JAK Inhibition for treatment of psoriatic arthritis in Down syndrome

People with Down syndrome (DS), the condition caused by trisomy 21 (T21), present with increased prevalence of several autoimmune conditions relative to the general population. This includes celiac disease, autoimmune skin conditions, and arthropathies. While it is now well established that T21 causes hyperactivation of interferon and downstream Janus kinase (JAK) signaling, the therapeutic value of this observation remains to be defined. Here, we describe the first reported case of an individual with DS who was treated with the JAK inhibitor tofacitinib as a first-line therapy for severely debilitating psoriatic arthritis (PsA). This resulted in near complete resolution of the patient's clinical symptoms, psoriasis, and normalization of inflammatory markers.