

# Anifrolumab Treatment in Monogenic Pediatric Lupus: A Case Report

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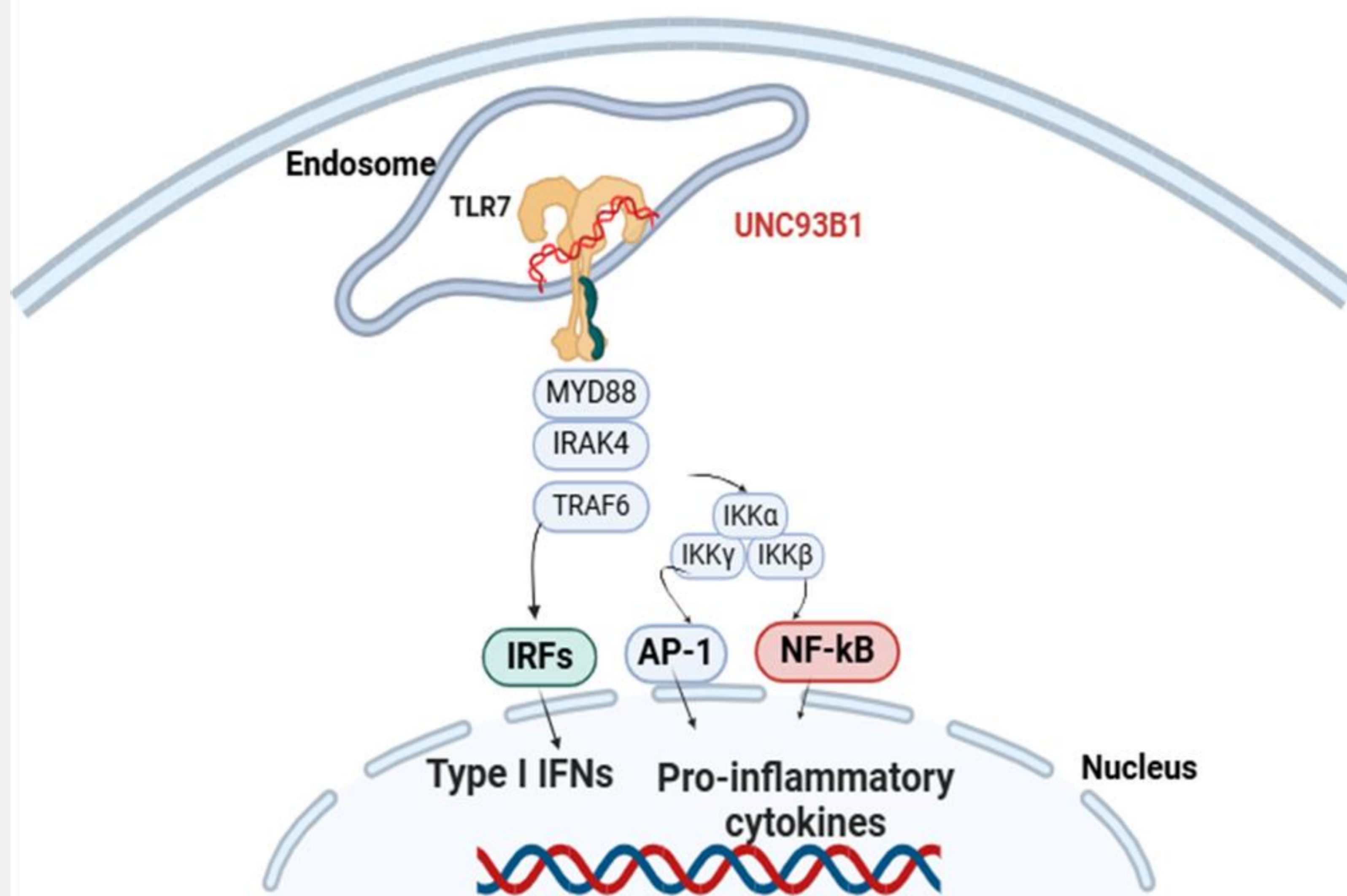
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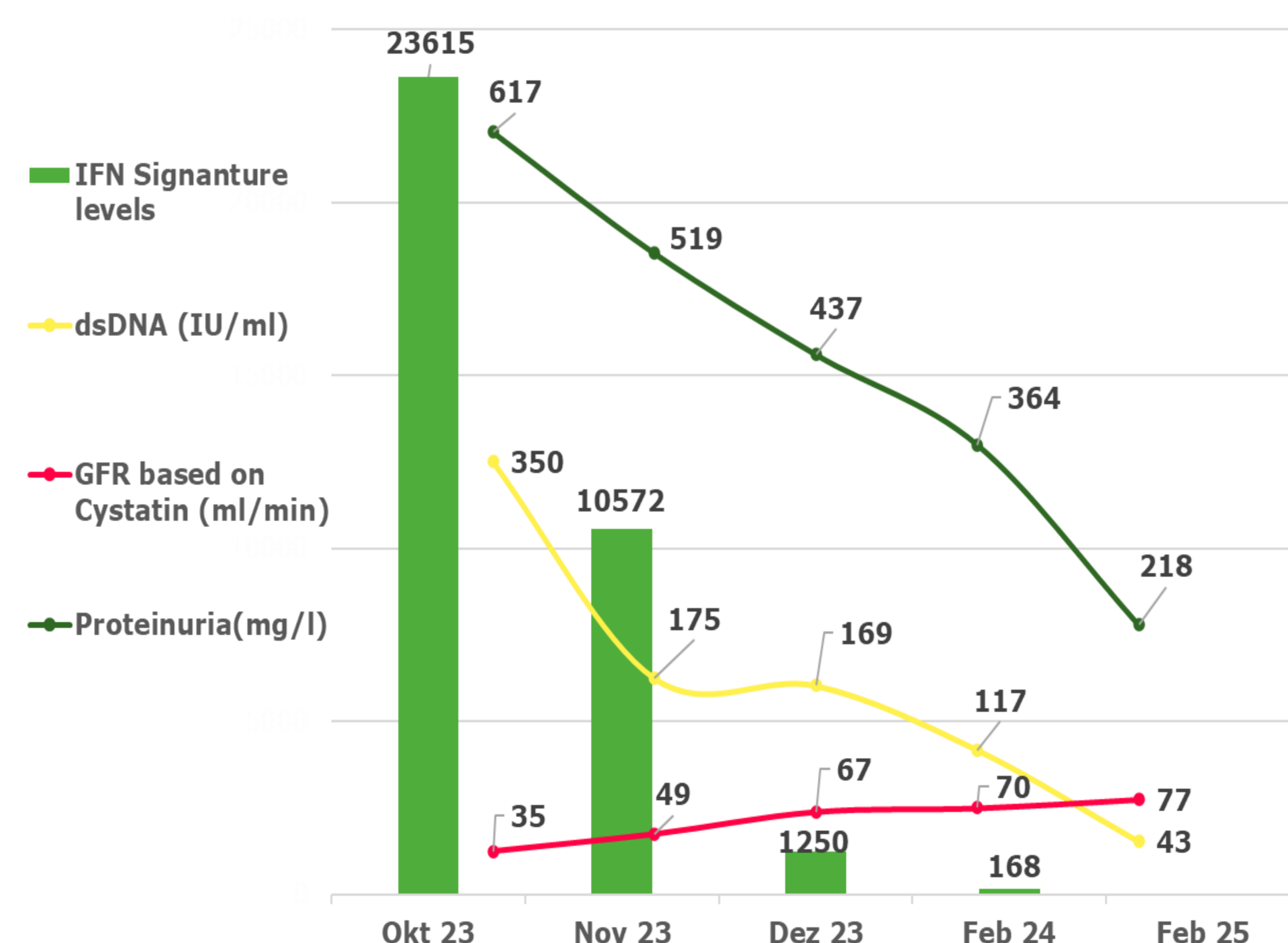
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## Background

