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RESEARCH ARTICLE

A RARE GENODERMATOSIS IN A PATIENT WITH NEUROPSYCHIATRIC DISORDER

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Abstract

Darierdisease(DD)is a rare congenital acantholytic disorder characterised by persistent eruptions of greasy, hyperkeratotic papules in seborrhoeic areas, extremities and rarely in intertriginous areas.Nail abnormalitiesand mucous membraneinvolvement also occurs. A neuropsychiatricdisordercan be present occasionally. We are reporting a rare case of Darier disease in a male child born in a consanguineous marriage coexisting with Attention Deficit Hyperactivity Disorder (ADHD).

Introduction:-

Darierdiseasealso known askeratosis follicularis, dyskeratosisfollicularis or Darier- White disease isarelativelyrareautosomal dominantskindisorder withabnormalkeratinizationand lossof epithelial adhesion. Althoughthiscondition has adominantgenetic inheritance,the report of sporadic cases is approximately 40–50%, presumablyofnew mutation or incompletepenetrance[1].The peak age of onset of Darier disease is between 6-20 years but may present in infants orold age. Apart from skin, nail and mucosal changes,Neuropsychiatric component with Mood disorders, including bipolar disorder, major depression, suicidal ideation and suicide attempts have been reported with high prevalence among individuals with DarierDisease[2].Darier disease has achronic course with fluctuations in disease severity. Treatment of Darier disease is challenging, avoidance of exacerbating factors, special care should be taken to avoid infections, emollients, soap substitutes and cotton clothing and sunscreens are advised. Oral retinoids and cyclosporine, physicalmodalities like dermabrasion,electrosurgical excision,CO2 laser, photodynamic therapy have limited role due to recurrence.

Case Report:

A eight year old boy born out of2nddegree consanguineous marriage diagnosed with Attention deficit hyperactivity disorder came with complaints of occasionally itchy skin lesions over the body since 3 years. History of summer exacerbation of the lesions was present. Noneofthe family members hadsimilar complaints. On examination, Multiple skincolouredto hyperpigmented keratotic papules were seen over both the cheeks, neck, chest, axillae, groins, both upper and lower limbs.Few tinypapules were noted over both the palms.Coarse pittingwas noted over left ring finger nail. Oral mucosa, genitals and hair were normal. Punch biopsy taken from apapule on right leg on histopathological examination showed a circumscribed focus of hyperkeratosis with acantholytic dyskeratosis and supra-basal cleft formation.Within thefocus the epidermis showed scattered acantholytic dyskeratotic cells (corpronds).The stratumcorneum shows a column of parakeratotic dyskeratotic cells (grains). Underlying dermisshows sparse superficial perivascular lymphocytic infiltrate.Mild papillomatosis is present, confirming the clinical diagnosis of Darier disease.Thepatient was advised oral antihistamines,topicalretinoid and urea containing creams, emollients and oral vitamin A.

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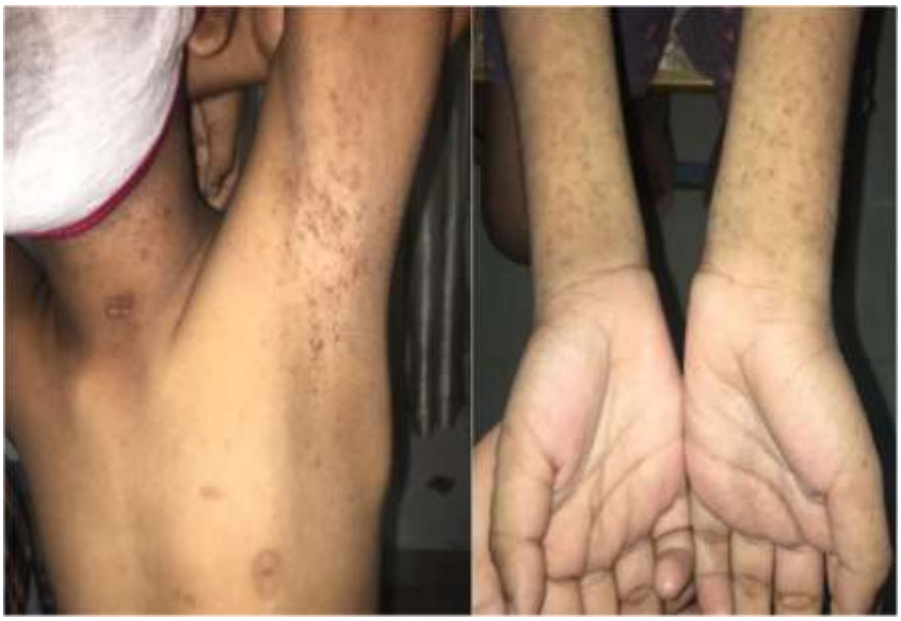


