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Mal De Meleda Syndrome With Poikiloderma And Bony Changes: A Case Report.

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Abstract*: Mal de Meleda is a rare type of autosomal recessive genodermatosis characterized by presence of transgradient palmoplantar keratoderma, nail changes, scleroatrophy, perioral erythema and angular cheilitis since birth or early childhood. We report a 46 year old female born out of non-consanguinous marriage having palmoplantar keratoderma, flexion deformity due to contracture of fingers, scleroatrophy and conical tapering of fingers. Extensive thickening with poikiloderma and bony changes are new features observed in this patient of Mal de Meleda syndrome.*
Key words: *Keratoderma, Poikiloderma, Bony changes*

I. Introduction

Hereditary palmoplantar keratodermas are a heterogenous group of disorders characterized by hyperkeratosis of palms and soles since birth or childhood. They may be either localized or diffuse, restricted to palms and soles or may extend to dorsal areas (transgradiens). Transgradient palmoplantar keratodermas include variants like Loricrin keratoderma, Vohwinkel syndrome, Syberts keratoderma, Huriez syndrome, Olmsteds syndrome and Mal de Meleda syndrome. Mal de Meleda syndrome is a rare autosomal recessive condition named after Croatian island of Meleda [1] which shows presence of palmoplantar keratoderma, angular cheilitis and knuckle pads on interphalangeal joints. Although contractures [2] and pseudosclerodermatous appearances are known to occur secondary to dense hyperkeratosis, poikiloderma and bony changes have not been reported yet.

II. Case History

A 46 year old female born out of non consanguinous marriage presented to the outpatient department with history of thickening of skin of palms and soles since birth. Thickening progressed slowly over the years gradually spreading to the dorsum of both the hands and feet. Over last 10 years there has been massive thickening of palms and soles causing inability to work. There was no history suggestive of photosensitivity, growth retardation, mental retardation, exposure to chemicals and long term topical therapies like corticosteroids. No other family members were affected.

On examination there was bilateral diffuse keratoderma on palms and soles along glove and stocking distribution**.** Both the soles showed thickening along with scaling extending up to ankle [Figure 1]. Dorsum of both feet showed thickening, atrophy, scaling and dyschromia at places [Figure 2]. Both the palms were thickened with loss of dermatoglyphics. The keratoderma extended to the dorsum of hand and well beyond the wrist [Figure 3]. Left hand had atrophic parchment like skin on the dorsum along with conical tapering of fingers [Figure 4]. Right hand showed more thickening with flexion deformity of the fingers [Figure 5]. Nails were dystrophic, thickened and showed pigmentation with hypercurvature. There were no digital constrictions or loss of digits. Mucous membranes, skin on perioral areas, knees, elbows were normal.

Routine investigations were within normal limits. Skin bipopsy was done from lateral border of left hand which showed massive hyperkeratosis, acanthosis, accentuated granular layer and orthohyperkeratosis. X-ray feet did not reveal any abnormality. X ray of both the hands showed radio ulnar subluxation, periarticular osteoporosis, diffuse osteopenia and carpo-metacarpal joints involvement in the form of reduced joint spaces [Figure 6]. Thinning of middle and distal phalanges along with interphalangeal joint deformity without significant joint destruction was also evident [Figure 7].

III. Discussion

Heriditary palmoplantar keratodermas are often difficult to distinguish and diagnosis is usually made on the basis of clinical features, distribution of keratoderma , presence or absence of associated anomalies and elucidation of underlying genetic defect.

The present case was diagnosed as having Mal de Meleda syndrome on the basis of following observations. Patient had progressive palmoplantar keratoderma since birth with no tendency to improve with age. Presence of glove and stocking type of palmoplantar keratoderma in this patient is a predominant finding of Mal de Meleda syndrome [3, 4]. Nails showed subungual hyperkeratosis with dystrophy and curving. Dorsum of the hand showed scleroatrophy and parchment like skin with conical tapering of fingertips. Presence of contracture of fingers causing mutilation and disability. Although perioral erythema, constriction bands are frequent in Mal de Meleda it was not seen in this patient.

