

SELECTIVE IMMUNOGLOBULIN M DEFICIENCY (SIgMD)- HYPERPHENYLALANINEMIA SYNDROME

Bandika V.L; Dormohamed N; Faraj I; Kassim F; Mbuga M.

Corresponding author: bandikav@yahoo.com

Department of Paediatrics and Child health, Coast Provincial General Hospital. Mombasa.

INTRODUCTION

SIgMD is a rare dysgammaglobulinemia, characterised by isolated low serum immunoglobulin M (IgM). SIgMD can be syndromic or non syndromic, manifesting with life-threatening invasive infections caused by encapsulated bacteria, fungi and viruses. To date, it's occurrence in the background of congenital hyperphenylalaninemia due to co-factor defect has not been described. In poor resource setting the diagnosis is missed due to low index of suspicion and unavailability of expensive diagnostic tests.

METHODOLOGY

We present a 21 month old male from non consanguineous parents with persistent fevers, recurrent episodes of diarrhoea, convulsions, vomiting and gross developmental delay since birth. He had evidence of sequential multi systemic severe infections. An older male sibling succumbed from a similar illness at 6 months. Several investigations were done: elevated blood inflammatory markers. Blood, urine cultures and sensitivities grew multiple organisms such as klebsiella, pseudomonas, acinetobacter and candida species with multiple antibiotic resistance pattern. Serum amino acid profile and urine organic acid analysis revealed hyperphenylalaninemia secondary to co-factor defect. Quantitative immunoglobulin assay showed low IgM (25.5mg/dL). Tuberculosis and Human immunodeficiency virus were excluded. Multidisciplinary clinical approach using various antibiotic combo and dietary interventions was implemented. Our patient did not benefit from prophylactic antibiotics due to severe sequential septicaemia.

DISCUSSION

To our knowledge SIgMD with hyperphenylalaninemia has not been described. Relationship between the two is quite obscure thus making us hypothesized of a possible syndromic relationship. This needs to be examined further. For timely diagnosis, a high index of suspicion is needed.

CONCLUSION

Patients with SIgMD are susceptible to recurrent severe infections that often lead to mortality. It's occurrence with hyperphenylalaninemia suggests a syndromic presentation.