Journal of Pakistan Association of Dermatologists.2023;**33(1)**:335-338.

Case Report

Mid-face Toddler Excoriation Syndrome (MiTES)

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Abstract Genetically heterogeneous group of sensory neuropathies have variable clinical presentation with insensitivity to pain as a prime feature. Chronic scarring with midface erosions mostly found in infants and toddlers and may be the only primary manifestation of chronic insensitivity to pain. In these cases, parents report that lesions are self-induced but, doctors attribute fabricated illness by parents. Thorough clinical and laboratory workup has to be done to distinguish between sensory neuropathies, Factious illness and neurometabolic disease. Midface toddler excoriation syndrome is a recently described entity, clinically characterized by excoriations and sequelae of long-term scratching, over mid-face in toddlers. In this case we have proposed the recognition of this entity in young adult, would encourage further scientific reports, and help to elucidate the cause.

Key words
Excoriation, neuropathy, midface.

Introduction

Heterogeneous sensory neuropathies may present with varied clinical presentation. Among which insensitivity to pain is one of the features in this group of genetic diseases.

Hereditary sensory-autonomic neuropathy (HSAN) type VIII is caused by pathogenic variations in the PRDM12 gene. HSAN clinically presents as partial or global insensitivity to pain, recurrent infections, temperature dysregulation and absence of corneal reflex with altered tears production.[1] However they may present with various other manifestations which include severe soft tissue injuries, self-mutilation, dental caries, premature loss of teeth, submucosal abscesses and mandibular osteomyelitis.[2]

Facial scarring and diabetic foot-like ulcers may be the manifestation of mild, localized phenotype of HSAN type VIII in few cases.[1] In recent times, a new terminology Mid-face Toddler Excoriation Syndrome (MiTES) has been suggested to depict this mild phenotypic variant. These patients clinically manifest as self-inflicted excoriations, ulcerations, hyperpigmented scars on the midface especially on forehead, nose and nasolabial folds during early infancy.[3] Here we describe a rare case of midline excoriation in an adult patient which is first of its kind to the best of our knowledge.

Case report

A 20 years old man presented with skin lesions on the face for 3 years. He reported persistent intense picking and scratching of central face without any obvious pain or distress. He was born at term to a non-consanguineous marriage

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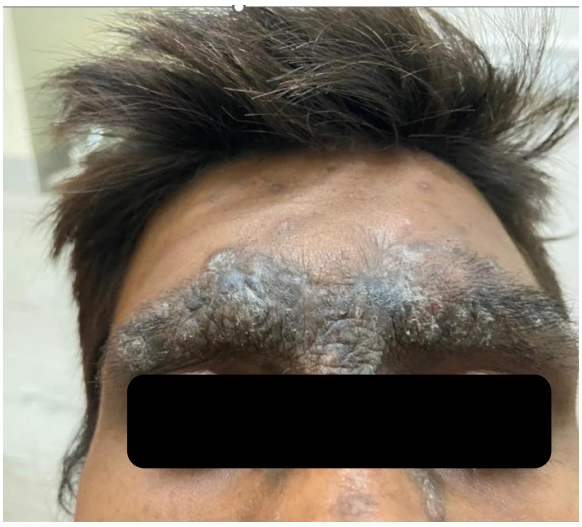

Figure 1 Post-inflammatory hyperpigmented lichenified plaques and atrophic scars confined to the midline of face with madarosis.

of Indian parents with no relevant family or medical history. His brother reported that he scratched the lesions repeatedly. He did not observe any blisters or photosensitivity. No other family members had similar history.

Skin examination revealed lesions confined to the midline of face (**Figure 1**). There were post-inflammatory hyperpigmented lichenified plaques and atrophic scars, madarosis and excoriations on the forehead and medial aspect of nose. Physical examination was otherwise normal.

Investigations like Complete blood counts, antinuclear antibodies (ANA), serum uric acid, ammonia, lactate, MRI brain, nerve conduction studies and EMG were normal. Biopsy from the lesion showed acanthosis, hyperkeratosis and increased dermal collagenization with a sparse lymphocytic infiltrate which was consistent with lichen simplex chronicus. There were no histopathological evidence of granulomas or vasculitis.

Psychiatric evaluation was done to rule out fabricated and induced illness (FII). Patient reports intense urge to itch and compulsion to scratch which was limited to midline of face. He had disturbed sleep causing major concern to him due to itch. He denied any stressors/ stressful life events/ anxiety and depression symptoms. The diagnosis of obsessive compulsive and related disorder (skin picking disorder) was made according to DSM 5. Personality assessment using Eyesanks personality questionnaire revealed more of neurotiscism traits. Intelligent Quotient assessment using Binet Kamat test was 108 which is normal.

We had started clomipramine 25 mg gradually increased to 50 mg over a period of 2 weeks along with Fluoxemine 50 mg, topical emollients and steroids. Treatment produced limited improvement in the lesions but, sleep was optimized and impulse to scratch had reduced over three months.

