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CASE IMAGE
Pediatric Rheumatology

Camptodactyly and rash: Common clues for Blau syndrome and H syndrome

Kamptodaktili ve döküntü: Blau sendromu ve H sendromunun ortak ipuçları

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Camptodactyly is defined as permanent flexion contracture of the fingers at the proximal interphalangeal joints. It may be an isolated anomaly, may be a component of a genetic syndrome, or caused by a systemic disease (1).

CASE 1 – A 9-year-old boy was admitted to the clinic with complaints of rash and swellings in the joints. The rash started when the child was 6 months old on the distal parts of the arms. Initially, it was slightly erythematous macular rash and later involved nearly all of the body and became scaly. He had the diagnosis of atopic dermatitis during infancy and later on was diagnosed as a mild form of ichthyosis vulgaris. Around 1 year of age, the joints of the wrist and ankle were swollen. He was diagnosed as juvenile idiopathic arthritis and treated with methotrexate at the age of 3 for 3 years. The mother mentioned some improvement in the rash, but the joints were the same and also fingers were involved for the past 2 years. On physical examination, there was boggy synovitis on bilateral wrists and ankle joints. Bilateral camptodactyly was evident in the fingers (Fig. 1a). Furthermore, there was scaly tan-colored rash on the abdomen, arms, and legs (Fig. 1b). The ophthalmologic examination did not show any sign of uveitis. Maternal grandmother had similar joint problems and rashes and died at

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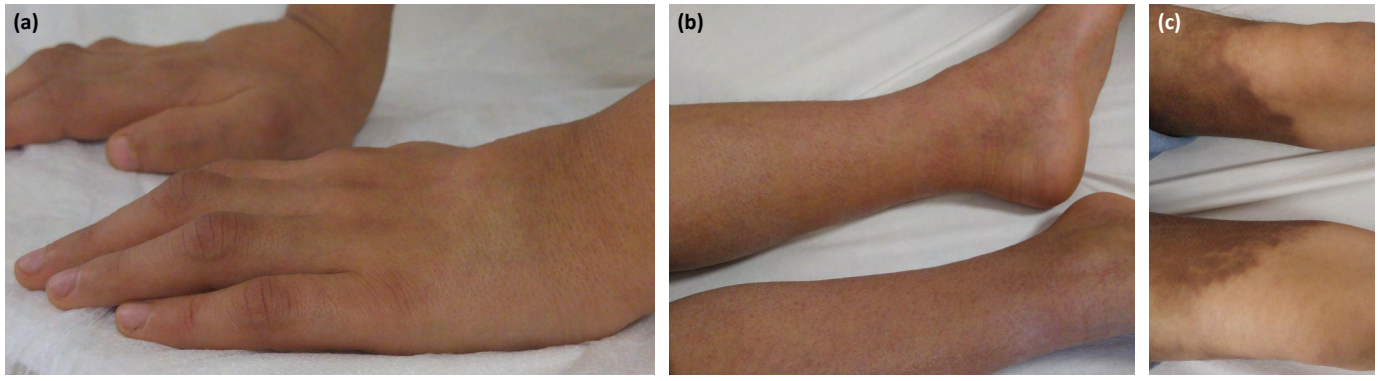

Figure 1. (a) The view of the camptodactyly of the Blau syndrome. **(b)** The view of the ichthyosiform rash on the lower legs of Blau syndrome. **(c)** The view of the hyperpigmented and hypertrophic rash of the H syndrome on the upper legs.

the age of 38 secondary to cardiac problems. The phenotype was compatible with Blau syndrome and genetic analysis revealed a heterozygous mutation in the *NOD2* gene (exon 4, c.1001G>T, p.Arg334Leu, p.R334L). Methotrexate and prednisolone were started. The rash resolved but did not lead to any change in arthritis. Adalimumab was added to treatment on the 6th month that led to resolution of arthritis.

CASE 2 – A 13.5-year-old-girl was referred to pediatric rheumatology with complaints of rash and inability to open fingers fully for 1 year with the provisional diagnoses of juvenile idiopathic arthritis and localized scleroderma. She had the diagnosis of insulin-dependent diabetes mellitus for the past 6 months and was also evaluated for delayed puberty. On physical examination, she had large discrete hypertrichotic, hyperpigmented, and indurated plaques on the legs and lower back (Fig. 1c). She had prominent bilateral camptodactyly on all fingers. Combining hyperglycemia, hyperpigmentation, hypertrichosis, and camptodactyly made us consider H syndrome. Genetic analysis revealed a homozygous deletion in exon 3 of the SLC29A3 gene.

Blau syndrome is a multisystemic granulomatous disorder that has the onset before the age of 4 and caused by mutations in the NOD2 gene. The disease is characterized by the triad of polyarthritis, rash, and uveitis. The familial autosomal dominant form is called Blau syndrome and sporadic form is called early-onset sarcoidosis (2–4). H syndrome is a genetic genodermatosis caused by mutations in the SLC29A3 gene. Some of the clinical manifestations are hyperpigmentation, hypertrichosis, hyperglycemia, hearing loss, and hypogonadism (5). Camptodactyly may be a component of both disorders (1, 5).

In children presenting with multisystem involvement, one disease can explain the phenotype instead of labeling the patient with different coincidental diagnoses.

REFERENCES

1. Poline J, Bourrat E, Meinzer U. Camptodactlyly in pediatric practice: Blau syndrome. J Pediatr 2020;221:257–9.
2. Kisaarslan PAÇ, Sözeri B, Şahin N, Özdemİr Çiçek S, Gündüz Z, Demirkaya E, et al. Blau syndrome and early-onset sarcoidosis: A six case series and review of the literature. Arch Rheumatol 2019;35:117–27.
3. Wu D, Shen M. Two Chinese pedigrees of Blau syndrome with thirteen affected members. Clin Rheumatol 2018;37:265–70.
4. Çakan M, Keskindemirci G, Aydoğmuş Ç, Akı H, Hatipoğlu N, Kıyak A, et al. Coexistence of early onset sarcoidosis and partial interferon-γ receptor 1 deficiency. Turk J Pediatr 2016;58:545–9.
5. Tekin B, Atay Z, Ergun T, Can M, Tuney D, Babay S, et al. H syndrome: A multifaceted histiocytic disorder with hyperpigmentation and hypertrichosis. Acta Derm Venereol 2015;95:1021–3.