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Generalised Woolly Hair with Hyper-Extensibility of Digits: A Rare Case Report

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Abstract: ***Woolly hair is an infrequent inborn scalp hair irregularity identified by short, firmly coiled hair that may occur in the hair covering other parts of the body as well. Various conditions have been reported to be associated with woolly hair, the most frequent associations include palmoplantar keratoderma, keratosis pilaris, ichthyosis and cardiac abnormalities. Here, we report a case wherein a 13 year old boy presents with woolly hair associated with hyper-extensibility of joints of all the digits. [51]***

Keywords: Woolly hair, Hyper-extensibility of joints

1. Introduction

Woolly hair is an uncommon congenital hair abnormality that affects the structure of scalp hair. While the hair growth rate typically follows the norm, the phase in which the hair actively grows (Anagen phase) is cut short, leading to shorter hair. The hair shaft itself displays characteristics such as an elliptical cross-section, axial rotation, and the formation of kinks. There are primarily two types of woolly hair: Autosomal dominant woolly hair (also known as Hereditary woolly hair) and the rarer Autosomal recessive Familial woolly hair. [51]

2. Case Report

A 13-year-old Asian boy, product of non-consanguineous marriage, presented with history of light coloured,coiled hair on the scalp since birth. He also gave associated history of extreme flexibility of all ten digits of hands and feet, noticed since the age of 7 years. There was also history of Chest pains, breathlessness and recurrent ear infections. Single episode of epistaxis and hematuria occurred in 2017, for which treatment was taken elsewhere (details not available). There was no history of seizures, dizziness, headache, visual disturbances or any joint swelling. He was born at term through normal vaginal delivery without any complications and there was no delay in developmental milestones.

Examination of scalp revealed thin, tightly-coiled hair with hypo-pigmentation throughout. They were short and thinner in diameter. The eyebrows had scant hair[Fig 1-5]. Also hyperextension of all the metacarpophalangeal and metatarsophalangeal joints was elicited. [Fig 11-17]

Examination of Nails, Palms and Soles were normal [Fig 6-10] . Dental examination revealed normal findings. Eye examination was normal. There were no cardiac manifestations or any other systemic involvement.

Laboratory investigations revealed Hb-9gm/dl, normal biochemical parameters, peripheral smear revealing normocytic normochromic anaemia with anisopoikilocytosis and thrombocytopenia, ultrasonography revealed situs solitus and ECG, ECHO and Chest X Ray showed normal findings.

Patient and the attenders were explained in detail regarding the nature of the condition and the current unavailability of treatment for woolly hair. They were also explained in detail regarding the importance of regular follow ups and serial investigations for early detection and treatment of any associated abnormalities.

