

UNIT OF AXIS JOURNALS

International Peer Reviewed Medical Journal Committed for Excellence

Recurrent infections in a five year old boy

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CASE REPORT

ABSTRACT

Children are prone to develop recurrent illness in the form of upper respiratory tract infections, gastroenteritis, pustulosis and otitis media owing to developing immunity and environmental exposures. Majority of such children improve on symptomatic treatment. However, few of them might develop severe life threatening infections affecting vital organs. Primary immunodeficiency is an entity in which children develop recurrent infection due to defect in the development of immune system. This may affect plasma cells, T lymphocytes, natural killer cells or peripherally circulating leucocytes. We present a 5 year old male child with such diagnosis. A five year old boy was admitted with history of repeated infections since one year of age. His serum IgA, IgG and IgM levels were markedly decreased. Flow cytometry showed absence of B cells. He was started on intravenous immunoglobulin 10 gm every four weeks following which there was marked reduction in intercurrent infections. Primary immunodeficiency should always be suspected in differential diagnosis if history of repeated infections exist.

Key Words

Primary immunodeficiency, recurrent infections.

Received on 18 Oct 2014

Accepted on 17 Nov 2014

Published on 30 Nov 2014

INTRODUCTION

Pediatric patients presenting with recurrent episodes of infections is common.¹ However identifying patients with immunodeficiency is a challenge. Here we present a case of primary immunodeficiency to highlight which subset of patients with history of recurrent infections should be screened for immunodeficiency.

CASE REPORT

A five year old male child born out of non consanguineous marriage was admitted with complaints of cough and cold since 5 days. Further mother informed that baby was alright till one year of age after which he developed several episodes of loose motions which used to subside with treatment. He also had history of ear discharge and boils over body. At the age of two and a half years he was admitted to a hospital with complaints of fever and convulsions. Blood investigations and lumbar puncture was suggestive of pyogenic meningitis for which he received a

course of intravenous antibiotics and improved. Now since last two years child develops repeated episodes of fever, cough and cold which responds to treatment. Patient has received antitubercular therapy for these complaints. His elder sibling died at the age of 8 years and had similar medical history. Younger sister was alive and healthy. On examination, child was well nourished. There was no lymphadenopathy. Systemic examination revealed added sounds in respiratory system. There was no significant finding in any other system. Investigations revealed a normal haemogram and a normal chest skiagram. Echocardiography revealed a normal heart structure with normal functioning of chambers. His IgA levels were 13 mg/dl (normal range 25-160mg/dl), IgG were 40 mg/dl (460-1240 mg/dl), IgM were 10 mg/dl (45-200 mg/dl) and IgE were 26.50 U/l (< 150 IU/l). Flow cytometric immunotyping for B cells, T cells and natural killer cells was done which revealed normal CD3 cells 95.99 %, normal CD16 and CD56 natural killer cells.

However the percentage of CD19 B cells was markedly decreased 0.06% (normal 10-31%) which is consistent with X linked agammaglobulinemia. Patient is on intravenous immunoglobulin 10 grams every four weeks and the number of intercurrent infections has reduced.

DISCUSSION

Recurrent infections are amongst the most frequent diagnostic dilemmas for primary care physicians. Clinicians should have a high index of suspicion to identify immunodeficiency. Evaluation of immune function should be done in children with unusual, chronic or recurrent infections like one or more systemic bacterial infections; two or more serious respiratory or documented bacterial infections; serious infections occurring at unusual sites (liver, brain abscess); infections with unusual pathogens (aspergillus, Serratia); infections with common childhood pathogens of unusual severity.¹

X linked agammaglobulinemia (XLA) is a recessive primary immunodeficiency disorder with a reported prevalence of 1/10,000 in the general population.²

X-linked agammaglobulinaemia (XLA) is a humoral immunodeficiency disease characterised by severe hypogammaglobulinaemia, defective B cell development, and extremely decreased numbers of mature B cells.³ The gene responsible for XLA is Bruton's tyrosin kinase (BTK). The BTK gene is a cytoplasmic tyrosine kinase that plays a critical role in the development of B cells.⁴

The clinical diagnosis of XLA depends on a positive family history of immunodeficiency, recurrent bacterial infections before the age of 5 years, life-threatening bacterial infections in early childhood, and considerably low levels of all isotypes of serum immunoglobulins.⁵

X linked agammaglobulinemia can present at any age and is associated with autoimmune disorders.⁶

CONCLUSION

Primary immunodeficiencies are often missed due to either lack of awareness or availability of laboratory tests. X linked agammaglobulinemia is a common immune disorder which if identified early can avert mortality from life threatening infections.

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Please cite this paper as: Goyal S, Goyal V. Recurrent infections in a five year old boy. *Inter J Medical Sci Res Prac* 2014; 1 (3): 103-104.

ACKNOWLEDGEMENTS

Nil

PEER REVIEW

Double Blinded externally peer reviewed.

CONFLICTS OF INTEREST

Nil

FUNDING

Nil