Genetic diagnosis of Systemic Lupus Erythematosus complicating hyper IgE syndrome

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Case Study: The diverse genetic or environmental factors are associated with the development of Systemic Lupus Erythematosus (SLE). SLE complicating hyper IgE syndrome has been rarely reported. The case is the single child from non-consanguineous Korean parents. He suffered from the recurrent serious infections since his neonatal period. At 2 years of age, he developed acute renal insufficiency associated with a prolonged febrile illness. Severe ecchymosis was noted on his face along with reddish skin rashes on his abdomen and legs. Immunologic investigation revealed he had low complements with a positive anti-nuclear antibody. Renal histology was consistent with focal lupus nephritis, class III. Along with the immunosuppressive therapy, however, he experienced recurrent febrile episodes (6 admissions within 6 months). With a suspicion of primary immunodeficiency underlying SLE, targeted exome sequencing was done, and it revealed a missense mutation in the DOCK8 gene and possibility of exonic deletion as well. The missense mutation was paternally inherited and the deletion of exon 1-8 was from his mother confirmed by multiple ligation probe amplification analysis. This report indicates that a genetic factor such as primary immunodeficiency should be considered in children experiencing severe SLE course. Considering the primary immunodeficiency genetically confirmed, the benefit of bone marrow transplantation can be expected.