RA or imitator?

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H syndrome is an autosomal recessive multisystemic disease with a very low prevalence rate, characterized by indurated cutaneous hyperpigmentation, hypertrichosis, and various systemic manifestations.

A 37-year-old woman was referred to the rheumatology department with a prediagnosis of rheumatoid arthritis due to deformity in the hands and feet and mild sensorineural hearing loss since the age of 17. On examination, the patient’s height was lower than average (147 cm [SD –2.9]). Well-defined, bilateral, hyperpigmented, reddish indurated plaques presented over the medial aspects of the thighs (Figure 1). Also, there were telangiectasias between both breasts. The patient also exhibited camptodactyly, specifically 2nd to 5th proximal interphalangeal (PIP) joint flexion deformity (Figure 2A), and a severe case of hallux valgus (Figure 2B). Moreover, the patient had splenomegaly on abdominal examination. Gene analysis was done on our patient on suspicion of a genetic disease. Whole genome sequencing showed a mutation in \textit{SLC29A3}, confirming H syndrome.\textsuperscript{1,2}

REFERENCES