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**Case Report**

# Twisted tressess: a rare case series of familial woolly hair

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## ABSTRACT

Woolly hair is an uncommon congenital anomaly of the scalp hair presenting with strongly coiled hair involving a localized area of the scalp or covering the entire side and occurring in non-black people. Among the syndromes presenting with woolly hair, the most known are the Naxos syndrome, the Carvajal-Huerta syndrome, the woolly hair/hypotrichosis, the ectodermal dysplasia-skin fragility and the tricho-hepato-enteric syndrome. Case characteristics: herein, we report a case of autosomal recessive familial woolly hair of an Asian female child and member of a family, her younger brother affected by a localised woolly hair, not associated to complications. Children with woolly hair must be examined completely and monitored regularly to rule out cardiofaciocutaneous and Noonan syndrome.

**Keywords:** Kinky hair, Light microscopy, Naxos syndrome, Carvajal syndrome, Woolly hair

## INTRODUCTION

Woolly hair is a rare congenital structural anomaly of scalp hair marked by generalized or localized occurrence of curly hair, which usually demonstrates slow growth and/or easy breakage, sometimes associated with hyperpigmentation occurring in an individual of non-Negroid origin. It was first described in 1907 by Gossage in a European family and classified by Hutchinson et al in 1974 as: localized circumscribed variant (woolly hair nevus), autosomal dominant variant (hereditary woolly hair), autosomal recessive variant (familial woolly hair).[1,2]

## CASE REPORT

An eleven-year-old Asian female presented with chief complaints of very short, curly, brittle hair since birth. There was history of difficulty in combing and also there was history of itching and dryness. The hair never grew longer than the current length. There was no history suggestive of systemic involvement and no history suggestive of physical and mental retardation. Similar history was also present in her younger sibling, a nine years old male. There was history of consanguineous marriage in the parents of the patient. There was no history of similar complaints in the parents.

On examination, the scalp hair were hypo-pigmented, coarse, short, thin, lusterless, dry and tightly curled (Figure 1). Broken hair shafts were visible. Body and pubic hair were sparse, shorter, lighter with absence of axillary hair. Hair pull test was negative. Seborrheic dermatitis was also present over scalp. Nail and teeth examination was normal. Sibling of the patient was examined and had similar findings (Figure 2). Systemic examination was within normal limits in both the siblings. Routine investigations were done including complete blood counts, liver function test, renal function test, routine urine examination, chest radiograph and

nothing abnormal was detected. Light microscopic of the hair was done and it showed thin and hypo-pigmented shaft (Figure 3).

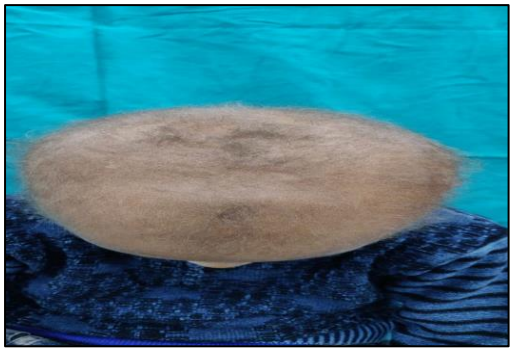

**Figure 1: Short, sparse and hypopigmented hair over the scalp.**

**Figure 2: Sibling of the patient showing dry, coarse, lustreless scalp hair.**

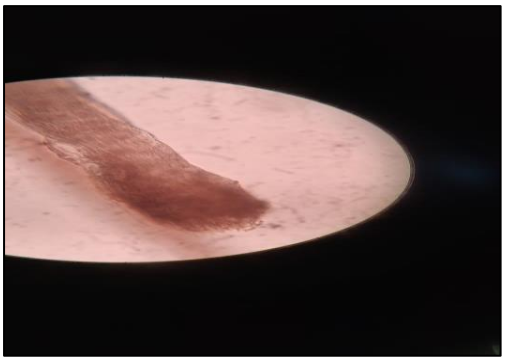

**Figure 3: Light microscopy showing hypopigmented and thin hair shaft.**

KOH examination of the hair was normal. Cardiac evaluation including Electrocardiogram and Echocardiography was within normal limits. Genetic analysis could not be done due to the non-availability of the facilities for genetic studies in the institution. Owing to the unavailability, Electron microscopy of the cut section could not be done. A diagnosis of autosomal recessive woolly hair syndrome was made based on history, clinical examination and investigations.

