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SCIENTIFIC LETTER

Disseminated Tuberculosis in an Infant with IL-7 Receptor Deficiency

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To the Editor: A 5-mo-old girl was admitted to our hospital as she was suffering from pneumonia. Her respiratory status worsened rapidly to respiratory failure and non-invasive ventilation combined with anti-infection treatment and systemic tests were initiated immediately. The infant's immune index showed greatly diminished T-cell count but normal numbers of B-cells and natural killer (NK) cells with normal immunoglobulin levels. Moreover, the chest computed tomography (CT) showed extensive infiltrations with consolidations. Next-generation sequencing (NGS) of bronchoalveolar lavage fluid indicated the existence of tuberculosis (TB), supported by TB-DNA of the secretions (5.13×10^2 copies/ml). In addition, genetic analysis was performed for suspected congenital immunodeficiency, indicating amino acid changes in exon 1 of Interleukin-7 receptor α (IL-7RA) gene (c.37delT) from her mother and in exon 4 of IL-7RA (c.361dupA) from her father, both of them leading to frameshift mutations. Thus, severe combined immunodeficiency disease (SCID) due to IL-7RA gene defects complicated with TB infection was diagnosed and anti-TB agents combined with supportive therapy were prescribed. However, during the next 1 y, several infections involving respiratory and digestive systems and recurrent suppuration from her BCG scar were observed. At the age of 18 mo, severe disseminated TB infection causing hepatosplenomegaly, peritoneal effusion, tubercular meningitis and uncontrolled fever threatened her life and eventually her parents dropped.

IL-7 receptor deficiency abolishes T cell development and function ($T^-B^+NK^+$), resulting in SCID within the first six months of life [1]. Due to the lack of adaptive immunity, affected patients suffer from severe, persistent infections, often with opportunistic pathogens, and generally die in infancy if hematopoietic stem cell transplantation (HSCT) is not performed [2], as shown in this child. Newborn screening may aid in early recognition [3]. Our case reveals a novel mutation of IL-7RA gene and emphasizes that early diagnosis and treatment of SCID is critical to prevent mortality, minimize morbidity and improve quality of life.

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Declarations

Conflict of Interest None.

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