Correspondence

Autoimmune Polyglandular Syndrome Type - 1

Sir,

Autoimmune polyglandular syndrome type -1 is a rare disorder which is characterized by mucocutaneous candidiasis, hypoparathyroidism and adrenal insufficiency. Patients must satisfy at least two of these criteria to confirm the diagnosis of this syndrome. A fifteen years old female was admitted in October 2010 to the Department of Medicine CSMMU Lucknow India with a history of mild fever and weakness of three-weeks duration. On general examination her blood pressure was 70/50 mmHg, pulse was 120/minute. She improved only partially after administration of intravenous fluids (0.9% normal saline and vasopressors (dopamine 15 microgram/kg/minute, norepinephrine 20 microgram/minute). On the basis of hypoparathyroidism, low serum cortisol and shock; a diagnosis of Autoimmune polyglandular syndrome type-1 was suspected. For the confirmation of diagnosis following specific investigations were done. She was started on prednisolone and fludrocortisone in addition to her previous medications. Her condition improved and she was discharged in good condition. On follow up, she showed normal growth parameters, normal blood pressure and normal blood glucose, serum electrolytes and calcium levels.

In majority of cases, autoimmune polyglandular syndrome type -1 (APS-1) occurs in childhood and may occur among siblings. It is anautosomal recessive disorder with mutation of the autoimmune regulator (AIRE) gene on chromosome 21q22.3. It is a sporadic autosomal recessive disorder. With regard to genetic susceptibility, APS-1 is unique among autoimmune endocrine disorders, because it has no HLA antigen association. However an increased frequency of HLA-A28, HLA-A3 has been documented in APS-1 more so than in normal controls. The genetic locus responsible for the disease has been localized to the short arm of chromosome 21 near markers D21s171 on band 21p22.3. A Finnish study concluded that the mutation R257X is responsible for 82% of cases. Frequency of mucocutaneous candidiasis is lower in the patients without the R257X allele and patients carrying the DRB1*03 allele had significantly higher prevalence of Addison’s disease. This and the DRB1*04 allele have been shown to be associated with non-autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED) autoimmune Addison’s disease.

So HLA-DRB1*03 and DRB1*04 when present only responsible for phenotypic presentation in APS-1 and non APS subject. A monogenic mutation of AIRE (autoimmune regulator), which codes for a putative transcription factor featuring 2 zinc motifs, is believed to be the likely pathogenic paradigm for APS-1. Treatment of this disease is that of the individual components, such as antifungal treatment for mucocutaneous candidiasis, calcium and vitamin D supplementation for hypoparathyroidism, and steroid replacement for adrenal insufficiency.

This is the first case seen in our department over the last 10 years. This shows the importance of looking for such symptoms which may uncover the diagnosis of other similar cases.

References

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Gangrene and Neurological Complications of Malaria

Sir,

We read the interesting case report entitled symmetrical peripheral gangrene and neurological manifestations in plasmodium falciparum malaria by Kotokey and Kaushik published in July 2011 issue of JAPI (vol. 59, page no. 449-451). The article is worth documenting and there are only few case reports earlier mainly from Indian subcontinent/south east Asia.

1. Neurological manifestations in malaria without loss of consciousness as a presenting illness besides sequelae in survivors of cerebral malaria and delayed complications as post malaria neurological syndrome (PMNS) are documented earlier from India in quite a large number. These were mainly convulsions, psychosis, extra pyramidal rigidity, and ataxia, either alone or in varying combinations. All such patients had complete recovery with appropriate antimalarial treatment in due course of time. Of course not all such cases are being categorized as severe and complicated malaria as per WHO (World health organization) but should be treated on the line WHO guidelines meant for severe and complicated malaria.1

2. Computed Tomography of the reported patient showed atrophy. It would have been worthwhile if the authors had got done the Magnetic resonance imaging which probably could have defined the site of lesion for coxsosporial signs to some extent. Furthermore it was not clear that the atrophy was sequelae of malaria or there was some other alternative cause for that.

3. The authors used 5% dextrose for quinine infusion, but 10% dextrose would have been a better option as per our experience.2

4. Last but not least quotation of reference is also of utmost importance for any article and the authors need to review the quotation of reference number two.

References