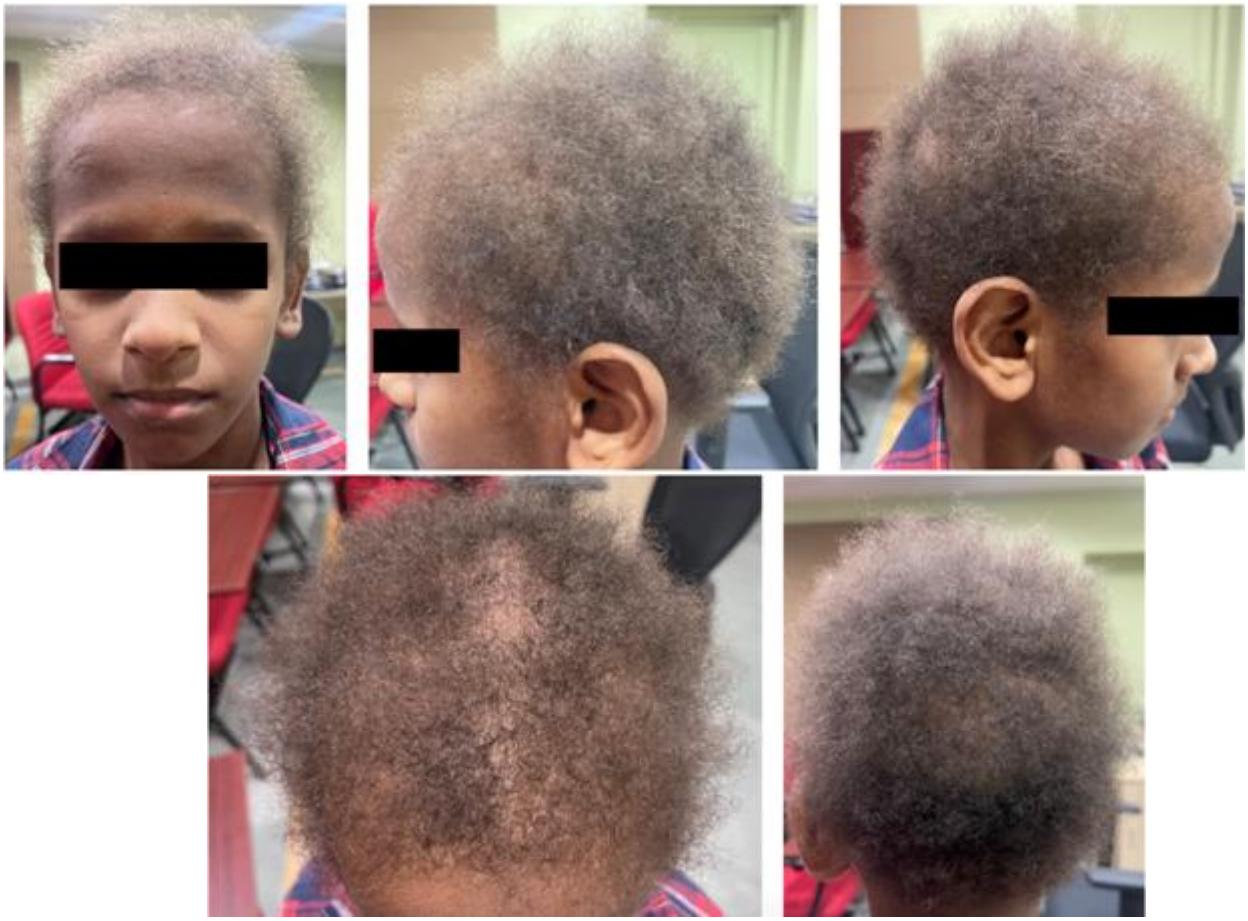


Figure 1-5: Thin, poorly pigmented, tightly coiled, sparse woolly hair present throughout the scalp.

