CASE REPORT

Young female with repeated chest and gastrointestinal infections

Qurat Ul Ain, Rabia Rasool, Saliha Afsheen

District Headquarters Hospital, Jhelum Pakistan

ABSTRACT

Common variable immunodeficiency disorder (CVID) is a primary immunodeficiency disorder characterised by reduced serum levels of immunoglobulin G (IgG), along with immunoglobulin A (IgA) and/or immunoglobulin M (IgM).[1] The range of clinical manifestations is broad, including acute and chronic infections, inflammatory and autoimmune disease, and an increased incidence of cancer and lymphoma.[2] We present a case of a young female with bronchial asthma, repeated chest infections, and a prolonged episode of gastroenteritis. After a detailed history, examination, and investigations, she was diagnosed with CVID. She was managed with IV fluids, antibiotics, and antiprotozoal medications. After one month of follow-up, she was doing well.

Keywords: Immunodeficiency, Immunoglobulins, CVID, Respiratory tract infection, IgA, IgG, IgM, asthma, gastroenteritis

BACKGROUND

CVID is a genetic immune disorder that results in decreased levels of immunoglobulin (antibodies).[3,4] This includes IgG and at least one other immunoglobulin, either IgA or IgM.[4] The degree and type of immunoglobulin deficiency vary from individual to individual, leading to a variable clinical presentation. It is typically seen in the second to fourth decades of life.[4] Respiratory tract infection (RTI) is a prominent clinical presentation often manifesting as recurrent sinusitis, bronchitis, or pneumonia.Other common manifestations include gastrointestinal symptoms, such as chronic diarrhea or malabsorption, and rarely autoimmune conditions.[5] Early diagnosis and appropriate management are crucial in improving outcomes in individuals with CVID. To improve the recognition of immunodeficiency disorders, they should be suspected in every patient with recurrent bacterial infections.

CASE REPORT

A 27-year-old female, resident of Jhelum, has been in poor health since she was 18. She had been suffering from respiratory symptoms including shortness of breath and cough, sometimes associated with fever. In the initial two years, her symptoms were seasonal and used to be relieved by taking oral medications but gradually her symptoms aggravated. She then consulted general practitioners (GPs), who prescribed her nebulization and IV medication. She had been using ipratropium and beclomethasone nebulization on an as-needed basis for the last seven years. She often used antibiotics for an associated fever.

She presented to the medical OPD of our hospital with a history of loose stools and vomiting for two months, aggravated for the last week. Stools were non-bloody and foul-smelling, and were associated with colicky abdominal pain. Vomiting was non-projectile, small volume, and non-bloody. She took treatment from her GP but her condition did not improve. She had a prior history of the passage of long worms in her stools (commonly ascariasis in our part of the world). The patient had experienced severe body aches and pains for the past two years. She had difficulty combing her hair and also had difficulty standing from a sitting position. She has developed hair loss over the past couple of years but there was no history of mouth ulcers, photosensitivity, or skin rash. She was married with three kids. Her husband was a shopkeeper and she belonged to a poor socioeconomic class. She lived in a two-room house and used well water for drinking and domestic purposes. She practiced hand hygiene in daily life.

On examination, she was dehydrated. Her pulse rate was 100/min and her blood pressure was 100/60 mmHg. She was afebrile and had a respiratory rate of

Correspondence: Dr. Qurat Ul Ain, Department of Internal Medicine, District Headquarters Hospital, Jhelum Pakistan

Email: dr_qurat74@yahoo.com

This case report can be cited as: Ain QU, Rasool R, Afsheen S. Young female with repeated chest and gastrointestinal infections. 2024; 33(4): 214-217.

DOI: https://doi.org/10.61529/idjp.v33i4.300

Receiving date: **05 Apr 2024** Acceptance Date: **19 Nov 2024**
Revision date: **15 Nov 2024** Publication Date: **30 Dec 2024**

20/min. The abdomen was soft and non-tender. The chest examination revealed bilateral expiratory rhonchi.

She was admitted to the medical ward. Routine blood test reports were normal. A stool examination routine (stool RE) showed the presence of pus cells. A stool culture did not reveal any organism (the sample was obtained after the start of treatment). Specific labs are tabulated as follows. (Table 1) Her vitamin D3 level was normal. Thyroid function tests (TFT) were within the normal range. C-reactive protein (CRP) was raised. Hepatitis B surface antigen, HIV, and Hepatitis C serologies, all were negative.

