Volume 9, Issue 6, June – 2024 International Journal of Innovative Science and Research Technology
ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/IJISRT24JUN1954

Dariers Disease with Oral Manifestations : A Clinical Rarity : Case Report

Dr. Saurav Shankar Das[1]
Post Graduate
Department of Periodontology, AJIDS

Dr. Nandini N. Krishnamurthy[2]
Head of Department,
Department of Periodontology, AJIDS

Dr. Grace Mary Joseph[3]
Alumni
Department of Periodontology, AJIDS

Dr. Shijal Narayan[4]
Post Graduate
Department of Prosthodontics, AJIDS

Dr. Riolla Sanchia Dsouza[5]
Post Graduate
Department of Periodontology, AJIDS

Abstract:- Darier disease (DD), also known as keratosis folicularis and White disease , is an inherited autosomal dominant genodermatosis. DD often develops in childhood, persists through adolescence and causes small papules predominantly in seborrheic areas such as the face , chest and back. The oral lesions are asymptomatic and comprise multiple white papules in the buccal mucosa and the soft and hard palate , giving a cobblestone appearance. Histologically , DD is characterized by corps ronds and grains in addition to suprabasal cleavage. There are no currently validated curative treatments available for DD, with the majority of cases treated symptomatically.

This is a case report of DD with varied manifestations intraorally and extraorally. Diagnosis was established on the basis of clinical , histopathological and dermatological findings.

Patient reported to the OPD with a chief complaint of loose teeth. A holistic treatment approach was carried out based on clinical and histological findings. Functional and esthetic demands were met with satisfactory results.

Keywords:- *Dariers Disease , Extraction , Complete Denture ,Biopsy.*

I. INTRODUCTION

Keratosis follicularis is also known as darriers disease or darrier white disease.[1]It is an autosomal dominant inherited genodermatosis.It is characterized by greasy hyperkeratotic papules in seborrheic regions with nail abnormalities and mucous membrane changes. among with these palmoplanktar pits and acting keratosis has also been noted.[2] The disease was first reported by Darier and white in the year 1889.[3] White was the first to recognise the genetic nature of keratotic follicularis in mother and daughter. It is usually seen more in males when compared to females with a prevalence rate of 1:1,00,000.[4]Usually the oral lesions are asymptomatic.

The given case report describes Dariers Disease with intra and extra oral findings which was symptomatically releaved and the functional and esthetic demand of the patient was achieved with a multidisciplinary approach.

II. CASE 1: DARIER DISEASE WITH ORAL MANIFESTATIONS

A 39 year old female patient came to OPD with a chief complaint of mobility of tooth and difficulty to chew food . Patient also showed concerns about multiple small spots all over the body.

Patient gave history of tooth mobility and difficulty to chew food since 6 months. There were no aggravating or relieving factors. Patient also gave history of appearance of multiple small spots all over the body which did not aggravate on exposure to sunlight.

Intra oral examination revealed Grade III mobility and Pocket Probing Depth>10mm with respect to upper and lower anteriors and posteriors.

