Background: Monogenic auto-inflammatory conditions are increasingly described with variable features. We present a case of an ulcerating scarring skin condition with erysipelas-like flare episodes, remaining undiagnosed until aged eleven, when genetic testing confirmed SAVI.

Methods: The patient’s health visitor identified widespread skin motting in the first weeks of life. At one month old, his mother described livedo reticularis on his lower legs, feet and arms, which then ulcerated. The ulcers gradually healed, however in some areas scarring remained, particularly on the back of his legs. He was systematically well during these episodes including the absence of fever. However, his mother commented that his skin would flare at the time of intermittent illness. By one year of age, he complained of digital and abdominal pain during episodes. He also suffered from rhinorrhoea. A skin biopsy reported only mild capillary dilatation and blood tests revealed only a mildly elevated ESR. The initial working diagnosis was cutis marmorata telangiectatica congenita. By age nine, the livedoid rash persisted over his limbs with full resolution elsewhere. There were chronic skin changes on the ear helices with ulceration, scarring and mild cartilage loss. Episodes of painful erythematous swelling over his ear helices and over the sides and dorsum of his feet and toes occurred every few months with subsequent ulceration. Multiple medications were trialed including nifedipine, azithromycin, steroid creams and ibuprofen gel. At age eleven, these episodes had progressed with associated arthralgia of MCPs/MTPs and reduced mobility. A full systemic work up did not reveal any additional organ involvement. Following multiple negative skin biopsies, a biopsy to deep fat reported polyarteritis nodosa (PAN) like vasculitis with panniculitis, prompting use of systemic steroid medication which rapidly resolved the flares. Six months of sub-cutaneous methotrexate (15mg/m2) weekly, as a steroid sparing agent, failed to control disease flares and mycophenolate mofetil has been commenced. Parents consented to a genetic panel for autoinflammatory/vasculitic conditions. This identified a recognised pathogenic mutation in the STING gene known to alter type 1 interferon signalling. The result confirms a diagnosis of SAVI. The genetic mutation found was heterozygous for c.817G>A p.(Cys272Tyr) like pathogenic variant in the TMEM173 gene. The same variant has been reported in one other case in the literature.

Results: SAVI is a rare genetic autoinflammatory condition characterised by systemic inflammation and small vessel vasculopathy in infancy. This can result in severe skin, lung and joint disease. The prevalence is unknown. Only a few cases have been reported in the literature.

Conclusion: This case highlights the clinical spectrum of SAVI including early onset cutaneous vasculitis affecting the extremities that often progresses to chronic skin changes, and joint disease. Previous reported cases typically describe fever, failure to thrive and respiratory involvement which are absent in our patient.