

Case report

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Common variable immunodeficiency syndrome with right aortic arch: a case report

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Abstract

Background: Common variable immunodeficiency syndrome predominantly affects adults. It is characterized by low production of all the major classes of immunoglobulins. We report a case of common variable immunodeficiency syndrome with right aortic arch. An association of right-sided arch and common variable immunodeficiency syndrome has not been previously reported.

Case presentation: A 41-year-old female patient presented with a history of recurrent pneumonia, sinusitis, otitis media, diarrhoea, cystitis since childhood. Biochemical and immunocytochemical analysis revealed common variable immunodeficiency syndrome and radiological evaluation confirmed right aortic arch and aberrant left subclavian artery.

Conclusion: Common variable immunodeficiency syndrome syndrome is a clinical entity that should be kept in mind in patients with recurrent infections of different sites.

Background

Common variable immunodeficiency (CVID) is the most prevalent primary immunodeficiency, and predominantly affects adults. The primary defect remains unknown, but it seems peripheral blood T lymphocyte dysfunctions in a substantial proportion of CVID patients, which may impair T-B cell collaboration. CVID involves low levels of most or all of the immunoglobulin (Ig) classes, a lack of B lymphocytes or plasma cells that are capable of producing antibodies, and recurrent bacterial infections [1].

Case presentation

A 41-year-old woman admitted to infectious diseases clinics with productive cough, fatigue and postnasal drip for three days. She reported repetitive attacks of sinusitis, otitis media, diarrhoea (giardiasis and amebiasis were detected many times), cystitis and pneumonia since childhood.

She had a history of symptomatic therapy (non-specific antibiotics). She had tonsillectomy (15 years ago), appendectomy (8 years ago), adenoidectomy and paranasal sinus operation (5 years ago). She has no history of smoking.



Figure 1
Right displacement of aortic arc imaging with plain chest radiography

On admission, she was oriented and well cooperated, body temperature was 38°C, pulse rate was 106 beats/min, blood pressure was 120/80 mmHg, respiratory rate was 22 breaths/min, and she had wheezing. On chest examination, early inspiratory crackles were auscultated on both lower lobes of the lungs. There was a postnasal mucopurulent secretion.

Laboratory examination revealed haemoglobin: 8.8 g/dL, haematocrit: 27%, RBC: 2.8 M/uL WBC: 30000/mL with 94.2% of neutrophils and platelet: 207.000/mL. Bone-marrow aspiration was normal. Her biochemical results were within normal limits with a decreased globulin level of 1.2 g/dL, erythrocyte sedimentation rate was 43 mm/h. In urine, leukocytes (especially neutrophils) were detected. Non-homogenous infiltrations were present on the paracardiac areas of both lungs on admission chest radiography. C-Reactive protein level was 42 mg/L (normal range: 0–6 mg/L). Anti-Streptolizin-O titres increased. The levels of IgA, IgE, IgM and IgG were 30 mg/dL, 3.2 IU/mL, 25 mg/dL and 100 mg/dL respectively by serum assay. IgG subgroups were IgG-1: 92 mg/dL, IgG-2: 22 mg/dL, IgG-3: 13 mg/dL and IgG-4: 2 mg/dL. Serum albumin 52.1%, alpha-1 globulin 7.1%, alpha-2 globulin 19.6% beta globulin 13.6% and gamma globulin 7.7% levels were detected with protein electrophoresis. Immu-



Figure 2
Right displacement of aortic arc with computerized tomography

nocytochemical analysis revealed these levels CD3: 76.45%, CD19: 7.7%, CD4: 11.9%, CD8: 51.4%, CD4/CD8: 0.23, CD5: 70.5% and CD45: 86.3%. The diagnosis of common variable immunodeficiency (CVID) syndrome was established with immunocytochemical tests. She had no autoantibodies.

The patient was seronegative for HbsAg, HCV, HIV-1, and HIV-2. Sputum culture revealed *Pseudomonas aeruginosa*. Throat culture and stool examination was normal. The right displacement of aortic arch was noticed on plain chest radiography (Figure 1) and high-resolution computerized tomography (CT) scanning of the thorax revealed right aortic arch, aberrant left subclavian artery (Figure 2), and bronchiectasis on both lower lobes of lungs (Figure 3). With magnetic resonance (MR) angiography, right aortic arch and aberrant left subclavian artery was confirmed. In CT scanning of the face, bilateral chronic maxillary sinusitis was detected.