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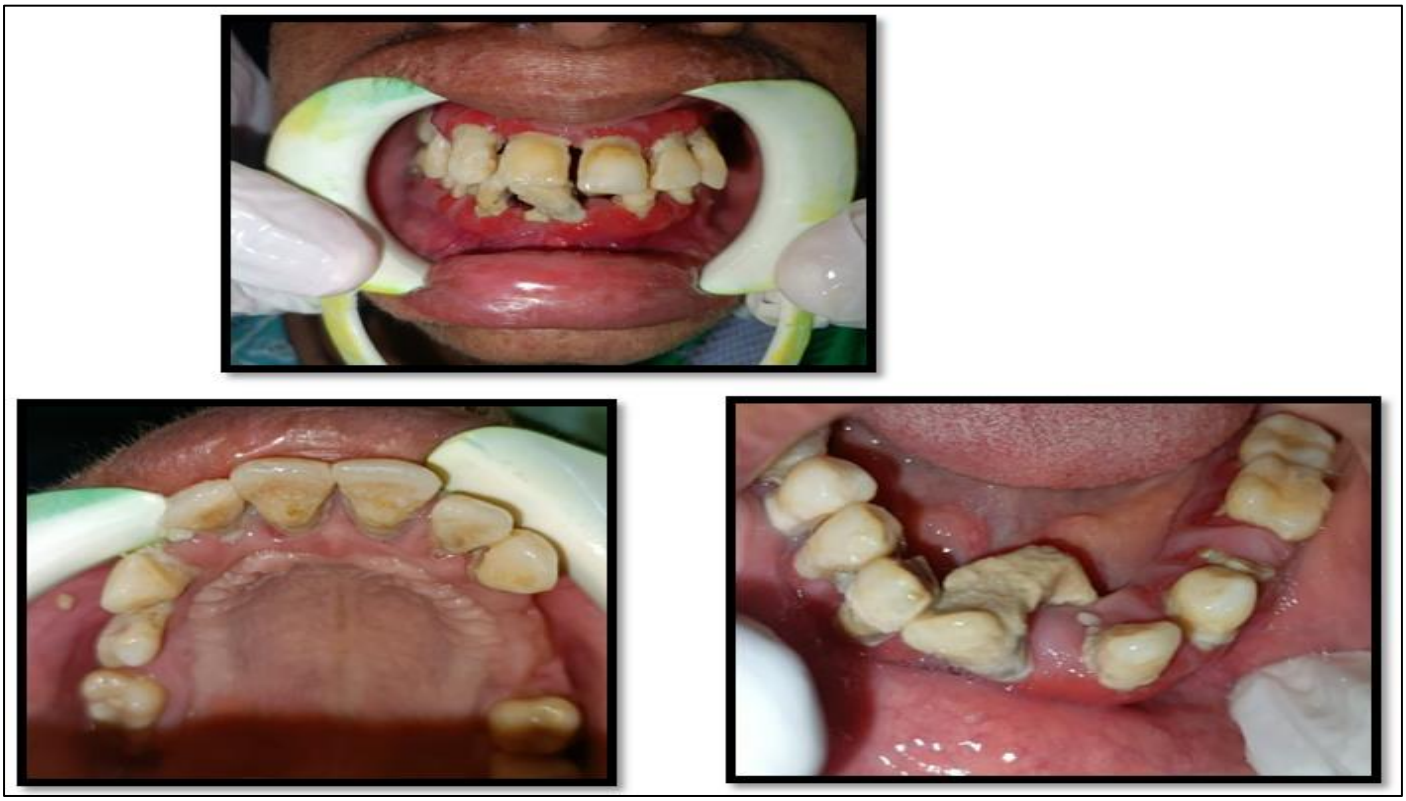

Fig 1 Intra – Oral Examination

Skin examination revealed generalized keratotic papules with brittle nails. Multiple hyperkeratotic papules were noted around the oral cavity with lesions on face, forehead, neck thighs and behind the auricle.Acantholytic lesions were noted on mucosa of lower lip.

