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RESEARCH ARTICLE

ATRICHIA WITH PAPULAR LESIONS : A CASE REPORT

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Abstract

Atrichia with papular lesions is a rare Autosomal recessive disorder characterized clinically by complete and irreversible hair loss shortly after birth, and is associated with the development of keratinfilled cysts all over the body. We present a 10 year old girl with extensive alopecia and papular lesions over scalp,upper limbs,lower limbs, abdomen and back . A clinical diagnosis of Atrichia with papular lesions was made after excluding Vit D dependent Rickets. An accurate diagnosis is important to prevent unnecessary treatment of APL with systemic steroids when misdiagnosed as alopecia universalis.

Introduction:-

Children affected with atrichia with papular lesions (APL) are usually born with normal hair which is shed during the first month of life and the hair never regrows thereafter. During the first to second decade of life, patients develops a diffuse papular rash, which has been noted to be particularly prominent over the cheeks and scalp but can involve almost any part of the body.These patients are nonresponsive to any therapy.

Case Report:

A10 year old girl presented with complete loss of hair over the scalp since 1month of her age. Patient was born with scanty hair over the scalp at birth, later she lost hair by 1 month of her age.History of development of hyperpigmented skin lesions all over the body since 5 years. There is no delay in milestones. None of her family members had similar skin lesions. History of consanguinous marraige present.

On examination:

Multiple hyperpigmented hyperkeratotic papules of size 2 mm to 3 mm noted all over the scalp with follicular papules over upper limbs,lower limbs,abdomen and back. Complete absence of hair noted all over the body. Mucosa,nails, teeth and sweating are normal. Clinically no bony abnormality, no dysmorphic features and no systemic involvement present.

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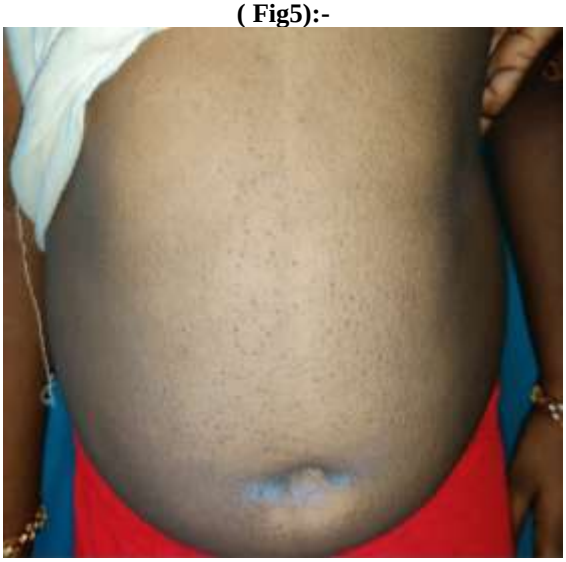
(Fig.1):-

(Fig.2):-

(Fig 3):-

(Fig4):-

(Fig5):-



(Fig 6):-

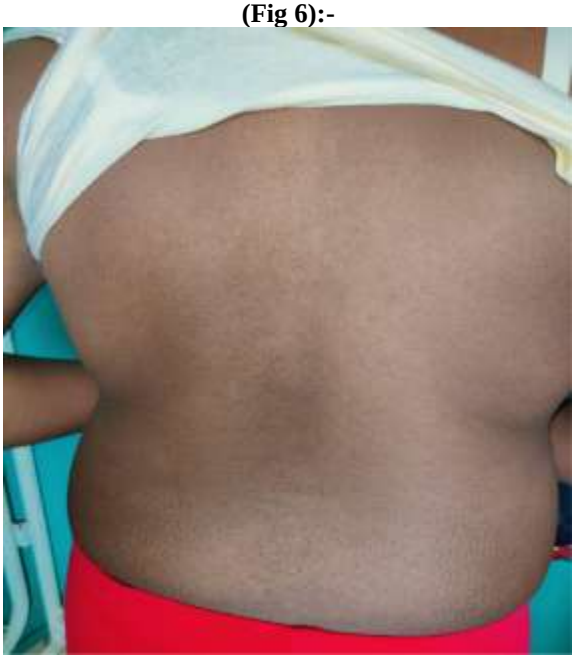


Fig 1 and Fig 2 : Multiple hyperpigmented hyperkeratotic papules over scalp with complete loss of scalp hair.
Fig 3 to fig 6 : Follicular papules over upper limb and lower limbs, abdomen and back respectively.

Management:
There is no treatment for APL.
An accurate diagnosis is critical to prevent unnecessary treatment.

Camouflage/wigs helps for better cosmetic appearance.
For Scalp : wigs, hair weaving, prostheses of human/ acrylic hair.
For Eyebrows: microblading/ tattooing.
For Eye lashes: False eyelashes.

Discussion:-

Atrichia with papular lesions is caused by mutations in the HR gene, encoding a transcription co-repressor factor called hairless. Hairless may regulate hair cycling through its effect on the WNT signalling pathway, through its effect on polyamine synthesis or through additional targets. The hair matrix cells in APL appear to undergo a premature and massive apoptosis. Regardless of its exact mechanism of action, downregulation of hairless is associated with abnormal catagen and interferes with normal hair cycle leading to the abnormal development of hair follicles into epidermal cysts that manifest at the clinical level as papules. Affected individuals display sparse to normal hair at birth, develop coarse, wiry and twisted hair in early childhood,followed by the development of generalized alopecia in adulthood.

Diagnostic criteria for Atrichia with papular lesions:

A)Major criteria:(4 out of 5 required for diagnosis)

1. Permanent and total loss of hair in the first months after birth.
2. White and milialike papules on face, scalp, arms, elbows, thighs, or knees from infancy or childhood.
3. Mature hair follicle structures replaced by follicular cysts in scalp histology.
4. Mutation(s) in the human hairless gene through genetic testing
5.Clinical and/or molecular exclusion of rickets due to lack of Vitamin D.

B) Minor criteria: (supplementary criteria)

1. Cousin marriage
2. Absence of secondary axillary,pubic, or body hair growth, and/or sparse eyebrows and eyelashes
3. Healthy development, including normal bones, teeth, nails, and normal sweating.
4. Whitishhypopigmented streaks on the scalp
5. Lack of response to any treatment.

Conclusion:-

Atrichia with papular lesions is rare Autosomal recessive disease. It is caused by a mutation in the zinc finger domain in the human hairless gene of the 8p12 chromosome. It can be easily misdiagnosed as alopecia universalis or vitamin Ddependent rickets type IIA. Hence a high index of suspicion should be kept in mind in patients presenting with generalized alopecia to avoid misdiagnosis and to avoid unnecessary treatments especially with steroids.

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