Discussion

The anatomical distribution and age of onset of this condition reflects as that of MiTES. It is characterized bilateral symmetrical self-inflicted deep, scars located on mid-face due to chronic habitual scratching since infancy. The autosomal recessive disorder HSAN VIII due to mutation in gene PRDM12, clinically characterised by congenital insensitivity to pain with ulceration on extremities. MiTES is an extension of the milder phenotypic spectrum of Bi-allelic mutations in PRDM12 gene. It is distinguished by self-inflicted severe mutilating lesions mid-face and extremities with no significant evidence of generalised pain insensitivity.[4] Hence, MiTES is a genetically heterogeneous entity, resembles HSAN VIII clinically and etiologically but lacks ulceration of the acral parts.[5]

Clinical history and examination were not consistent with ecthyma, leishmaniasis, lupus vulgaris, anthrax or tropical ulcer. Skin fragility

and facial scarring can be features of erythropoietic protoporphyria (EPP), but it begins in early childhood with subtle presentations. There was no histological evidence for granulomatous disease, lupus, vasculitis or pyoderma. None of the above conditions explained lichenificaton, madarosis and scarring. The skin lesions look traumatic but patient admits that the damage is self-inflicted due to irresistible urge to scratch. This case typifies a condition that poses a significant diagnostic dilemma.

He had significant facial scarring with madarosis though scratching may be witnessed but the damage appears disproportionate. These lesions in an adult would suggest dermatitis artefacta (DA). It is characterized by deliberately self-inflicted skin lesions presenting as skin disease.[6] However, bilateral symmetrical distribution of lesion on forehead makes DA unlikely. Trigeminal trophic syndrome (TTS) is a disorder of the elderly which was excluded in our case due to its distribution and age of presentation.

There was no evidence for sensory neuropathy due to leprosy or diabetes. Lesch-Nyhan syndrome which manifests as dystonia, mental retardation, aggressive, impulsive, self-mutilating behaviours due to excess uric acid production. It was eliminated as in this case plasma uric acid levels were normal. Hereditary insensitivity pain syndromes can sometimes lead to self-injury. However gross sensation in our patient was normal but we could not eliminate PRDM12 gene mutation causing this localized disorder of sensation. We arrived at the diagnosis of 'Midface Toddler Excoriation Syndrome' (MiTES) for this distinctive, localized midface skin condition occurring in context of mild congenital insensitivity to pain. It needs further elucidation to differentiate MiTES from other organic and FII. Management requires multidisciplinary approach with symptomatic treatment to reduce itch, promote healing with topical steroids, as well as psychological support and interventions as per case-to-case basis.[3]

Chronic lichenified excoriated lesions on the face are rarely seen in adults. Cutaneous differential diagnosis like blistering diseases, vasculitis, chronic infections and self-mutilation due to neurometabolic disease should be excluded. The above mentioned probable organic diseases has to be excluded before attributing such lesions to abuse, fabricated or induced illness.[3] The salient features of Munchausen syndrome by proxy (MBP) are proof of damage, repetitive behaviour, unconscious motivation and doctor shopping and confession by the perpetrator.[7] We would like to propose thorough psychological evaluation of these patients to exclude body focuses repetitive disorder, underlying depression or anxiety.[7] As in our case, lesions were self-inflicted, organic diseases and Fabricated or induced illness (FII) was excluded. The clinical pattern closely resembles MiTES. Due to paucity of literature among adults, we require further reports to clarify the etiopathogenesis and acknowledge the existence of this condition.

Conclusion

MiTES is a recently recognised condition, characterised by severe, chronic, scarring, self-inflicted, midface excoriations, commencing in infancy which resembles fabricated and induced illness. It as an autosomal recessive disorder of pain sensation which is probably genetically heterogeneous presentation. Bilateral distribution and localized to mid-face distinguish it from other causes of self-inflicted skin injuries. New understanding about MiTES, which can masquerade as factitious disease, will facilitate appropriate management.

References

1. Chen Y-C, Auer-Grumbach M, Matsukawa S, Zitzelsberger M, Themistocleous AC, Strom TM, *et al*. Transcriptional regulator PRDM12 is essential for human pain perception. *Nat Genet*. 2015;**47(8)**:803-8.
2. Elhennawy K, Reda S, Finke C, Graul-Neumann L, Jost-Brinkmann PG, Bartzela T. Oral manifestations, dental management, and a rare homozygous mutation of the PRDM12 gene in a boy with hereditary sensory and autonomic neuropathy type VIII: a case report and review of the literature. *J Med Case Rep*. 2017;**11(1)**:233.
3. Srinivas SM, Gowda VK, Owen CM, Moss C, Hiremagalore R. Midface toddler excoriation syndrome (MiTES): a new paediatric diagnosis. *Clin Exp Dermatol*. 2017;**42(1)**:68-71.
4. Moss C, Srinivas SM, Sarveswaran N, Nahorski M, Gowda VK, Browne FM, *et al*. Midface toddler excoriation syndrome (MiTES) can be caused by autosomal recessive biallelic mutations in a gene for congenital insensitivity to pain, PRDM12. *Br J Dermatol*. 2018;**179(5)**:1135-40.
5. Kumar P, Das A. Midface toddler excoriation syndrome. Indian J Dermatol Venereol Leprol 2021;87(4):528-9.
6. Moss C. Dermatitis artefacta in children and adolescents. *Paediatric Child Health*. 2015; **25(2)**:84-9.
7. T. Narang, S. Singh. Response to: Midface toddler excoriation syndrome (MiTES) can be caused by autosomal recessive biallelic mutations in a gene for congenital insensitivity to pain, PRDM12. *Br J Dermatol*. 2019;**180(2)**:431.