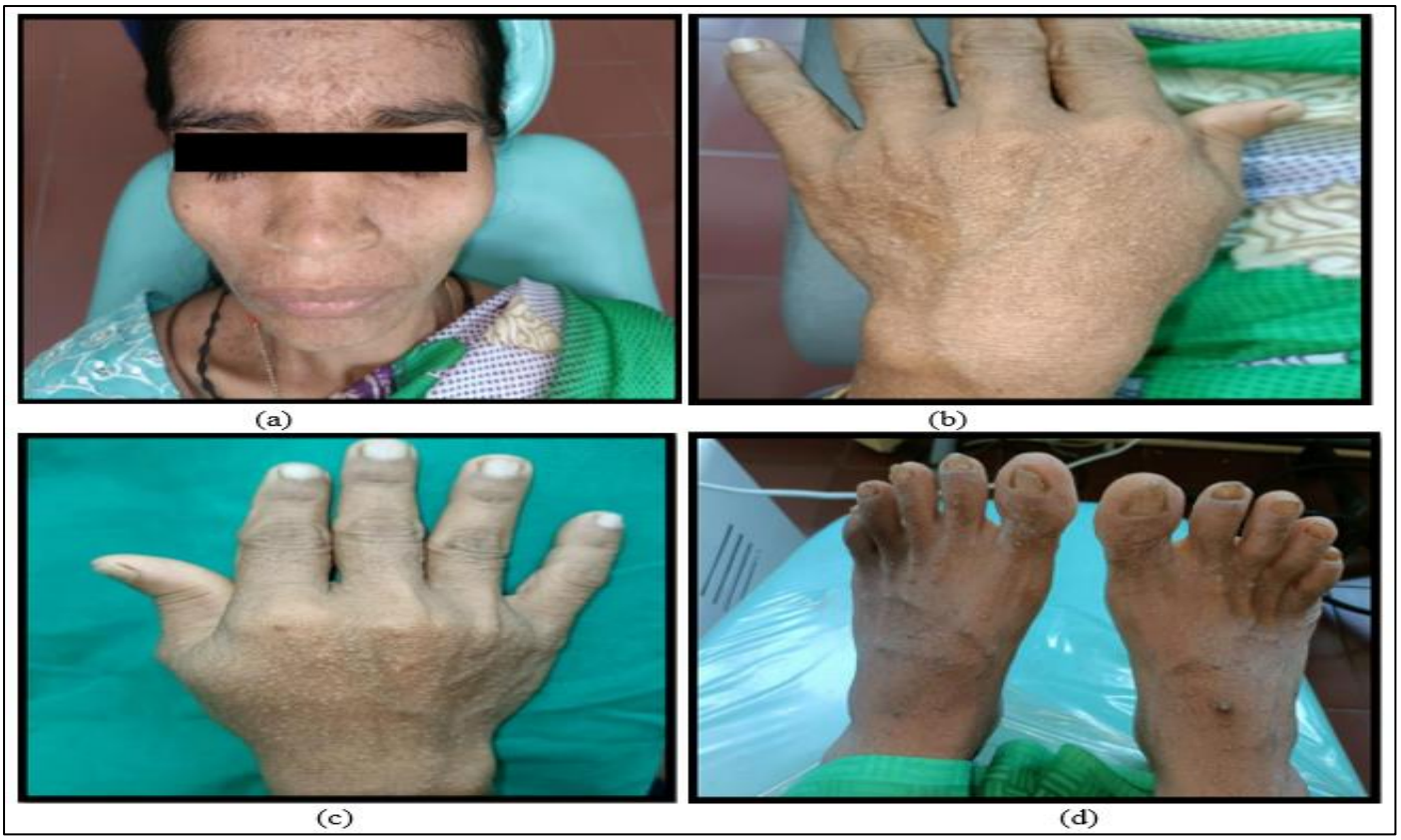

Fig 2 (a) Multiple hyperkeratotic papules noted around the oral cavity with lesions on face, forehead ; Fig .2(b,c,d) Multiple hyperkeratotic papules noted on the upper and lower limbs.

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The orthopantamogram (OPG) revealed generalized bone loss upto apical third of tooth root in relation to maxillary and mandibular anteriors and posteriors.

After visual and radiographic examination patient was given a differential diagnosis of Darriers disease; Adenoma sebaceum;Seborrheic dermatitis;Acroderma pigmentosa and a provisional diagnosis of Generalized periodontitis Stage IV , Grade C.

Patient was referred to Department of Dermatology for biopsy and diagnosis.

