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Mendelian Susceptibility to Mycobacterium Disease (MSMD): A Single Center Experience

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Abstract

Background: Mendelian susceptibility to mycobacterium tuberculosis(MSMD) is a rare group of inherited conditions characterized by selective predisposition to mycobacterial infection. We here demonstrate the clinical and molecular data of patients diagnosed with MSMD in our center.

Methods: One hundred infants and children with mycobacterial infection not responding to triple antituberculous therapy were evaluated for possible underlying Inborn errors of Immunity(IEI). Their clinical and immunological data were evaluated, and patients with MSMD apart from chronic granulomatous disease and other IEI were described.

Results: Out of the 100 patients screened,18 patients were suspected to have MSMD based on their clinical data and normal basic immunological tests. They were 12 females and 6 males(ratio 2:1). Their median age of onset, age at diagnosis and diagnostic lag were 6.5, 24 and 19 months respectively. Seventeen patients received live attenuated BCG vaccine at birth. Ten patients had recurrent fungal infections, four had disseminated salmonella infections and 3 patients had history of encephalitis. Genetic diagnosis was performed to eleven patients and it showed IL12RB1 mutation in all. Subcutaneous interferon gamma was administered to two patients to control infection but for a short course due to un-sustained availability and high cost of the drug. All patients were from consanguineous families, mainly from upper Egypt(13/18 patients) and four had positive family history. Three patients died out of fulminant infection soon after diagnosis. None of the patients did hematopoietic stem cell transplantation.

Conclusion: IL12RB1 mutation is elicited as the only mutation in our small cohort; however, more patients need to be screened to confirm this finding and determine other possible mutations causing MSMD in Egypt.