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Internal Medicine

VARIED PRESENTATIONS OF PRIMARY IMMUNODEFICIENCY DISORDER IN TERTIARY CARE SET-UP

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KEYWORDS :

Introduction:
Primary Immunodeficiency Disorders (PIDs) are largely Mendelian inherited genetic illnesses. Deleterious mutations in 420 genes have been found, and more than 450 diseases have already been defined. PIDs' overall prevalence has been calculated to be five to ten per 100,000 people in other countries. In the past 1.5 years, there have been numerous admissions for recurring infectious diseases at our facility. Among these, four young individuals with suppurative illnesses and a history of previous repeated admissions raised our suspicions about primary immunodeficiency disorders.

Case Summary:
First case was a 16 year old girl presenting as febrile illness with cough and breathlessness diagnosed to have left lower lobe consolidation with sympneumonic effusion. There was past history of similar lower respiratory tract infections with documented evidence of immune thrombocytopenia treated with Rituximab. On further investigations she had deficiency of IgA/G. Eventually diagnosed as Common Variable Immunodeficiency Disorder. Second case was an 18 year old male who presented with perennial cough with expectoration with weight loss and growth retardation. He had hypo-pigmented patches since 6 years and recurrent ICU admissions for lower respiratory tract infection. X-ray and CT chest revealed bronchiectatic changes. Serum Ig levels and lymphocyte subset analysis was suggestive of low IgG/A and low B cell fractions and Genetic studies revealed X linked lymphoproliferative disorder.

Third case was 14 year old male who presented with seasonal exacerbations of cough with mucopurulent expectoration since childhood with failure to thrive. Examination revealed multiple telengiectasia on oral mucosa. X-ray, CT chest showed bronchiectatic changes. Serum IgA/G were low with normal B and T cell number. Patient was diagnosed as Ataxia telangiectasia presenting as common variable immunodeficiency disorder. X 4th case was a 25 year old male who presented with headache with altered sensorium and past history of abdominal TB. On enquiry he had a history of recurrent ear infections and meningitis in the past. CSF analysis confirmed Pyogenic meningitis in current admission. Serum IgG/A/M were low with normal IgE.

All these patients were advised monthly Intra venous immunoglobulin, however only the first and forth patient could manage to receive treatment. Third patient eventually succumbed to the illness.

Discussion:
These individuals had the typical symptoms of common illnesses, but a considerable prior history of recurrent infections that required indoor management suggested that they may have had primary immunodeficiency disorder (PIDs). PIDs can manifest in a variety of ways, including meningitis, recurrent respiratory infections, and otitis. Several PIDs exhibit a confluence of recurring infections, inflammation, and autoimmunity, posing treatment problems. Despite the fact that the underlying mechanism of disease activity is still not fully understood, just 10% of instances are known to have a genetic component, and 25% of patients also have an autoimmune condition. Also it is noteworthy that malignancies like lymphoma are more common in some PIDs, such as X linked lymphoproliferative disease.

Conclusion:
Prudent follow up, strict compliance to IVIG and early detection of infections and malignancy are the main objectives while managing PIDs.

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