Figure 1:- Hyperkeratotic papules over the neck, axilla and forearms.

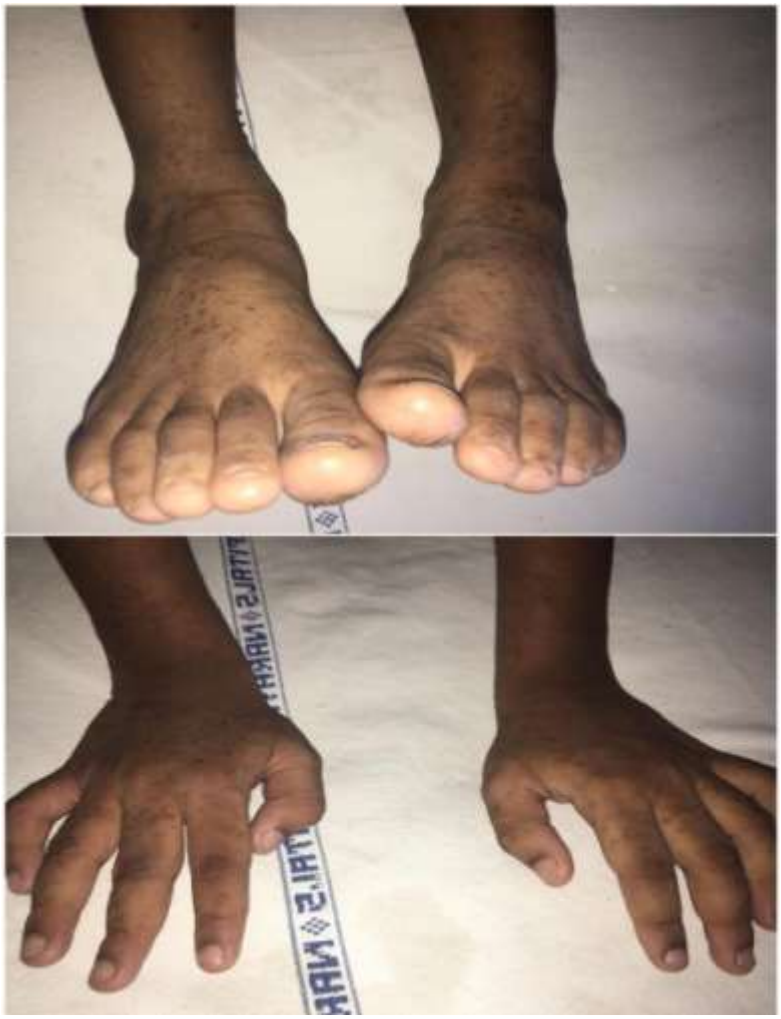
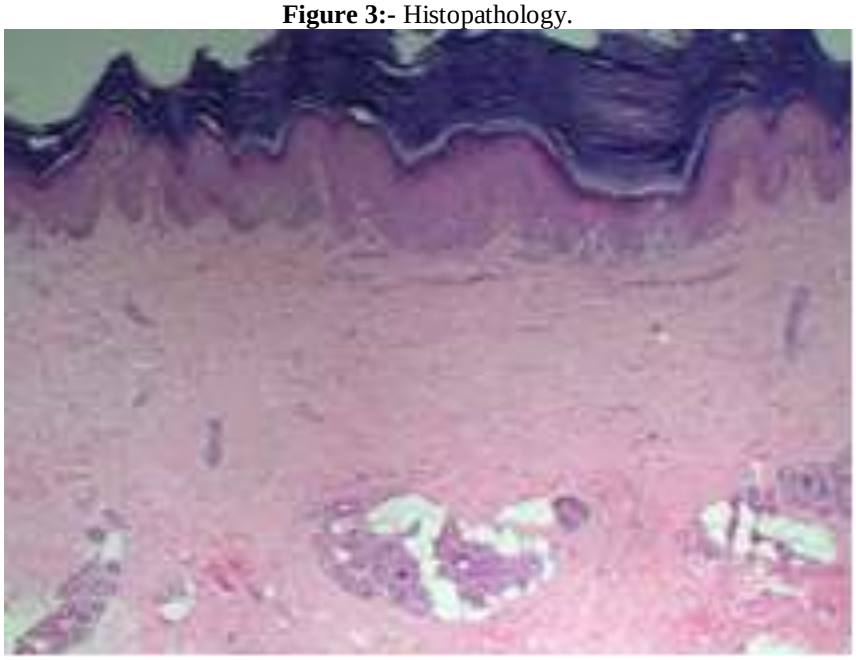