Pulmonary function tests revealed, FVC: 2180 mL (65.1%), FEV1: 1610 mL (55.7%), FEV1/FVC: 86%, PEF: 3250 mL (48%) with negative reversibility. These findings did not resolve completely after antibiotic treatment. Fiberoptic bronchoscopy was scheduled but could not be achieved because of bronchospasm.

The patient had received salbutamol inhaler form, intravenous immunoglobulin (IVIg) 400 mg/kg/day for 5 days and imipenem/cilastatin 2 g/day intravenously in the



Figure 3
Bronchiectasis on both lower lobes of lungs with computerized tomography

intensive care unit. Intravenous immunoglobulin prophylaxis 400 mg/kg/day for one day and bronchodilator therapies were recommended once a month after discharge from the hospital.

Discussion

Common variable immunodeficiency (CVID) is a diagnostic category, which includes a heterogeneous group of males and females, mostly adults, who have in common the clinical manifestations of deficient production of all types of major immunoglobulin classes [1]. In our case, diagnosis was confirmed also by the deficiency of all types of immunoglobulin classes. The majority of these panhypogammaglobulinemic patients have normal numbers of B-lymphocytes that are clonally diverse but phenotypically immature. B-lymphocytes are able to recognize antigens and can proliferate in response to these antigens but fail to differentiate to become plasma cells [1].

The development of the abnormal branching patterns of the aortic arch, arteries is traditionally explained by transformation of the primitive embryonic pharyngeal arch arterial system due to obliteration of some of its vascular segments. Based on this concept, the isolation of an aortic arch artery can be explained by obliteration of vascular segments proximal and distal to this artery, whereas its connection to a pulmonary artery can be explained only by the deficiency obliteration (persistence) of the distal portion of the right or left sixth pharyngeal arch artery. The segments of the bilateral aortic arch system that nor-

mally regresses include the distal portion of the sixth arch and the right-sided dorsal aorta [2].

In differential diagnosis, all primary immunodeficiencies including combined immunodeficiencies, predominantly antibody deficiencies, other well-defined immunodeficiency syndromes (Wiskott-Aldrich syndrome, Ataxia telangiectasia, DiGeorge anomaly, etc) and acquired ones should be kept in mind [1].

In adults who present with chronic pulmonary infections with unexplained bronchiectasis it is important to consider the diagnosis of CVID [3,4]. Our patient had also a history of recurrent lower respiratory tract infection attacks since her childhood and was diagnosed bronchiectasis on lower lobes of both lungs with HRCT. Intestinal diseases, including chronic giardiasis and amebiasis, intestinal malabsorption, and atrophic gastritis with pernicious anaemia are common in these patients as seen in our case. Oral disseminated mycoplasma infections have also been reported [1,5]. In medical history of our patient there were recurrent giardiasis and amebiasis infestations. These patients also may present with signs and symptoms of highly suggestive lymphoid malignancy including fever, weight loss, splenomegaly, generalized lymphadenopathy and lymphocytosis. Routine histological examination of lymphoid tissues usually reveals germinal centre hyperplasia, which may be difficult to distinguish from nodular lymphoma [6].

The monthly administration of intravenous immunoglobulin in adequate doses is an essential part of the prevention and treatment of all these complications [7]. The patient was given prophylactic IVIG monthly.

For the diagnosis of vascular anomalies, MR angiography is the best choice [8]. We noticed the right displacement of aortic arcus on plain chest radiography and we confirmed this and the aberrant left subclavian artery with MR angiography. This vascular abnormality could cause progressive dysphasia and dyspnoea [2,9], but our patient had none of these symptoms. Right aortic arch could be seen with some congenital anomalies such as Ullrich-Turner syndrome [10]. CVID syndrome with right aortic arch and aberrant subclavian artery is a very rare finding; because in computer searched base investigation it is seen that CVID with vascular abnormalities has not been previously described. Various cardiovascular abnormalities have been reported in T cell deficiencies, but such anomalies are very rare in B cell deficiencies including CVID as in our case [11].

Conclusion

In the current report, a CVID case that has cardiovascular anomalies was presented, but we couldn't say that there is

a specific relationship between them in the current literature. We think that the issue needs to be clarified with further studies. CVID syndrome is a clinical entity, which should be kept in mind in patients with recurrent infections of different sites.

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Written consent was obtained from the patient for publication of this case report.

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