+NK^+ SCID.

At the same admission she also presented progressive erythema at the site of BCG vaccination on her arm (Fig. 3) and responded well to anti-TB antibiotic therapy and the erythema was re-

solved. Also received prophylactic Co-trimoxazol and anti-fungal therapy during the hospital stay.

She was discharged with ganciclovir, anti-fungal and anti-TB drugs and prophylactic Co-trimoxazol. She was scheduled for bone marrow transplantation which was successfully pre-formed with no further complications.

Discussion

SCID, a profound defect in T cell differentiation and development, results in complete lack of adaptive immunity (1-3). The disease is considered a medical emergency due to its high mortality rate if left untreated. Treatment is based on haematopoetic cell transplantation which if is done successfully, it is a cure for the disease (5,6).

We reported a case of SCID which presented with CMV pneumonia. She was clinically healthy in the past and had gained weight normally and undergone vaccination program with no complication until the age of 5 months. Her CMV infection rapidly progressed and she was admitted in PICU for severe respiratory distress but responded well to anti-viral therapy. Also experienced activation of Mycobacterium at the site of previous BCG vaccination, which resolved with anti-TB antibiotic therapy.

Cell mediated immunity is the major defense against CMV infection. Inadequate immune response of CD-4 T Cells leads to defective immunity against CMV infection leading to high morbidity and mortality burden of the infection in SCID patients (9).

Sever pneumonia with viral etiology, particularly CMV pneumonia, should raise the possibility of the presence of a life threatening immune deficiency (4).

Although few studies have been performed on CMV pneumonia as the first manifestation of SCID, given the importance of early diagnosis of SCID, one should consider these severe infections as an indication to assessing the immune system in affected patients to preserve a better approach, treatment and prognosis

Conflicts of interest: None declared.

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