12º CONGRESSO BRASILEIRO DE ALERGIA E IMUNOLOGIA EM PEDIATRIA

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Trabalhos Científicos

Título: Disseminated Histoplasmosis Caused By Il12rb1 Gene Mutations In Two Brazilian Siblings

Autores:

Resumo: DESCRIPTION: A 4-year old Brazilian boy was referred for evaluation with a history of fever, paleness, and dry cough. At presentation he had diffuse abdominal pain with liver and spleen 4,5 cm and 3 cm below the right costal margin, respectively. Cervical, submandibular and inguinal lymphadenopathy was also noted. Bone marrow aspirate analysis disclosed Histoplasma capsulatum. He responded well to antifungal treatment and was discharged receiving itraconazole prophylaxis. The patient developed chronic progressive CNS histoplasmosis at the age of 6 and his condition worsened with the development of hydrocephalus requiring a ventricular peritoneal derivation at the age of 7. His previous history included a tuberculous adenitis at the age of 1. During the patient's follow-up, his 4 year-old brother presented tuberculous adenitis followed by disseminated Salmonellosis in the next year. He also developed disseminated histoplasmosis at the age of 6. The parents are consanguineous and the patient's mother had 12 brothers and sisters, of whom eight (both male and female) died of an unexplained cause shortly after birth. Given the susceptibility to intracellular pathogens, a defect in the IL-12/23-IFN-y axis was suspected. DNA sequencing demonstrated a homozygous p.R283X mutation in the IL12RB1 gene, confirming the diagnosis of IL-12R?1 deficiency in both patients. No mutation was detected in the third younger sibling (1 year-old). COMMENTS: Pediatricians should be aware of this group of disorders, as proper diagnosis and prophylatic treatment can be life saving.