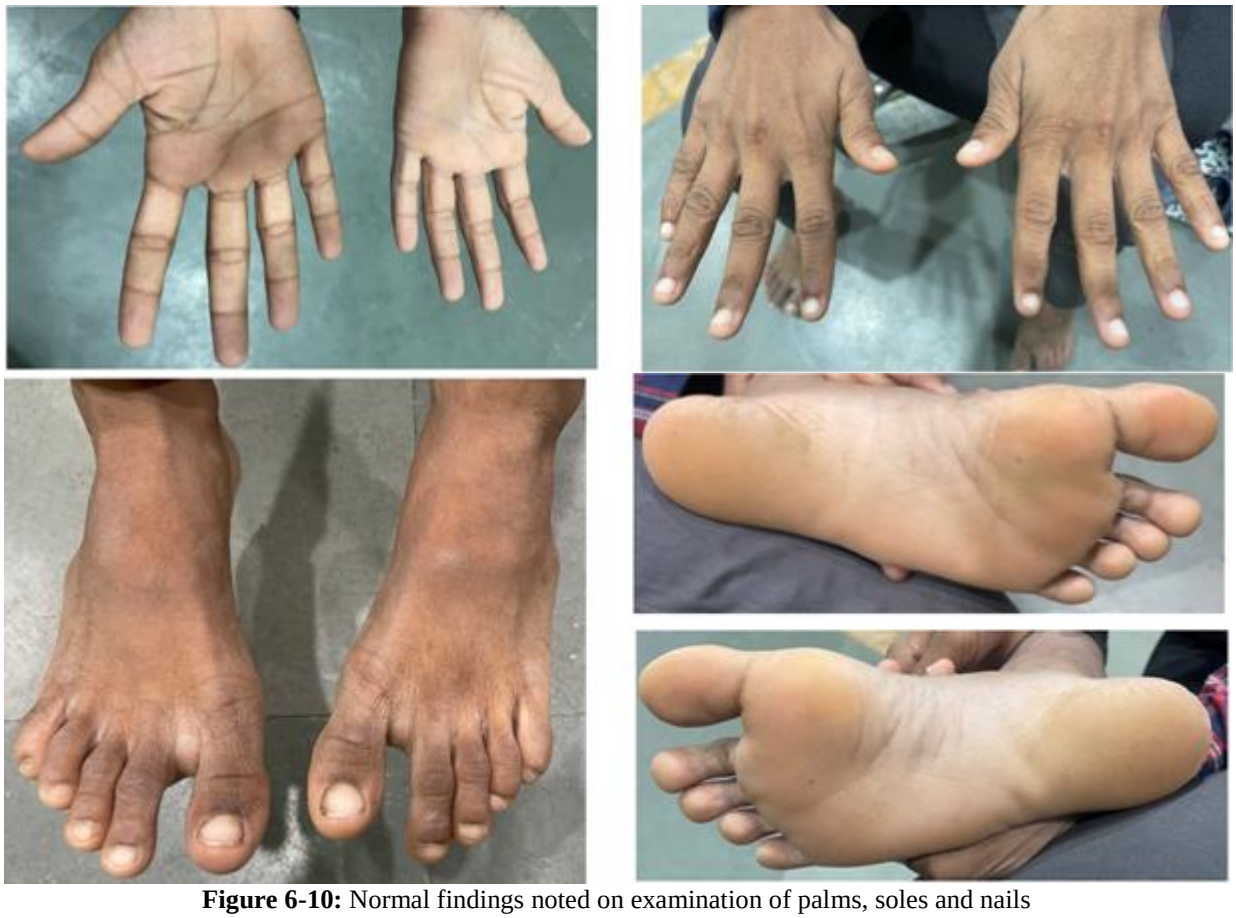


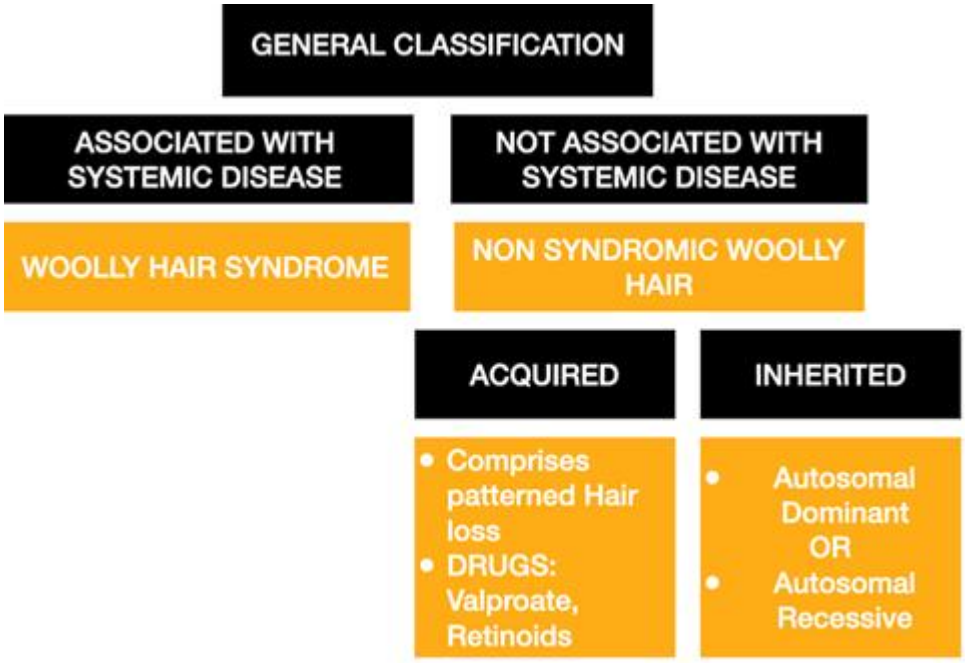
Figure 6-10: Normal findings noted on examination of palms, soles and nails

Figure 11-17: Hyper-extensibility of all the Metacarpophalangeal and Metatarsophalangeal joints of fingers and toes.

3. Discussion

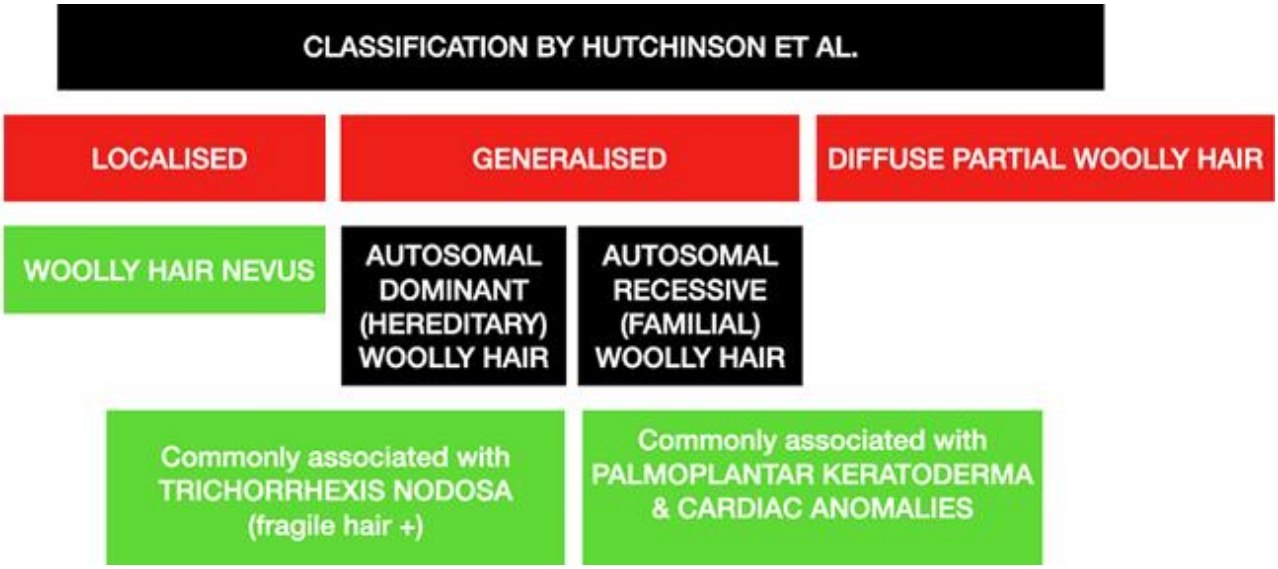
Woolly hair is an uncommon innate abnormality impacting the structure of scalp hair. This condition is characterised by tightly curled hair that can cover a portion or the entire scalp, primarily occurring in individuals of non-African origin. It was initially recorded by Gossage in 1907 within a European family. Woolly hair syndrome comprises a range of disorders marked by irregularities in the structure of the hair shaft. This leads to the distinctive feature of highly curly scalp hair, commonly observed in Asians and Caucasians.

