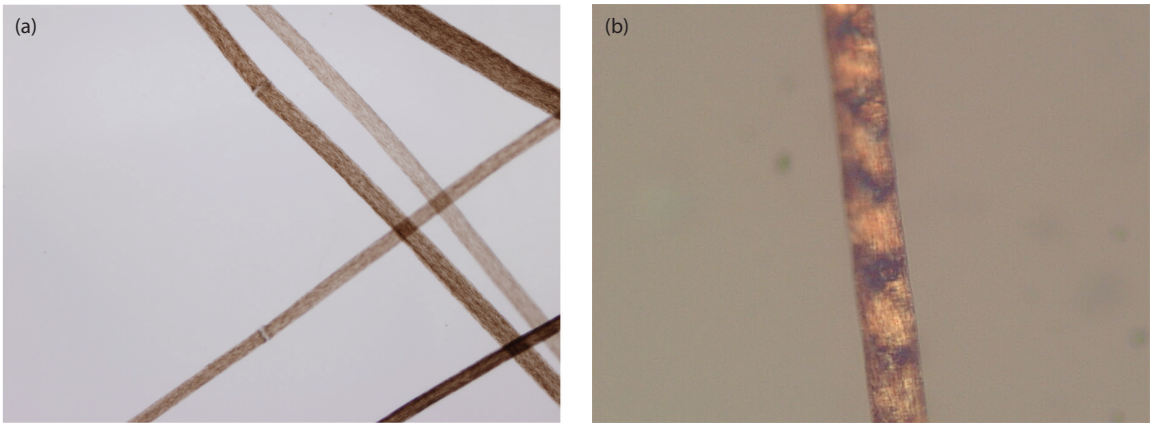

Figure 4.13 **(a) Flat hair with a typical transverse fracture. (b) Spotted and tiger-like hair surface.**

characteristic facial features, microdolichocephalia, and pleasant attitude. Other clinical findings may include mental retardation, growth delay, onychodystrophy (nail striae), and photosensitivity.[52]

Optical microscopy. Flat hair appears with typical transverse and clean transversal fractures (trichoschisis) (Figure 4.13a).

Polarized light. Alternating light and dark bands in the hair are observed, with a typical spotted tiger-like surface (Figure 4.13b).

Scanning electron microscopy. Completely rigid flat hairs like tapes are observed. Trichoschisis with a clean fracture is pathognomonic. The hair surface shows longitudinal crests and cuticle defects (Figure 4.14).

X-rays microanalysis. Sulfur content is lower than 50%.

Hair amino acid chromatography. Sulfured amino acids are clearly lowered.

Trichoscopy. Dermoscopy is not characteristic except for the trichoschisis.[62] This hair shaft may show a weaving contour and nonhomogenous structures appearing as grains of sand, alternating dark and light bands, and spotted tiger-like surface.

Trichonodosis

This is a frequent but rarely diagnosed hair dysplasia in which true knots are observed along the hair shaft. It may be suspected when the hair shaft shows an angle and changes abruptly its direction.[4,63,64] This condition is more frequently observed in individuals with curly hair in association with local trauma, scraping maneuvers, and ticks.[65] It may be found in the axillae and genitalia in association with pediculosis and acarophobia. It is not yet completely understood how double or even more complex knots are originated.

Clinical diagnosis. It usually occurs as an isolated finding. Suspicious arises when a hair changes abruptly its direction due to a true knot.

Optical microscopy. A simple, double, or even a complex true knot ("tie knot") is observed (Figure 4.15).

Scanning electron microscopy. Similar images to those observed by optical microscopy are seen. Amplification may reveal cuticle defects in the true knot area (Figure 4.16).

Trichoscopy. A simple, double or complex true knot is observed. Sometimes a "tie knot" or a "sailor knot" is observed in dermoscopy.