Figure 2:- Hyperkeratotic papules overlegs and dorsum of hands.

Figure 3:- Histopathology.



Discussion:-

Darier disease is a rarehereditary acantholytic dermatosis caused by heterozygous mutations in the ATP2A2 gene which encodes the sarco/endoplasmic reticulum Ca^{2+} ATPase isoform 2 (SERCA2), a calcium pump located in the endoplasmic reticulum (ER) membrane that plays a pivotal role in intracellular calciumsignalling,dysfunctionof which leads toimpaired processing of junctional proteins resulting in acantholysis and dyskeratosis due to increased apoptosis. ATP2A2gene is highly expressed both in the skin and in the brain[3]. It ischaracterized primarily by malodorous, warty, greasy, yellow to brown, hyperkeratotic papules, on the seborrheic areas of the chest, upper back, forehead, scalp, nasolabial folds, and ears.These lesionscan lead to large crusted plaques.Typical nailabnormalities are characterized by longitudinal white or red lines with ridges and distal V-shaped notches on the nail surface. In addition, papules may appear onmucosal membranes, mainly oral, pharynx, vulva and rectum. Thewhitish oral mucosal lesions mostly affect the hard palate andresemble nicotinic stomatitis.Ultra-violet B (UVB) irradiation, heat, friction, and infections of affected areas are clinically known to exacerbate symptoms[4]. Affected areas of skin are susceptible to secondary infections like Kaposi varicelliform eruption and also by bacteria, yeast and dermatophytes. Variousneuropsychiatricdisorders such as epilepsy, intellectual impairment, and mood disorders have been reportedin patients with Darier disease. Neuropsychiatriceffects in Darier disease have been hypothesized to be due to accumulation ofinsoluble SERCA2 aggregateswithin the neurons and contribution ofdisfigurement and isolation due to severe skin involvement or it can be aco-occurrence. We hereby report a case of8 year old boywho hadunique co-existenceofDarier disease and Neuro-psychiatric disorderi.eAttention Deficit Hyperactivity Disorderbeing born in a 2nd degree consanguineous marriage.This case has attained rarity because as per the available information there are no published case reports in India of Darier disease associated with Attention deficit hyperactivity disorder. This unreported association is pointing the need of further studies on ADHD and other neuropsychiatric disorders in patients with Darier diseasetosubstantiate the association.

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