HRCT Chest: Mild bilateral pulmonary infiltrates suggestive of acute inflammatory changes. No evidence mediastinal lymph nodes or pleual effusion. She was treated with normal saline, antiemetics, ceftriaxone, and metronidazole. Metronidazole was given due to the high prevalence of *Entamoeba histolytica*-associated enteritis in our area, though stool RE did not detect any vegetative form of the parasite. The patient showed significant improvement after a 7-day stay in the hospital and was subsequently discharged with OPD follow-up.

Ultrasound Abdomen:

1. Fatty liver
2. Mild splenomegaly

Table-I: Autoimmune antibodies and quantitative immunoglobulins.

Autoimmune Disorders		
C-ANCA-PR3	Negative	
Patient Value	0.42	
Reference ranges positive >1.1 negative <0.9 grey zone 0.9-1.1		
P-ANCA-MPO	Negative	
Patient Value	0.59	
Reference ranges *positive > 1.1 negative <0.9 grey zone 0.9-1.1*		
ANTI TISSUE TRANSGLUTAMINASE (IgA)	Negative	
Patient Value	4.86 U/ml	
Reference Ranges *positive >10.0U/ml*		
ENDOMYSIAL ANTIBODIES (IgA)	Negative	
Patient Value	3.91 U/ml	
Reference ranges *positive >10.0 U/ml*		
Immunology		
IgG	1.7 g/dl	***Reference Range*** 7-16 g/dl
IgA	50 mg/dl	***Reference Ranges*** 90-450 mg/dl
IgM	1.1 g/dl	***Reference Ranges*** 0.4-2.3 g/dl
IgE Levels	172.0 iu/ml	***Reference Interval*** *0-100 iu/ml*

DISCUSSION

Immunodeficiency syndromes result from defects in components of the immune system, including lymphocytes, phagocytes, and the complement system. These immunodeficiencies can be either primary or secondary. Primary immunodeficiency (PID) is a large heterogeneous group that results from inherited defects in the immune system, while secondary immuno-deficiency disorders result from external factors such as infections, medications, or underlying medical conditions.[3, 5]

CVID has an incidence of approximately 1 in 25,000 individuals and is more prevalent in Northern Europe.[2,4] The majority of patients present between the ages of 20 and 45, with no association with any particular race or gender. The overall mortality in CVID patients, according to age and sex-matched population controls, is around 20%.

"Variable" refers to the heterogeneous clinical manifestations of this disorder, which include recurrent bacterial infections, increased risk for autoimmune disease and lymphoma, as well as gastrointestinal disease. CVID is a lifelong disease.[4] The main cause remains unknown, though environmental and genetic factors may be involved.[2,6] While the specific environmental factors are unclear, genetic influences in CVID are believed to cause intrinsic B cell defects (such as CD19 deficiency), intrinsic T cell defects, and mutations in TNF receptors.[5] However, CVID can also be present without a known genetic defect.The

hallmark of CVID is severe and recurrent bacterial infections, especially those involving the respiratory tract.[5]Asthma may be the most common chronic respiratory manifestation, as it is the most often reported.[7] Some patients may also present with bronchiectasis. Other associations of CVID include autoimmune and granulomatous diseases, as well as an enhanced risk of malignancy, such as lymphoma and gastric carcinoma.[2,4]

Treatment typically involves the administration of immunoglobulin once per month throughout the patient's life.[8,9,10] Immunoglobulin therapies have been shown to increase life expectancy and reduce the development of autoimmunity. Bacterial infections are treated with longer courses of antibiotics, though prophylactic antimicrobials are not routinely recommended.

