Abstract Code: ASID021-010

Challenges in diagnosing auto-inflammatory disorders in low resource settin

Tinsae Alemayehu*
*tigisttinsae@gmail.com**
*Ethiopia**

Kirkos sub-city

Keywords: Auto-inflammatory disorder, Primary immune-deficiencies, Child, Ethiopia.

Abstract

Background: With increasing awareness of the syndromes and their presentation, more children with primary immune-deficiencies are being diagnosed in Ethiopia with each passing year. The clinical presentation of a young Ethiopian boy suspected of having an auto-inflammatory disorder and the diagnostic and therapeutic challenges in a low income country are outlined.

Case: A three year old Ethiopian boy presented with recurrent respiratory and intestinal infections starting from two years of age. Episodes were notable for subcutaneous nodules shifting positions with each febrile episode, arthralgia, marked leukocytosis and neutrophilia, normal serum immunoglobulin levels and remaining hematologic parameters, normal chest xray but mesenteric adenitis, varying levels but persistently elevated CRP even during periods between febrile episodes, negative autoimmune and oncologic markers and negative markers for HIV, TB and other infectious illnesses. He had no atypical post-vaccine reactions and his maternal uncle has a «high predisposition for illnesses». He was circumcised at ten days of age (uneventful) and shed his umbilicus at one week of age. Febrile episodes are being managed with short-term oral steroids while exploring ways to do genetic testing for auto-inflammatory disorders.

Discussion and conclusion: There are numerous auto-inflammatory disorders and are increasing in number by the year. Though some members of this group are associated with specific communities – familial Mediterranean fever among Mediterranean populations, hyperimmunoglobulin D syndrome among north Europeans etc, some others like Tumor necrosis factor receptor-associated periodic syndrome can be diagnosed globally. Clinical features of auto-inflammatory disorders overlap and genetic analysis for known mutations is vital for diagnosis. A paucity in genetic labs makes confirmation of such disorders difficult in Africa. But continuing education will help identify common clinical features and avoid unnecessary work-up and antimicrobial treatment for similar patients.