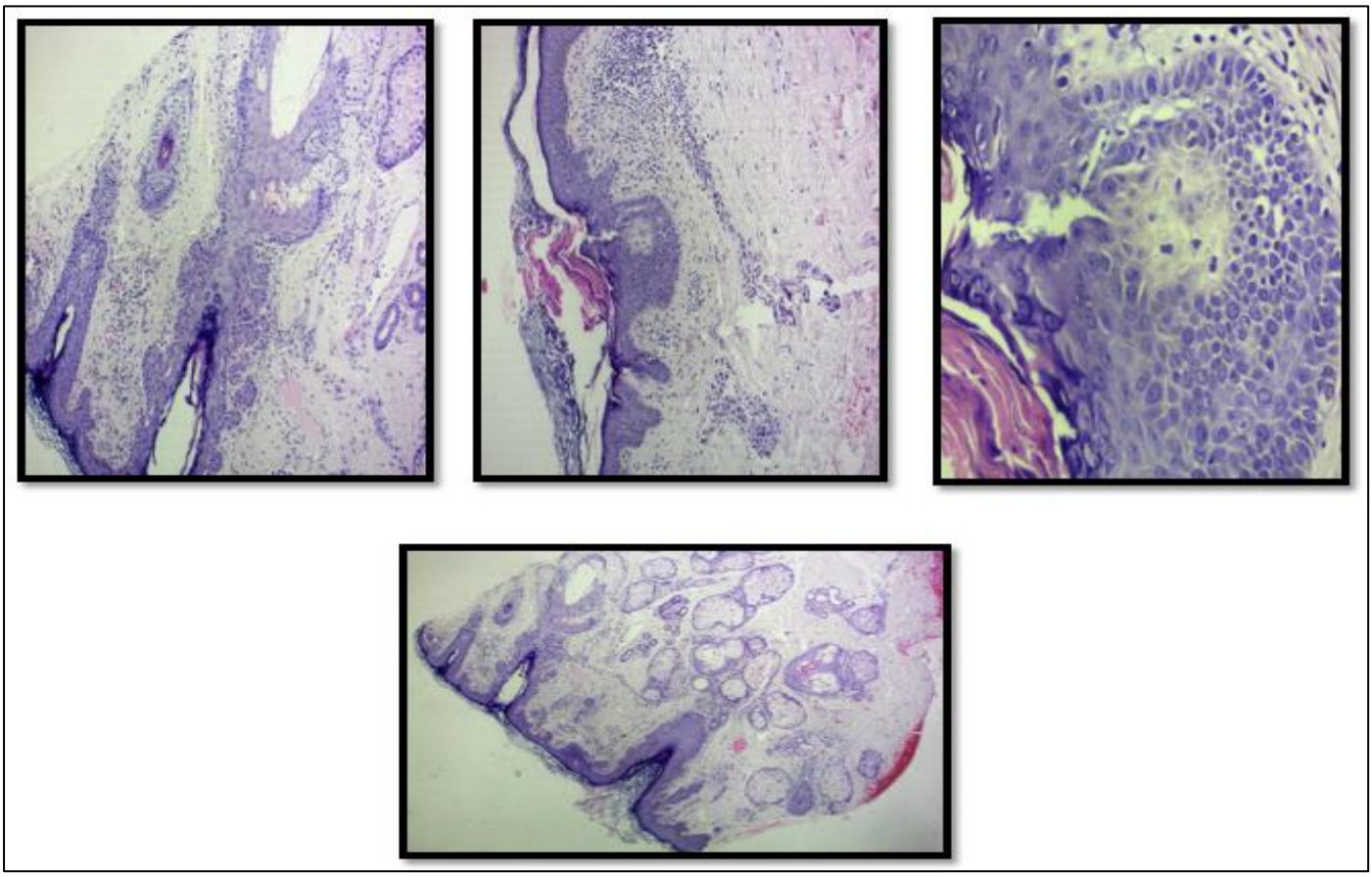

Fig 3 Histological Images Revealing Suprabasal Split in the Epithelium with Acantholytic and Dyskeratotic Cells Observed as Corps Ronds and Corps Grains

Following this , biopsy of the intra oral lesion was taken and sent to Department of Oral Pathology for final diagnosis.

