“PEARLY PAPULES IN A PATIENT WITH DOWN SYNDROME: A CASE OF MILIA-LIKE IDIOPATHIC CALCINOSIS CUTIS”

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KEY WORDS

milia-like, idiopathic, calcinosis cutis, Down Syndrome

INTRODUCTION

Calcinosis cutis is a condition characterized by the abnormal deposition of insoluble calcium salts within the skin and subcutaneous tissues. Five major forms have been described: dystrophic, metastatic, idiopathic, calciphylaxis and iatrogenic calcinosis cutis.

Milia-like idiopathic calcinosis cutis (MLICC) is a rare variant of calcinosis cutis that mostly occurs in childhood, it is characterized by firm, white-yellowish papules measuring 1–4 mm that resemble milia. It predominantly affects the dorsal hands and feet.There is a strong clinical link; aproximately two-thirds of MLICC occur in patients with Down Syndrome. This association has been linked with premature aging and increase intracellular calcium levels in fibroblasts of affected patients. Unlike other forms of calcinocis cutis, MLICC is not associated with an identifiable metabolic disorder and tissue damage so these lesions tend to resolve spontaneously in adulthood without scarring, as well serum calcium and phosphate levels remain within normal ranges.

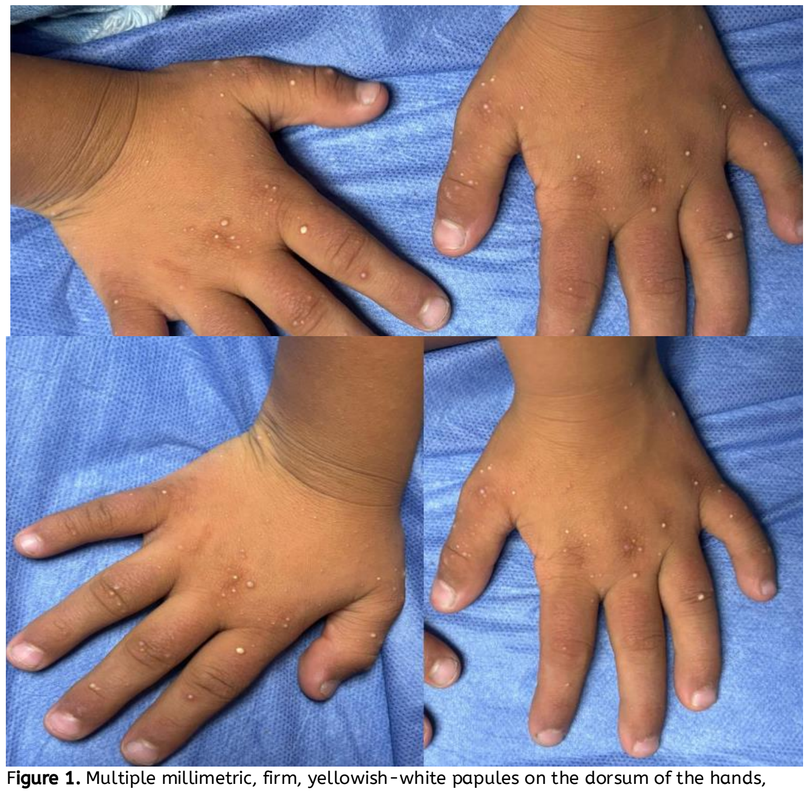

Figure 1. Multiple millimetric, firm, yellowish-white papules on the dorsum of the hands, consistent with MICC in a patient with Down syndrome.

CASE PRESENTATION

A 7-year-old female with Down syndrome presented with a 3-month history of asymptomatic skin lesions. She was initially diagnosed by a pediatrician with molluscum contagiosum; however, due to concerns regarding lesion spread, she was referred to pediatric dermatology.

General and systemic examinations were within normal limits. Physical examination revealed a monomorphic, symmetric dermatosis involving the dorsal hands, composed of multiple well-defined, smooth, firm, white-yellowish papules **(Figure 1)**.

Biochemical studies, including serum calcium, phosphate, parathyroid hormone and vitamin D were within normal range. Based on the characteristic clinical morphology, absence of symptoms and lab results, the patient was diagnosed with milia-like idiopathic calcinocis cutis and a conservative approach was adopted given the tipycally benign course.

DISCUSSION

The term idiopathic calcinosis cutis is reserved for cutaneous calcification occurring in the abscense of underlying tissue injury or a demonstrable systemic or metabolic disorder, with normal serum calcium and phosphate levels.Patients with Down syndrome have a predisposition to ectopic calcifications, including cutaneous involvement. Although pathogenesis remains unclear, lesions are thought to result from transepidermal elimination of calcium deposits. Diagnosis is mainly clinical; histopathology, when performed, shows circumscribed calcium deposits within the dermis surrounded by thickened collagen fibers and ocasionally multinucleated giant cells.

The differential diagnosis include molluscum contagiosum, common warts, cysts and other forms of calcinocis cutis.

Accurate diagnosis is essential to avoid unnecessary and potentially harmful treatments. Although spontaneous resolution is common, topical tretinoin and CO_2 laser have been reported in selected cases.

CONCLUSION

Although it is a rare and self-limited condition, MLICC should be considered in patients with Down syndrome. Early recognition helps prevent unnecessary, painful, and costly interventions, while appropriate counseling reassures caregivers about its benign and favorable course.

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