



ORAL PRESENTATION

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# Familial chilblain lupus caused by an activating mutation in STING

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Familial chilblain lupus is a monogenic form of cutaneous lupus erythematosus characterized by cold-induced cutaneous lesions at acral location. It is caused by loss-of-function mutations in the nucleic acid metabolizing enzymes TREX1 or SAMHD1. Gain-of-function mutations in STING (stimulator of Interferon genes) have been described in an infancy-onset autoinflammatory syndrome with fever, inflammatory cutaneous lesions and interstitial lung disease.

Here we report on a family with dominant chilblain lupus over 4 generations. Affected family members presented with acral inflammatory and partially necrotizing lesions beginning in early childhood. In some cases, low-titered ANAs and immune complexes were detectable. The family tested negative for TREX1 or SAMHD1 mutations. Exome sequencing revealed a heterozygous STING mutation segregating with chilblain lupus in the family. The mutation affects a highly conserved residue within the STING dimer interface and is predicted to be pathogenic. Quantitative RT-PCR analysis showed an increased expression of IFN-stimulated genes in blood cells of affected family members suggesting that the identified mutation has an activating effect on type I IFN signaling. Taken together, our findings demonstrate that gain-of-function mutations in STING can cause familial chilblain lupus and expand the spectrum of type I interferonopathies.

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