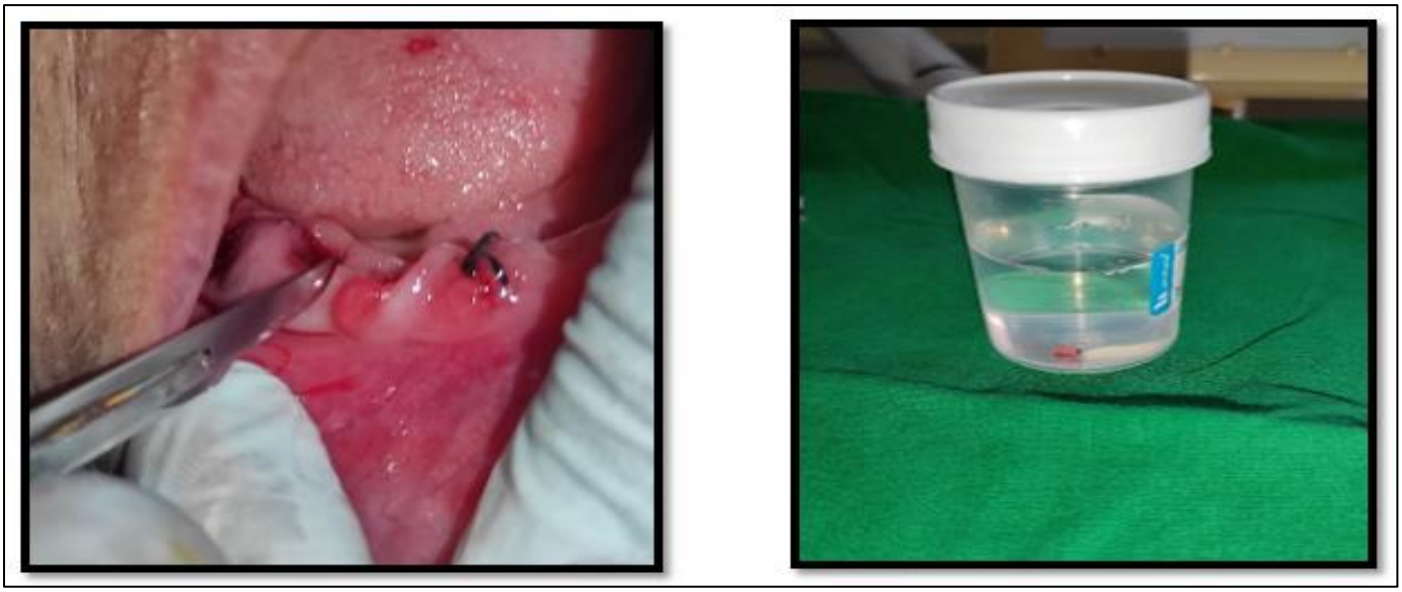

Fig 4 Biopsy of Gingival Socket Lining Epithelium

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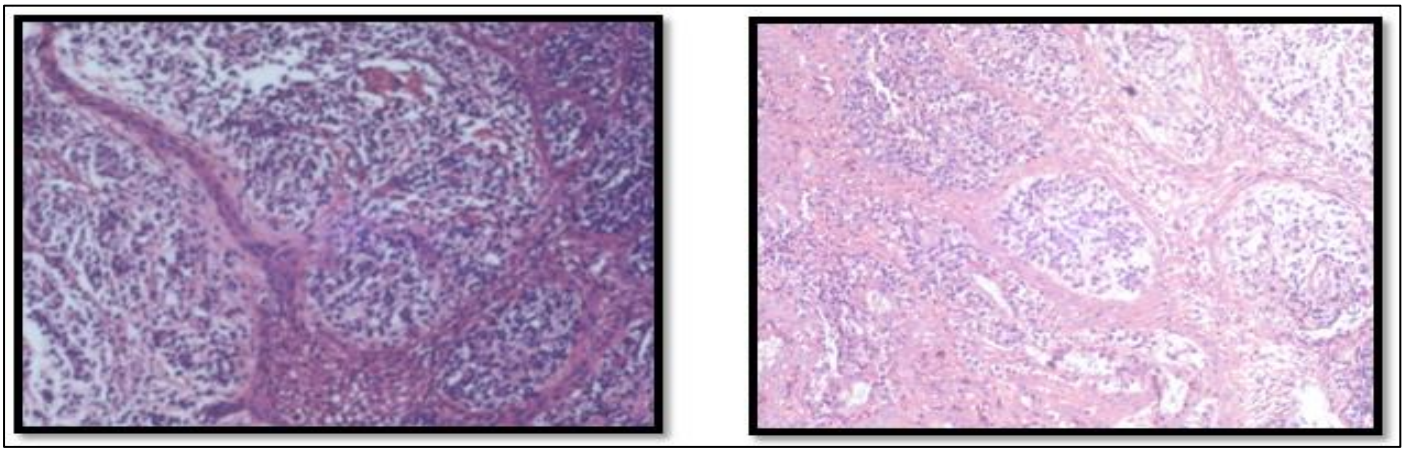

Fig 5 Given H&E stained section shows overlying hyperkeratotic epithelium with focal plaqued areas and granular layer. Degenarative epithelial cells in round and granular shape(corps and grains) seen in parabasal layer. Connective tissue shows perivascular lymphocytic infiltration along with other inflammatory cells in a fibro vascular stromal background.

In clinical correlation of the dermal and mucosal lesions the final diagnosis was suggested as DARIERS DISEASE.

A holistic treatment approach was followed with a goal of full mouth rehabilitation to meet the esthetic and functional demands.

An oral topical anesthetic gel was applied before the local anesthetic injection to prevent injection discomfort. an injection of Lignocaine hydrochloride 2% with adrenaline 1: 100,000 was administered. Subjective and objective evaluation of local anesthesia was obtained to ensure the success of pain control. A mucoperiosteal elevator was used to reflect the gingival tissues around the teeth. extraction forceps were used to luxate the teeth and the teeth were delivered without complications(Fig.6) .Copious irrigation with normal saline was followed by gauze packed in the extraction site and the patient was asked to bite for half an hour. Closure of the extraction sockets were achieved by silk sutures .Copious Verbal and written postoperative instructions were given and illustrated to the patient and the patient's guardian. the patient was kept under careful monitoring to ensure the proper healing of the periodontium.The patient had multiple visits to continue her treatment based on the agreed-upon treatment plan, including acomplete denture prosthesis. Followup visits were scheduled at 14 days (Fig.7),60 days (Fig.8) and at 3 months(Fig.9). Patient oral hygiene improvement was noticed in the followup visits. Following the extraction the patient was referred to the Dept of Prosthodontics for the fabrication of a complete denture so in to achieve both esthetic and functional need of the patient.