There is individual variation in clinical manifestations of Mal de Meleda which should be differentiated from other palmoplantar keratoderma syndromes. Grithers disease differs from Mal de Meleda by late age of onset (8-10 years), tendency to improve spontaneously with age and at times sparing of palms and soles. Conical tapering of finger tips as seen in Mal de Meleda is not seen in Greithers disease [5]. Huriez syndrome a rare autosomal dominant transgradient palmoplantar keratoderma characterized by congenital scleroatrophy of distal extremities, mild keratoderma of palms and to lesser extent of soles and very high risk of development of aggressive squamous cell carcinomas of skin[2]. Another prominent feature of Huriez syndrome is invariable family history [6] which was lacking in our patient. Although severe cicatrizing transgradient palmoplantar keratoderma as seen in this patient has also been reported in Olmsteds syndrome the absence of spontaneous amputation of digits, follicular keratoses on skin and oral mucosa differentiates it from Olmsteds syndrome. Acral keratoderma with mutilation as seen in our case is more often noticed in Vohwinkel however, absence of starfish keratosis, sensorineural deafness, circumferential bands and auto amputation of fingers [7] rules out this possibility. Camisa syndrome is a variant of Vohwinkel where there is absence of deafness but these patients have icthyosis [8] which was not evident in our case. Papillon Lefevre syndrome is characterized by palmoplantar hyperkeratosis, severe periodontitis and premature loss of teeth [1]. Presence of normal teeth and absence of periodontosis excludes the possibility of Papillon Lefevre syndrome. Recently Kharge P et al has reported occurance of poikiloderma in Huriez syndrome [9]. None of the authors mention the presence of poikiloderma in Mal de Maleda syndrome. Moreover very few reports of bony changes occurring in palmoplantar keratoderma have been reported in the literature. Kaviggeo L [10] et al has reported changes like destruction of distal phalanges, atrophy of proximal phalanges and atrophy of metatarsals in a patient with mutilating palmoplantar keratoderma. None of the reports mention the presence of bony changes in Mal de Meleda syndrome. The presence of radiological findings like radio ulnar subluxation, periarticular osteoporosis, diffuse osteopenia and subluxation indicates changes secondary to chronic disability and subsequent disuse of hand.

What's new ? : Non familial case of Mal de Meleda syndrome with massive palmoplantar keratoderma, poikiloderma and bony changes.

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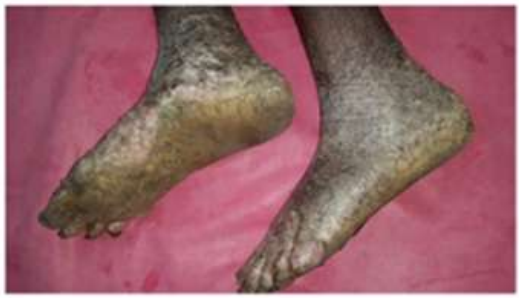

Figure-1 Soles showing diffuse thickening along with and scaling extending up to ankle.

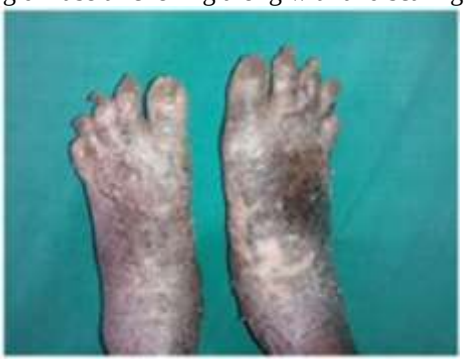

Figure-2 Dorsum of feet showing poikiloderma.

