Erythromelanosis follicuaris faciei: in two siblings with autosomal recessive inheritance

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Introduction

Erythromelanosis follicuaris faciei (EFF) is a rare underdiagnosed disease, where the neck is affected, the disease is called Erythromelanosis follicuaris faciei et colli (EFFC)[2,5]. The disease was first reported by Kitamura and collaborators in 1960[2,3]. The disease is characterized by well defined erythema, pigmentation and follicular papules mainly involving preauricular area, cheeks with extension to the sides of the neck. The etiology of the disease is still debatable but possible autosomal recessive mode of inheritance and possible polyetiologic influence (e.g., familial, environmental) has been suggested[2,4]. Herein we report EFF in two siblings in a family of first degree consanguinity.

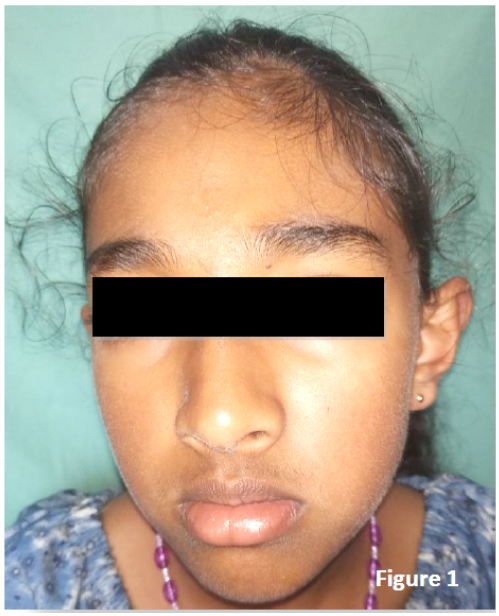

Figure 1

Cases

9 year old girl and a 3 year old boy, products of a consanguineous marriage presented with asymptomatic symmetrical hyperpigmentation and erythema of cheeks since 2 years and 3 months of age respectively. On examination there were symmetrical ill defined erythematous to hyperpigmented patches studded with tiny follicular papules (Figure 1, 2, 3, 4). In the elderly sibling multiple telangiectatic vessels were evident over the affected areas but there was no skin atrophy[2]. Mucus membranes, nails, hair were normal and systemic examinations were unremarkable.

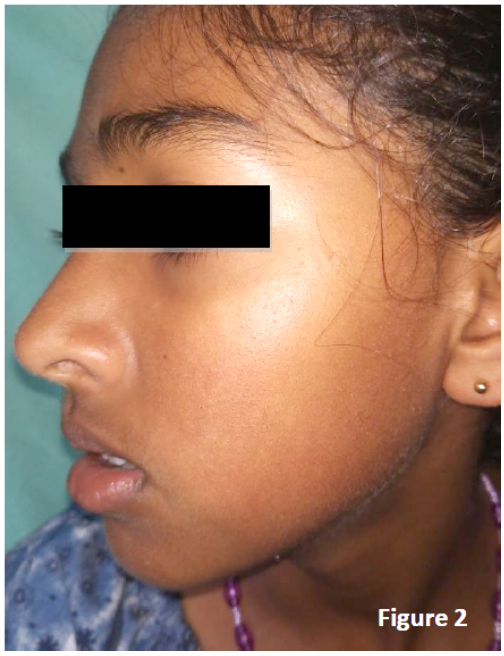

Figure 2

Hyperlinear palms were observed in both patients but there was no evidence of keratosis pilaris. Due to classic clinical features with possible autosomal recessive inheritance the diagnosis of EFF was made clinically without histopathological evaluation. Both children were advised on sun protection, a sunscreen and a bland emollient with a plan to follow up every six months.

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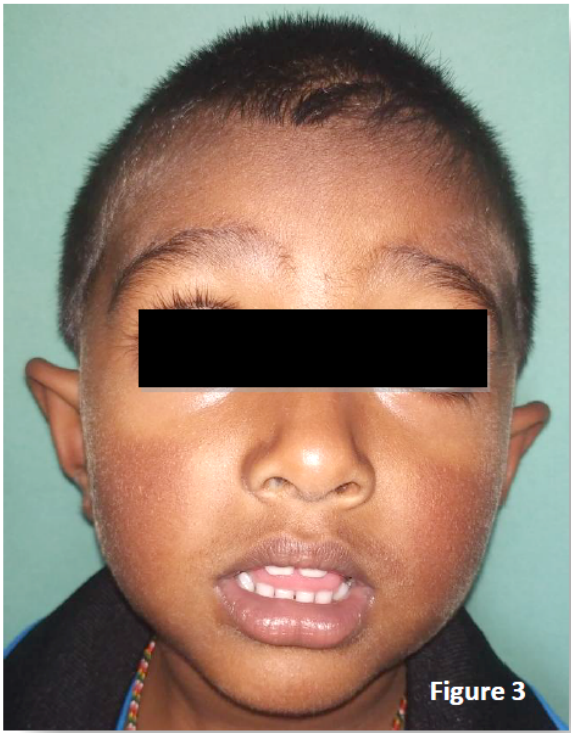

Figure 3

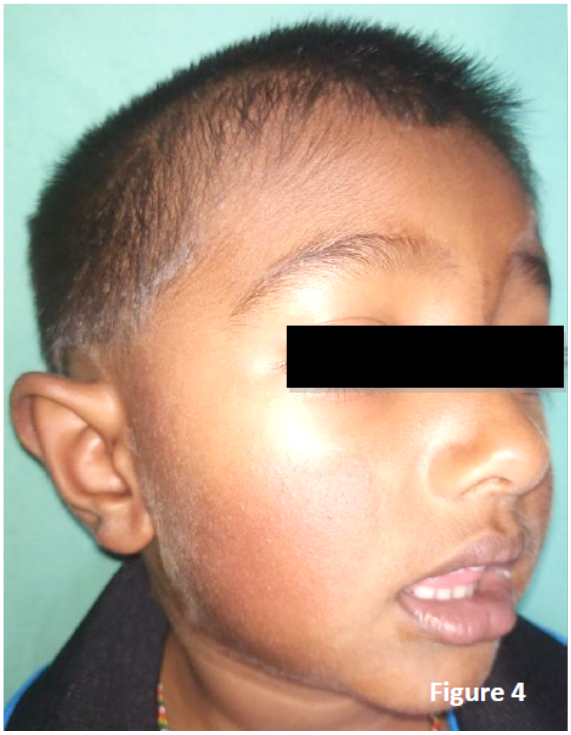

Figure 4

Discussion

Being a rare disease with an unknown etiology Erythromeanosis foicuaris faciei has been reported to occur with autosomal recessive inheritance and spontaneous mutations[2,4]. Having consanguineous parents, the above two siblings were thought to have autosomal recessive inheritance. The disease was thought to occur only in males but recently reported to have equal incidence among both genders[1,4]. EFF is characterized by well defined erythematous-brown patches in preauricular and maxillary areas and is associated with follicular papules over the affected areas and keratosis pilaris over classic sites[4]. In some patients telangiectasia can be observed over sun exposed areas.

Histology demonstrates follicular plugging, follicular dilatation, increased basal layer pigmentation, perivascular/periadenexial inflammatory infiltrate and dilated upper dermal blood vessels[2,4].

Emollients, keratolytics, metronidazole gel, hydroquinone, vitamin D analogues, chemical peels, glycolic acid peels and Laser has been offered as treatment with variable results but the disease recurs after discontinuation of treatment[1,2].

References

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