

# Woolly hair

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## Abstract

*Woolly hair is a rare congenital abnormality of structure of scalp hair. It is marked by extreme kinkiness of hair in Caucasians. Woolly hair is either already present at birth or appears in the first months of life. The curls, with an average diameter of 0.5 cm, lie closely together and usually make the hair difficult to comb; in addition, the hair may be more fragile than usual. The hair growth rate is usually normal but the anagen phase may be truncated, with the result that the hair does not grow to be long. The hair shaft exhibits an elliptical cross section, an axial rotation and a kinked formation. A circumscribed occurrence of woolly hair in the form of a woolly hair nevus is distinguished from the forms affecting the entire scalp. The latter forms are: autosomal dominant woolly hair (hereditary woolly hair) or, far less frequently, autosomal recessive hereditary woolly hair (familial woolly hair). Autosomal recessive woolly hair can be syndromic and therefore accompanied by palmoplantar hyperkeratosis and heart abnormalities. No treatment is currently available. Traumatic physical and chemical cosmetic treatments should be avoided. Woolly hair is most pronounced during childhood; the manifestations often become less severe in adulthood.*

**Keywords:** woolly hair, kinky hair, Naxos disease, desmoplakin, plakoglobin

## Disease name

Woolly hair

## Included diseases

- Woolly hair, autosomal dominant (hereditary woolly hair)
- Woolly hair, autosomal recessive (familial woolly hair)
- Woolly hair nevus

## Definition and diagnosis criteria

The phenomenon of woolly hair was first observed and described in a European family by Gossage in 1907 [1]. Woolly hair is a congenital structural anomaly of scalp hair occurring in Caucasians. It is either sporadic or genetic. Woolly hair is extremely curly, the average diameter of the curls goes up to approximately 0.5 cm. The hair texture resembles sheep's wool, whereas the hair color most often shows no unusual characteristics [4]. Mohr and

Hoffmann compared this anomaly of the hair shaft to the structure of curly hair in Black people [2, 3]. The curled hair of Black people lies typically separately, while the curls of woolly hair tend to merge [5]. The hair is difficult to comb and often fragile, which is probably due to the simultaneous occurrence of Trichorhexis nodosa [6].

#### **The various forms of woolly hair**

Three forms of woolly hair can be distinguished:

1. autosomal dominant,
2. autosomal recessive,
3. localized woolly hair in the form of woolly hair nevus [7].

In addition, diffuse partial woolly hair, which manifests in youth and adulthood, has been described. In this form 20 to 30% of the total hair is made up of woolly hair, which is scattered over the entire capillitium alongside the normal hair. The woolly hair is short (about 5 cm), thinner in diameter, exhibits variations in caliber and shows hypopigmentation [8].

#### **Associated anomalies**

The various forms of woolly hair may occur in association with other anomalies.

A simultaneous occurrence of ocular anomalies (cataracts) with hereditary woolly hair has been reported [11, 19]. Autosomal recessive woolly hair often appears syndromically with palmoplantar hyperkeratosis and heart anomalies. A further distinction is constituted by Naxos disease, which involves right ventricular cardiomyopathy with heart-rhythm disruptions and nonepidermolytic diffuse palmoplantar hyperkeratosis (Vörner-Unna-Thost type) [12]. A further concurrent occurrence of dilated cardiomyopathy and epidermolytic striated palmopantar keratosis has been observed [13].

Van Steensel reported a family in which most members presented with woolly hair and ectodermal dysplasia in the form of nail dystrophies, acral hyperkeratosis and changes in the periodontium [24].

Woolly hair nevus can be associated with melanocytic nevi and epidermal nevi, which are typically localized on the neck [14, 15]; retarded bone growth, tooth anomalies and disturbances in the development of language were described as well [16].

Both, woolly hair that affects the entire capillitium and diffuse partial woolly hair can be accompanied by loose anagen hair syndrome. Garcia-Hernandez described a patient with woolly hair and loose anagen syndrome whose family had no known history of woolly hair. Whether this was a case of the autosomal recessive variation or of a sporadic occurrence of woolly hair remains undetermined [17]. Chapalain observed two patients with diffuse

partial woolly hair and loose anagen hair syndrome [18].

#### **Differential diagnosis**

- Acquired progressive kinky hair
- Allotrichia circumscripta symmetrica
- Acquired partial kinky hair
- Drug-induced kinky hair

#### **Frequency**

Woolly hair is a rare disorder. No epidemiological studies on its frequency are available.