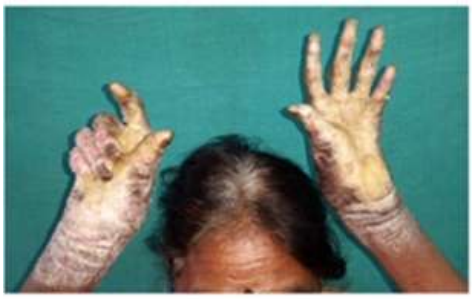

Figure-3 The keratoderma extending to the dorsum of hand and beyond the wrist.

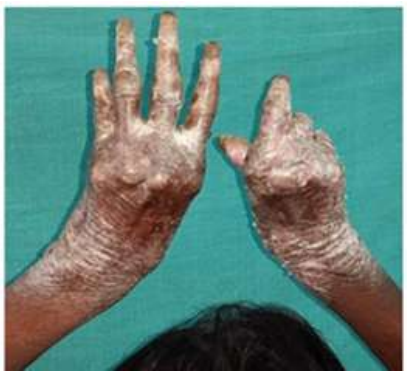

Figure-4 Atrophic parchment like skin on the dorsum of hand along with conical tapering of fingers.

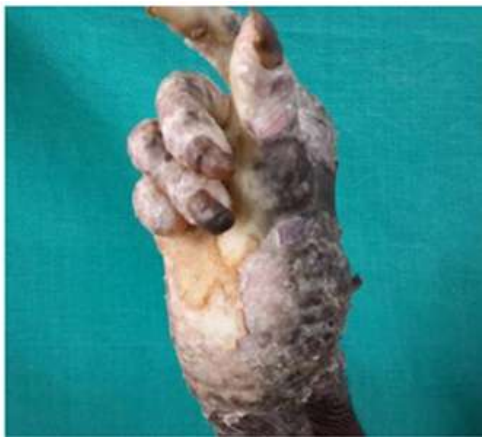

Figure-5 Flexion deformity of the fingers .

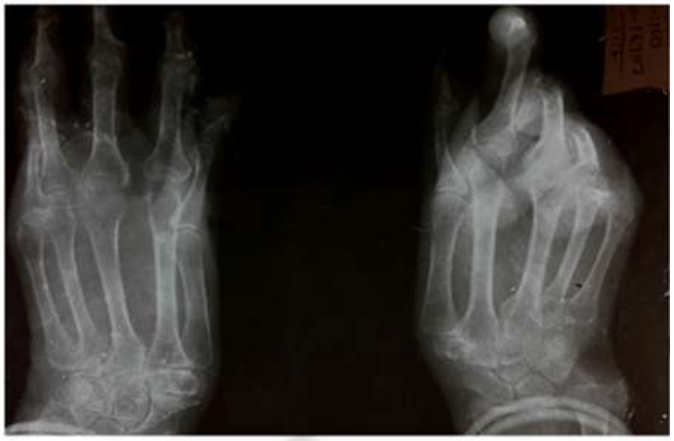

Figure-6 X ray of the hands showed radio ulnar subluxation, periarticular osteoporosis and diffuse osteopenia

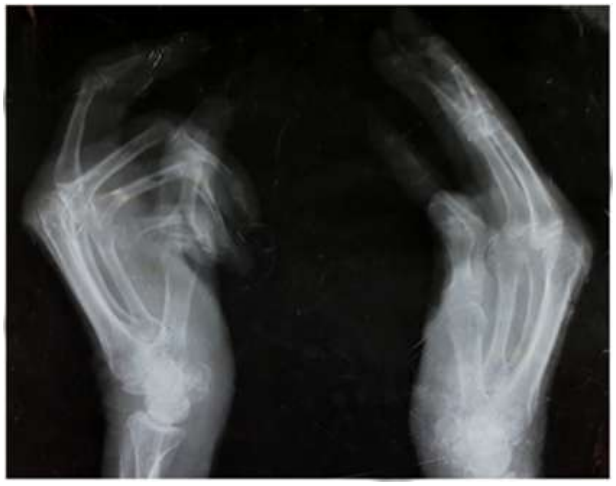

Figure-7 Thinning of middle and distal phalynges along with interphalyngeal joint deformity.