## DISCUSSION

Woolly hair was first described by Gossage in 1907 in a European family.[1] Woolly hair is extremely curly, with the average diameter of hair recorded up to a maximum of 0.5 cm. It is different from the curly hair of the black people, in that the curled hair of black people lies separately while the curls of woolly hair usually merge.[3] Presence of woolly hair in non- blacks is extremely rare.[4] Very few cases of familial woolly hair are reported in India.

Autosomal recessive woolly hair syndrome refers to a rare congenital structural abnormality of scalp hair characterized by extreme kinkiness, easy breakage and hypo-pigmentation of hair seen in Asians and Caucasians. Woolly hair can be present at birth or appear in the first months of life and is most pronounced during childhood as the manifestation often becomes less severe in adulthood. The curls tend to form tight locks which make hair difficult to comb and may not grow beyond 2-3 cm with an average diameter of about 0.5 cm. The eyebrows, the hair on arm, leg, pubic and axillary region may be short and pale. Hair growth is usually normal but the anagen phase may be truncated resulting in reduced length of the hair. Hair shaft exhibits an elliptical cross section, an axial rotation and a kinked formation.[5] Pathogenesis of isolated AR woolly hair has been shown to be allelic to AR localized hypotrichosis(non-syndromic) caused by mutations in LPAR6 and LIPH genes.8 But AR woolly hair has also been described in the context of complex cardiocutaneous syndromes.[6]

Naxos syndrome- woolly hair, arrythmogenic right ventricular cardiomyopathy and palmoplanter keratoderma (mutations in gene encoding plakoglobin).

Carvajal syndrome- woolly hair, palmoplanter keratoderma and heart disease (deletion mutation in desmoplakin)

Patients with woolly hair may in some cases be affected with Noonan's syndrome and tend to display a short stature, ptosis, borderline intelligence, a webbed neck and pulmonary stenosis.[9]

Classical clinical presentation establishes the diagnosis and detailed systemic examination is done particularly for cardiovascular system to rule out associated cardiocutaneous syndromes. No treatment is currently available for woolly hair. But manifestation often become less severe with age. Harsh physical/chemical cosmetic treatments should be avoided. Number of cases of AR familial woolly hair described in non-Negroid individuals are very few.[7]

## CONCLUSION

We are reporting this case because of its rarity and to highlight the use of light microscopy as a convenient

diagnostic tool. Children with woolly hair must be examined completely and monitored regularly to rule out any systemic involvement, especially of the cardiovascular system.

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## REFERENCES

1. Gossage AM. The inheritance of certain human abnormalities. Quartz J Med. 1907;1:331-47.
2. Hutchinson PE, Cairns RJ, Wells RS. Woolly hair. Clinical and general aspects. Trans St Johns Hosp Dermatol Soc. 1974;60:160-77.
3. Vasudevan B, Verma R, Pragasam V, Badad A. A rare case of woolly hair with unusual associations. Indian Dermatol Online J. 2013;4:222-4.
4. Ramot Y, Zlotogorski A. The twisting tale of woolly hair: a trait with many causes. J Med Genet. 2015;52:217-23.
5. Naveen KN, Shetty SR, Radha H, Athaniker SB, Gundannanavar MR. Woolly hair with trichoscopic features. Indian J Paediatr Dermatol. 2016;17:56-7.
6. Singh A, Mathur D, Yadav S, Nijhawan M, Aggarwal P, Soni S et al. Woolly hair a rare presentation. Journal of Evolution of Medical and Dental Sciences. 2013;2:10051-55.
7. Singh SK, Manchanda K, Kumar A, Verma A. Familial Woolly Hair: A Rare Entity. Int J Trichology. 2012;4:288-89.
8. Matsuno N, Kunisada M, Kanki H, Simomura Y, Nishigori C. A Case of Autosomal Recessive Woolly Hair/Hypotrichosis with Alternation in Severity: Deterioration and Improvement with Age. Case Rep Dermatol. 2013;5(3):363-67.
9. Chien AJ, Valentine MC, Sybert VP. Hereditary woolly hair and keratosis pilaris. J Am Acad Dermatol. 2006;54:S35-9.

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