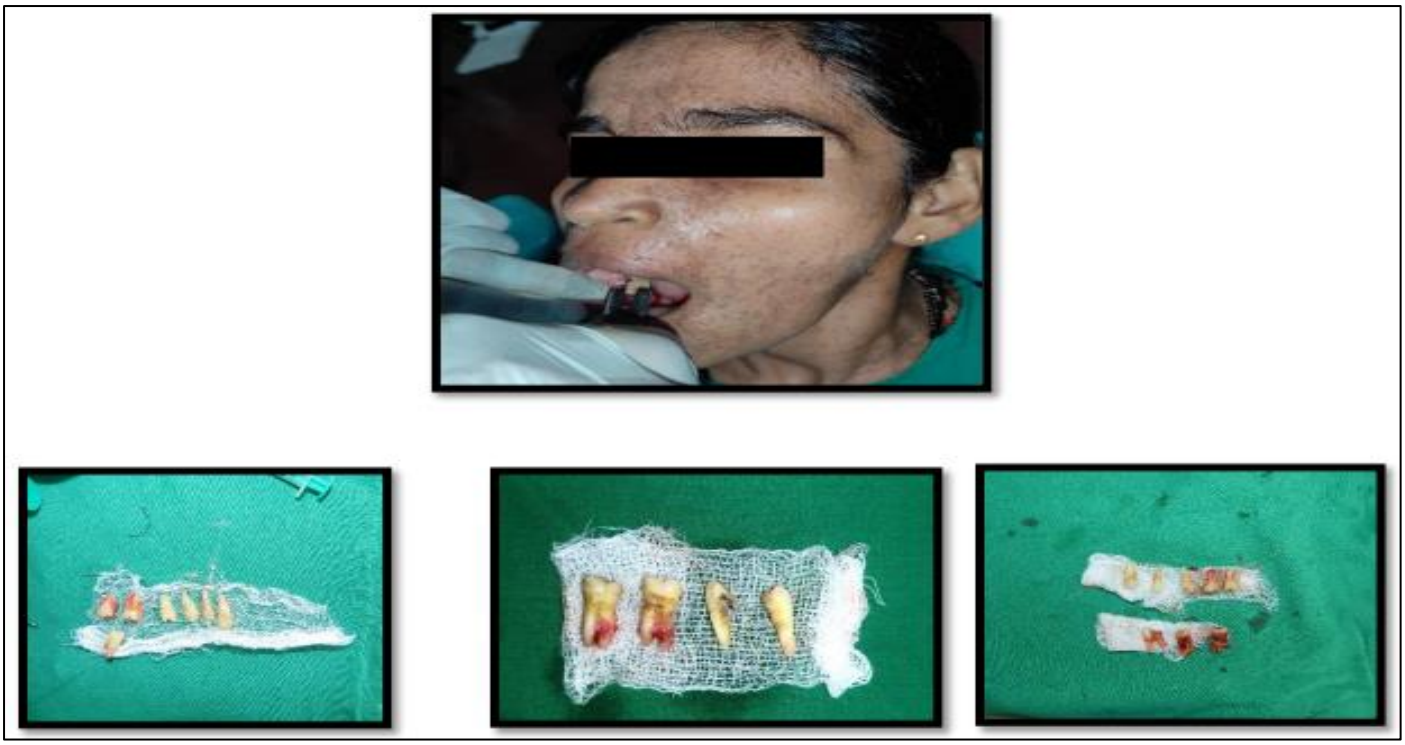

Fig 6 Atraumatic Full Mouth Extraction

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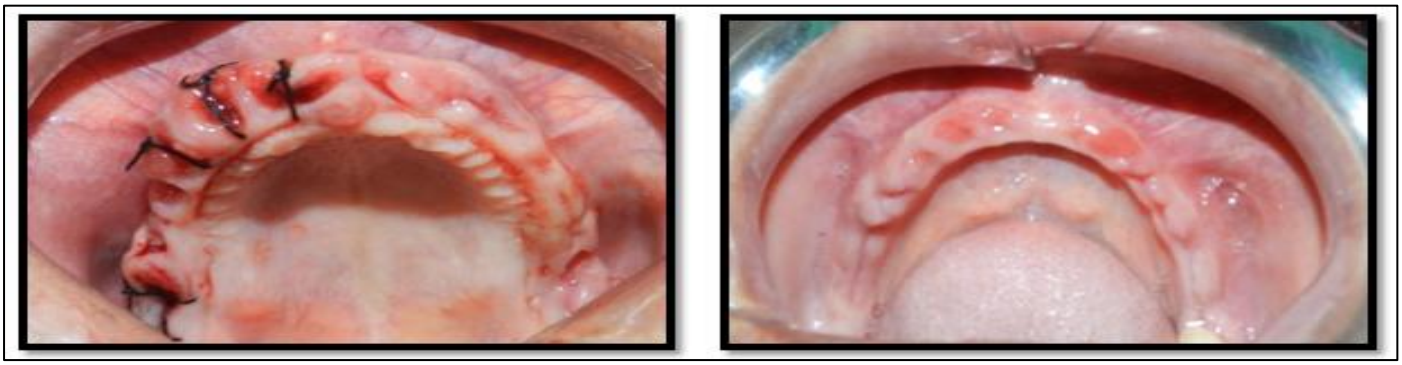