Hereditary woolly hair is likely inherited through an autosomal recessive pattern. It involves a significant reduction in the thickness of the hair shafts, which might also lack proper pigmentation. The condition is evident from birth and is defined by tightly coiled, fine hair. However, this hair typically only grows to a length of 2-3 centimetres due to a shortened growth phase (Anagen). The hair's colour is often lighter than that of unaffected family members, and the diameter of each hair strand is smaller than typical hair. When examining a cross-section of the hair follicle, an oval shape is noticeable rather than the usual round shape. Despite a normal hair growth rate and the balance between growth (Anagen) and resting (Telogen) phases, the anagen phase is shorter in those with this condition.



Flow-Chart 1: General classification of woolly hair.

Furthermore, during this phase, the hair roots tend to be dystrophic and lack a protective covering. [1-15]



Flow-Chart 2: Woolly hair classified according to Hutchinson et al.

Already reported associations with woolly hair include Keratosis Pilaris, Palmoplantar Keratoderma, Acral Keratoderma, Recurrent Bullous Impetigo, Icthyosis, Keratosis follicular spinulosa decalvans, Scarring alopecia, Pachyonychia Congenita, Nails showingYellowish brown hypertrophy & Wedge shaped subungual hyperkeratosis, Dental caries, Dental Agenesis, Increased Inter-dental space, Malformed teeth, deafness, facial and ophthalmological abnormalities.

Syndromes associated with woolly hair are Naxos syndrome, Carvajal syndrome, Noonan syndrome and Cardiofaciocutaneous syndromes amongst others. Naxos syndrome has autosomal recessive inheritance due to mutation in Plakoglobin gene, and is characterised by woolly hair, Non-epidermolytic diffuse palmoplantar keratoderma and Dilated cardiomyopathy with right ventricular dysplasia. Carvajal syndrome has similar inheritance pattern and features of Naxos syndrome, however, it is more commonly seen in younger age group and has predominant left ventricular involvement with mutation in Desmoplakin gene. [16-23]

Various conditions can cause hyper-extensibility of joints namely Ehler Danlos Syndrome, Osteogenesis Imperfecta-Type I and IV, Marfans Syndrome, Pseudoxanthoma Elasticum, Cutis Laxa Syndromes, Pseudoachondroplasia, Spondylo-epiphyseal dysplasia congenita, and Fragile X Syndrome amongst others. Beighton score is commonly used to diagnose Benign Joint Hyper-mobility Syndrome, Ehler Danlos syndrome, Marfans syndrome and Osteogenesis Imperfecta. These conditions are differentiated based on clinical features, lab investigations and genetic testing. [24-50]

Diagnosing this condition is primarily based on recognising its distinctive clinical features. It's important to conduct a thorough examination of the whole body, with special attention given to the cardiovascular system, in order to eliminate the possibility of various syndromes.Caution should be exercised to prevent any physical or chemical actions that could harm the hair. Presently, there is no available treatment for woolly hair. However, in certain individuals, the hair might naturally become darker and exhibit reduced curliness over time. [16-23]

This case has been reported due to its extreme rarity, with no single case reported until now in previous literatures with these features to the best of our knowledge. It is also important to bring to notice the occurrence of such conditions to be more aware of these associations and for further research to be done in this direction to prevent morbidity and mortality in these patients.

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