Our patient had a history of repeated asthma exacerbations. She presented to us with prolonged symptoms of gastroenteritis that did not respond to oral antibiotics. Gastroenteritis settled down after seven days of IV ceftriaxone and metronidazole therapy. Although stool culture and sensitivity did not identify the growth of any organism (the sample was taken after the start of antibiotics), a repeat stool RE showed the persistence of pus cells. Due to the presence of pus cells in repeat stool RE and low serum immunoglobulin levels, antibiotics were administered for an extended period. She had a past history of repeated sinusitis, as evidenced by her previous GP record. Even at presentation to our hospital, she had nasal crusting. Her autoimmune workup for Granulomatosis with polyangiitis and Churg Strauss syndrome was negative. She had reduced levels of IgA and IgG. Advanced molecular tests were unavailable at our setup. Her HRCT chest did not show any evidence of bronchiectasis. She was referred to a tertiary care hospital to receive IV immunoglobulins, but she could not go, and she is under regular OPD follow-up. She has been advised to seek medical attention if she experiences fever, cough, diarrhea, throat, ear, or skin infections. She has also been advised to use boiled water and maintain strict hygiene daily.

CONCLUSION

Due to the lack of awareness among healthcare providers, many individuals with CVID remain undiagnosed, leading to recurring infections and serious co-morbid conditions. Therefore, raising awareness among primary care physicians is crucial for the management and prevention of complications.

CONFLICT OF INTEREST

None

GRANT SUPPORT & FINANCIAL DISCLOSURE

Declared none

AUTHOR CONTRIBUTION

Qurat Ul Ain: Conceptualization, manuscript writing, final approval, accountable for every aspect of the work

Rabia Rasool, Saliha Afsheen: Manuscript writing, final approval, accountable for every aspect of the work

REFERENCES

1. Pescador Ruschel MA, Vaqar S. Common Variable Immunodeficiency. [Updated 2023 May 8]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan-. Available from:https://www.ncbi.nlm.nih.gov/books/NBK549787/
2. Bazregari S, Azizi G, Tavakol M, Asgardoon MH, Kiaee F, Tavakolinia N, *et al*. Evaluation of infectious and non-infectious complications in patients with primary immunodeficiency. Cent Eur J Immunol. 2017;42(4):336–41. DOI: https://doi.org/10.5114/ceji.2017.72825
3. McCusker C, Upton J, Warrington R. Primary immunodeficiency. Allergy Asthma Clin Immunol. 2018 Sep;14(S2): 61.DOI: https://doi.org/10.1186/s13223-018-0290-5
4. Ghafoor A, Joseph SM. Making a diagnosis of common variable immunodeficiency: A review. Cureus. 2020; 12 (1): e6711.DOI: https://doi.org/10.7759/cureus.6711
5. Filion CA, Taylor-Black S, Maglione PJ, Radigan L, Cunningham-Rundles C. Differentiation of common variable immunodeficiency from IgG deficiency. J Allergy Clini Immunol Practice. 2019;7(4):1277–84. DOI: https://doi.org/10.1016/j.jaip.2018.12.004
6. Justiz Vaillant AA, Ramphul K. Antibody Deficiency Disorder (Archived). 2023 May 23. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; Treasure Island (FL): StatPearls Publishing; 2024 Jan–. PMID: 29939682. Available from: https://www.ncbi.nlm.nih.gov/books/NBK507905/
7. Weinberger T, Fuleihan R, Cunningham-Rundles C, Maglione PJ. Factors beyond lack of antibody govern pulmonary complications in primary antibody deficiency. JClinImmunolo. 2019; 39 (4) 2019; 440–7. DOI: https://doi.org/10.1007/s10875-019-00640-5
8. Pilania RK, Chaudhary H, Jindal AK, Rawat A, Singh S. Current status and prospects of primary immunodeficiency diseases in Asia. Genes Dis. 2019; 7 (1): 3–11. DOI: https://doi.org/10.1016/j.gendis.2019.09.004
9. Westh L, Mogensen TH, Dalgaard LS, Jensen MB, Katzenstein T, Hansen ABE, *et al*. Identification and characterization of a Nationwide Danish adult common

variable immunodeficiency cohort. Scandinavian J Immunol. 2017;85(6):450–61.
DOI: https://doi.org/10.1111/sji.12551

10. Bonilla FA, Barlan I, Chapel H, Costa-Carvalho BT, Cunningham-Rundles C, de la Morena MT, *et al*. International Consensus Document (ICON): Common variable immunodeficiency disorders. J Allergy Clin Immunol Pract. 2016;4(1):38–59.
DOI https://doi.org/10.1016/j.jaip.2015.07.025