#### **Clinical description**

##### **Autosomal dominant woolly hair (hereditary woolly hair)**

Several genealogies of families with hereditary woolly hair have been described [2, 7, 9, 10, 11, 20]. Patients with this form exhibit strongly curled hair at birth or in the first few months of life. The curl diameter goes up to approximately 0.5 cm, the hair is difficult to brush and partially breaks off. Growth rate is normal, while the anagen phase can be interrupted or shortened. In cases of truncated anagen phase or simultaneous occurrence of Trichorhexis nodosa, which is responsible for increased hair fragility, the hair does not grow to be very long [6, 7]. The average diameter of the hair shaft can be normal or reduced and the hair color is typically normal, although several cases of hypopigmentation of the hair have been described [2, 7]. The woolly hair is most evident in childhood; in adulthood the severity of the disorder can lessen, and then adults often exhibit markedly wavy hair. Body hair is generally not affected by the occurrence of hereditary woolly hair.

##### **Autosomal recessive woolly hair (familial woolly hair)**

Familial woolly hair is present at birth. Tightly-curved, thin-caliber hair most often grows to be only 2 or 3 cm long due to the truncated anagen phase [7]. The hair is normally lighter than that of the unaffected family members, and in some cases it is white-blond [7, 22, 23]. The body hair is short, light, and relatively sparse and a rarefaction of lateral eyebrows is observed [7].

##### **Woolly hair nevus**

Woolly hair nevus was first described by Wise in 1927 [25]. In this variation woolly hair occurs in one or more circumscribed location(s) on the scalp. The remaining hair on the head exhibits no peculiarities. In some cases the hair color can be lighter than that of the healthy hair. In half of all cases woolly hair nevi are associated with lineal epidermal nevi. Retinal anomalies, retarded bone growth, teeth anomalies and disturbances in the development of speech have also been described [14, 15, 16].

### **Naxos disease**

In Naxos disease, which is associated with familial woolly hair, diffuse non-transgradient, non-epidermolytic palmoplantar keratoses occur alongside the woolly hair itself. These keratoses are sharply-defined and often exhibit fissures (Keratosis palmoplantaris Vörner-Unna-Thost). Additionally, patients present with right ventricular cardiomyopathy with disruptions of the heartbeat [12].

### **Dilated Cardiomyopathy with woolly hair and palmoplantar keratoderma**

In the patients observed by Carvajal-Huerta *et al.*, autosomal recessive hereditary epidermolytic striated palmoplantar keratosis and a dilated cardiomyopathy occurred along with woolly hair [13].

### **Management and treatment**

No treatment for woolly hair is currently available. Physically and chemically, traumatic cosmetic measures should be avoided.

Woolly hair is most evident in childhood; the manifestations often become significantly less severe in adulthood, especially in the autosomal dominant hereditary forms.

In syndromic occurrence of palmoplantar keratoses a symptomatic treatment involving the mechanical abrasion of the hyperkeratosis and moisturizing measures are effective. In the case of heart anomalies intensive internal and cardiologist care is necessary. The pharmaceutical treatment of arrhythmias and anticoagulants as a prophylactic measure for embolic thrombosis are recommended; in cases of disturbed stimulus conduction it may be necessary to consider implanting a pacemaker. The final option for treating therapy-resistant dilated cardiomyopathy (NYHA IV) is a heart transplant.

Depending on their size and location, woolly hair nevi can be excised. A serial excision is recommended for larger nevi.

### **Etiology**

Autosomal dominant hereditary woolly hair is more commonly encountered than the autosomal recessive form. The mode of transmission of the latter is still debated [7, 21].

The etiology of diffuse partial woolly hair and of sporadically occurring woolly hair nevus is unknown. A follicular mosaicism can likely be assumed, while an autosomal dominant transmission is also discussed for diffuse partial woolly hair [8].

### **Diagnostic methods**

A thorough dermatological examination with an evaluation of the entire integument including the hair, nails and cuticles is the basic prerequisite

for making a diagnosis and probably can draw attention to an associated syndrome.

The examination of the hair shafts is carried out by light and electron microscopy, revealing the elliptical cross section, variations in caliber, axis rotation and kinked formation as well as non-homogeneous keratinisation. In some cases Trichorrhexis nodosa is evident [7, 18, 26].

If necessary the anagen/catagen ratio can be determined using a trichogram. A scalp biopsy is generally not necessary. In cases of diffuse partial woolly hair an increase in intermediate follicles can be detected histopathologically [27]. An ophthalmologic examination is recommended for all patients with woolly hair.

If the presence of a syndrome is suspected, an extensive internal and especially cardiologist diagnostic investigation is necessary. Genetic analysis can be carried out if the gene locus is known. This is only the case for Naxos disease.

### **Genetic counseling**

A gene locus for hereditary woolly hair is yet to be found. The genetic evidence for an autosomal recessive transmission is incomplete, but seems obvious in view of the occurrence of woolly hair in a sibling pair with healthy parents [7, 21].

The gene locus responsible for Naxos disease has been discovered. It entails a plakoglobin genetic defect located on chromosome 17q21 [12].

A desmoplakin genetic defect located on chromosome 6p23-24 accounts for dilated cardiomyopathy in combination with familial woolly hair and epidermolytic striated palmoplantar keratoderma [13].

