Tumor Necrosis Factor Receptor-associated Periodic Syndrome Mimicking Systemic Juvenile Idiopathic Arthritis

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Background: We report two cases of tumor necrosis factor receptor-associated periodic syndrome (TRAPS) in patients in whom systemic juvenile idiopathic arthritis (JIA) had initially been diagnosed or suspected. One patient, given a diagnosis of systemic JIA, was a 10-year-old boy who had presented with recurrent episodes of spike-fever, skin rash, arthritis, and myalgia. The other patient was his 7-year-old sister, who presented with similar symptoms and was suspected of having systemic JIA.

Methods: Serum levels of soluble tumor necrosis factor receptor super family 1A (TNFRSF1A), TNF-alpha, Interleukin (IL) -6, and C-reactive protein (CRP) were measured in two siblings and JIA patients. In addition, DNA sequencing of the TNFRSF1A gene in two siblings was also performed.

Results: A detailed family history showed that their mother had an episode of recurrent fever, arthritis, and myalgia with an increased serum CRP after the delivery of a daughter. Both siblings had serum levels of soluble TNFRSF1A that were below the normal reference range, and that did not reach a level corresponding to that of systemic JIA. On TNFRSF1A gene analysis, a single missense mutation resulting in C30Y was found in both siblings.

Conclusions: Based on the clinical features and the TNFRSF1A mutation, both siblings were given a diagnosis of TRAPS. The serum levels of soluble TNFRSF1A, measured along with the CRP level, may be a useful screening marker for differentiating TRAPS from systemic JIA.

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