**Variants in UNC93B1 lead to selective hyperactivation of the toll-like receptor TLR7, resulting in the incorrect recognition of the body's own RNA. This triggers uncontrolled production of type I interferons and inflammatory processes, ultimately causing a form of monogenic systemic lupus erythematosus (SLE).**



**Figure 1.** TLR7 signaling pathway (created in BioRender.com)



**Figure 2.** Impact of Anifrolumab Therapy on IFN Signature, dsDNA, GFR and B cells levels

## Case Report

We have recently reported the case of a 15-year-old boy with **SLE caused by a homozygous variant in UNC93B1 (c.275A>G, p.E92G)**, presenting with severe hematologic, immunologic, neurologic, and nephrological manifestations. **Multiple treatments** (corticosteroids, hydroxychloroquine, mycophenolate mofetil (MMF), cyclosporine, rituximab, ruxolitinib) failed to achieve remission. In October 2023, treatment with **anifrolumab**, a monoclonal antibody that **inhibits the type I interferon receptor (IFNAR)**, was initiated. Remarkably, **renal function stabilized, B cell levels increased, complement factors normalized**, C-reactive protein (CRP) levels decreased, and the SLE Disease Activity Index (SLEDAI) significantly reduced. **The interferon signature improved** but remained elevated. Serious Adverse Event (SAEs), (temporary headaches) were reported, but **no significant severe adverse effects were observed**.

## Conclusion

In this case of monogenic SLE, Anifrolumab appeared to present **a personalized targeted therapeutical approach**. Further studies of patients with monogenic SLE, are necessary to definitively confirm the long-term safety and efficacy of Anifrolumab in this setting.

## Literatur

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