Fig 7 Healing (14 Days) Post Extraction

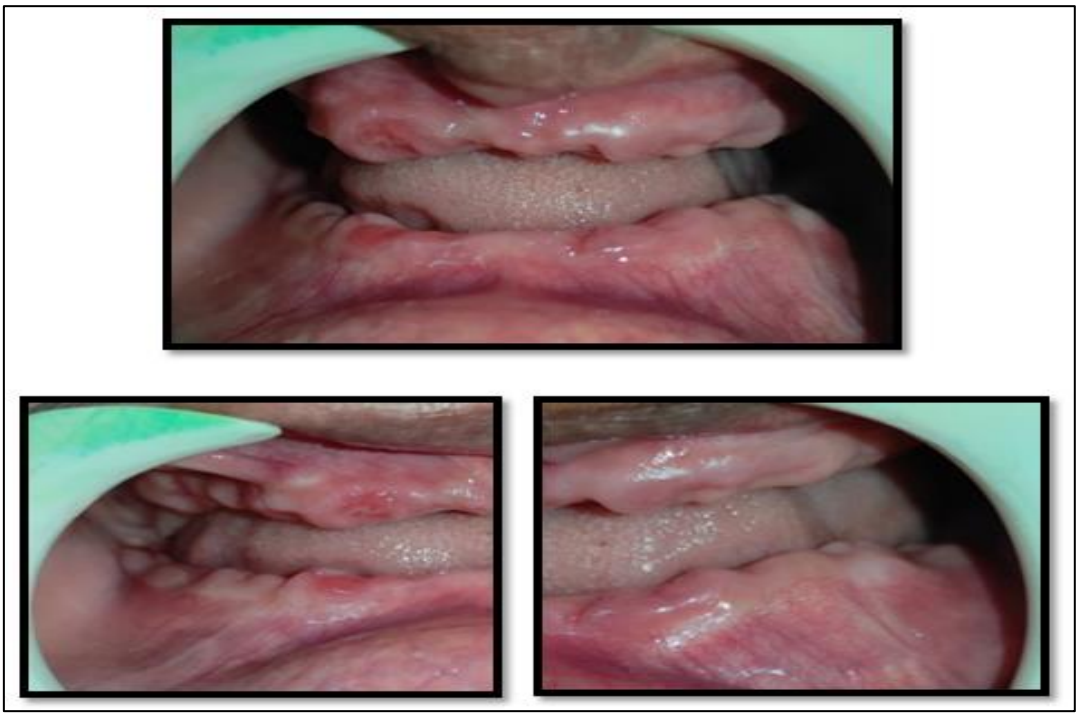

Fig 8 Healing (60 Days) Post Extraction

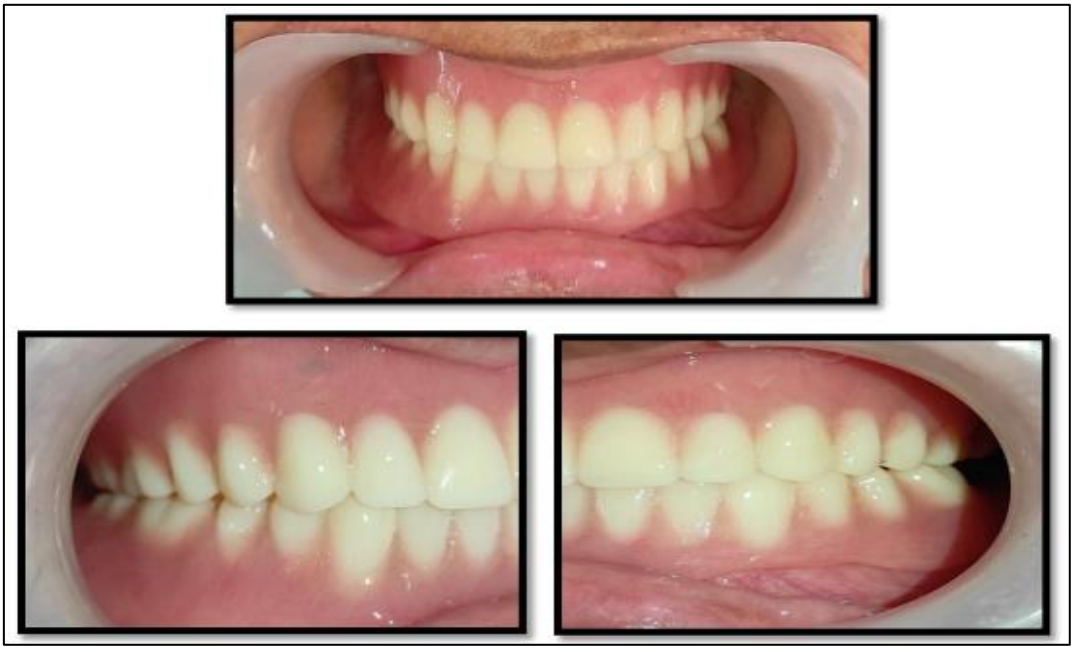

Fig 9 Functional and Esthetic Demands met by Full Mouth Complete Denture

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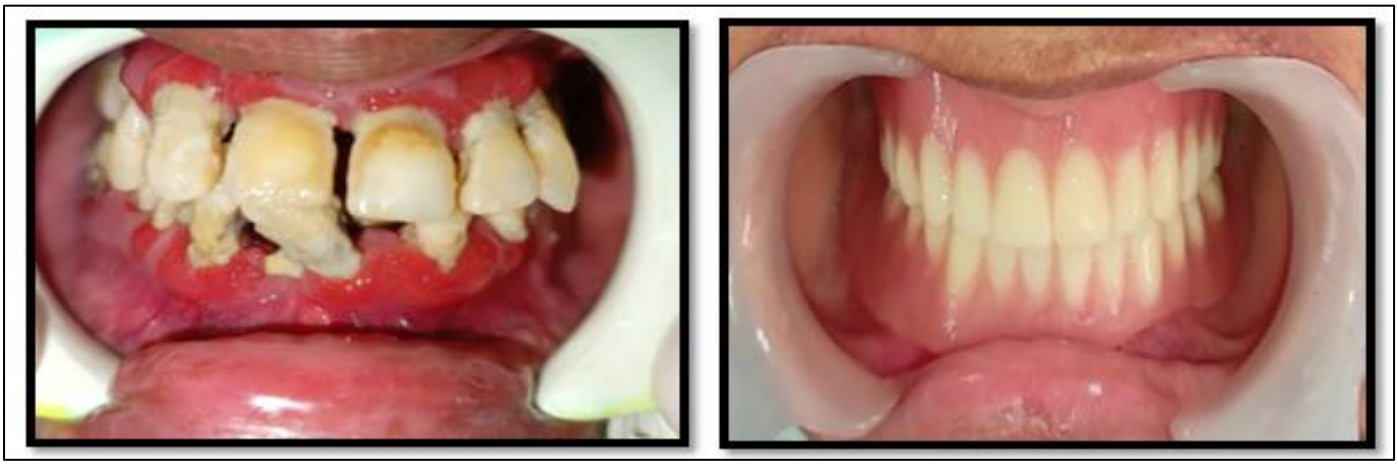

Fig 10 (a) Intra Oral Photograph (DAY : 1) ; (b)) Intra Oral Photograph (DAY : 90)

III. DISCUSSION

Darier disease is a well-established genodermatosis characterized by autosomal dominant inheritance and abnormal keratinization . While the exact mechanisms remain under investigation, the disease is thought to involve structural defects in cell adhesion complexes, particularly desmosomes. This dysfunction may contribute to abnormal interactions between epithelial cells, though the role of autoimmunity requires further exploration. [5] More recently, it has been related to mutations in the gene encoding the sarco/endoplasmic reticulum Ca2+ ATPase pump (SERCA2), resulting in abnormal organization or maturation of complexes responsible for cell adhesion, thus leading to the disturbance.[6]The implications of the Darier disease are more associated with cosmetic and esthetic than functional implications, since this is a benign dermatosis. However, depending on the severity of the disease and affected area, the patient has more complaints and the emotional status may be damaged by esthetic reasons.[7]Some of the clinical characteristic findings include nail abnormalities which is presented as alternate white and red longitudinal streaks (candicanc nails) and V shaped nicking of free nail edge[8] .Oral lesions include cobble stonning of oral mucosa, gingival hypertrophy and obstructive sialadenitis.[9] Skin lesions comprises of brown keratotic papules , which could be puritic, painful with distinctive odour.[10]

The diagnosis can be confirmed by skin biopsy and genetic testing followed by symptomatic relief of the patient.

IV. CONCLUSION

Dental professionals frequently detect oral signs of inherited conditions during routine examinations, often before these conditions are identified by medical doctors. These asymptomatic lesions can then be a prompt for genetic counseling to discuss the underlying condition and its potential transmission to offspring.. A biopsy is essential for a definitive diagnosis. Following the biopsy results, a referral to a dermatologist for further evaluation is recommended. To ensure optimal care, patients need to be fully informed about the potential complications of this disorder and the necessary management strategies. In severe cases, a psychologist should be involved to monitor and address any emotional challenges. This collaborative approach, with a multidisciplinary team, ensures comprehensive care for the patient.

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