### **Unresolved questions**

- The mode of transmission for the forms of woolly hair affecting the entire scalp, especially that of autosomal dominant woolly hair is known. At present it has not been possible to assign a gene locus to hereditary woolly hair, in contrast to the syndromes associated with autosomal recessive woolly hair. The question of whether diffuse partial woolly hair is a subform of hereditary woolly hair or if it must be viewed as a separate form remains unsolved.
- The etiology of the sporadically occurring woolly hair nevus is also unclear.

### **References**

1. Gossage AM: The inheritance of certain human abnormalities. *Quart J Med* 1907, 1: 331-347
2. Mohr OL: Woolly hair, a dominant mutant characteristic in man. *J Hered* 1932, 23: 345-352
3. Hoffmann E: Über einen Kräuselnäevus innerhalb sonst glatten Kopfhaares im Vergleich

zum erblichen Kraushaar und zur Lockenbildung nach Röntgenepilation. *Dermatologica* 1953, 197, 281

**4.** Schokking CP: Another woolly hair mutation in man. *J Hered* 1934, 25:337-340

**5.** Anderson E: An American pedigree for woolly hair. *J Hered* 1936, 27, 444

**6.** Rook AR, Dawber RPR: in: *Haarkrankheiten, Diagnose und Therapie*, Blackwell, p. 262

**7.** Hutchinson PE, Cairns RJ, Wells RS: Woolly hair: clinical and general aspects. *Trans St. John's Hosp Derm Soc* 1974, 60: 160-177

**8.** Ormerod AD, Main RA, Ryder ML, Gregory DW: A family with partial woolly hair. *Brit J Derm* 1986, 116, 401-405

**9.** Mori A: Contributio allo studio della eredità. *Ital Anat Embriol* 1925, 22, 123-135

**10.** Sanders J: Eine Familie mit Kraushaar *Genetica* 1936, 18 : 97-104

**11.** Taylor AEM: Hereditary woolly hair with ocular involvement. *Brit J Derm* 1990, 123: 523-525

**12.** Mc Koy G, Protonotarios N, Crosby A: Identification of a deletion in plakoglobin in arrhythmogenic right ventricular cardiomyopathy with palmoplantar keratoderma and woolly hair (Naxos-disease). *Lancet* 2000, 355: 2119-2124

**13.** Carvajal-Huerta L: Epidermolytic palmoplantar keratoderma with woolly hair and dilated cardiomyopathy. *J Am Acad Dermatol* 1998, 39: 418-421

**14.** Grant PW: A case of woolly hair naevus. *Arch Dis Childh* 1960. 35, 512-514

**15.** Jacobsen KU, Lowes M: Woolly hair naevus with ocular involvement. *Dermatologica* 1975, 86, 117-122

**16.** Al-Harzozi SA, Mahmoud SF: Woolly hair nevus syndrome. *J Am Acad Dermatol* 1992, 27: 259-260

**17.** Garcia-Hernandez MJ, Price VH, Camacho FM: Woolly hair associated with loose anagen hair. *Acta Derm Venereol* 2000, 80(5): 388-389

**18.** Chapalain V, Winter H, Langbein L, Le Roy JM, Labreze C, Nicolic M, Schweizer J Taieb A: Is loose anagen hair syndrome a keratin disorder? A clinical and molecular study. *Arch Dermatol* 2002, 138(4): 501-506

**19.** Werner S: Erblicher Star und feingelocktes Haar bei mehreren Mitgliedern derselben familie. *Acta Ophtal* 1928, 6, 382-389

**20.** Verbov J: Woolly Hair-Study of a Family. *Dermatologica* 1978, 157:42-47

**21.** Furando J, Gertalos MR, Fontarnau R: Woolly Hair. Estudo histologica e ultraestructurale en quatro caso. *Actas Dermosiphigraphicas* 1979, 70, 203

**22.** Gottheil WS: Peculiar woolly hair. 1919, 37, 489

**23.** Sweitzer SE: Woolly Hair naevus. *Achives of Dermatology and Siphiology* 1948, 58, 643

**24.** Van Steensel MAM, Koedam MI, Swinkels OQ, Rietveld F, Steijnen PM: Woolly hair, premature loss of teeth, nail dystrophy, acral hyperkeratosis and facial abnormalities: possible new syndrome in a Dutch kindred. *Brit J Derm* 2001, 145(1): 157-161

**25.** Wise F: Woolly hair nevus. A peculiar Form of birthmark of hair of the scalp, hitherto undescribed, with report of two cases. *Med J Rec* 1927, 125, 545-547

**26.** Reda AM, Rogers RS, Peters MS: Woolly hair nevus. *J Am Acad Dermatol* 1990, 22: 377-380

**27.** Guidetti MS, Fanti PA: Diffuse partial woolly hair. *Acta Dermatol Venereol* 1995, 75: 141-142