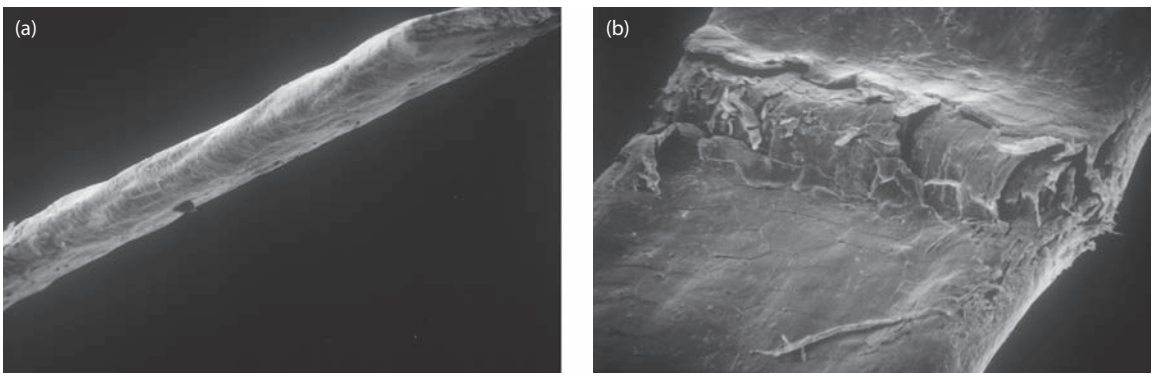

Figure 4.14 **(a) Flat hair like a tape. (b) Inicial trichoschisis at the SEM.**

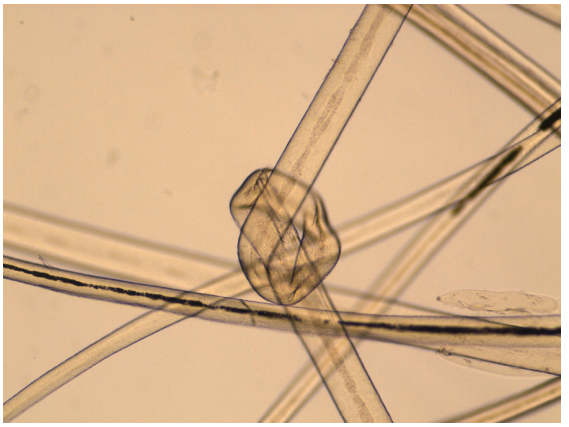

Figure 4.15 **True knot in the hair shaft. It may be simple, double, or more complex.**

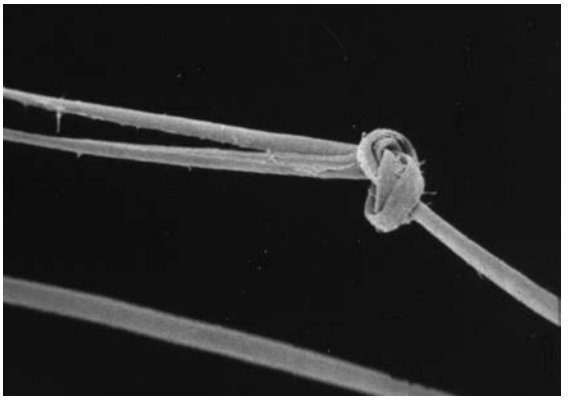

Figure 4.16 **True knot with cuticle defects in the area is observed using SEM.**

Distal trichorrhexis nodosa

This hair dysplasia is defined as a sharp fracture of the hair shaft due to a node. Clinical findings are different depending if the fracture occurs proximally or distally in the hair shaft.[66] In the former case, but not in the latter, alopecia is observed.

Distal trichorrhexis nodosa is an acquired diffuse condition, more frequently observed in long hair, and is caused by external physical–chemical factors and weathering in predisposed individuals.

Clinical diagnosis. This disorder is more frequently seen in young, long-haired women subjected to physical–chemical factors related to esthetic procedures. Hair usually shows some little balls in the distal end. The more distal, the closer the little balls appear. The hair shaft breaks at these little balls and final end of the hair shaft bifurcates; consequently, the hair of the affected individuals shows trichoptilosis and trichorrhexis nodosa.

Figure 4.17 **Long-haired individuals are more frequently affected by distal trichorrhexis nodosa. Hair shows little balls that appear as dust spots and bifurcated hair endings (trichoptilosis).**

Patients normally complain of no hair growth and that a haircut is not needed due to easy breaking off of hair when minimally manipulated (Figure 4.17).

The pull test is positive (Saboureaud sign) and hair easily breaks off at the point where little balls appear. The examiner usually gets short segments of distal broken hair shafts.

Optical microscopy. Fracture at a nodule is clearly seen, and both sides of the nodule show a chipping aspect of the cortex (Figure 4.18). When pulling apart two segments of the hair shaft, both ends look like a brush. Bifurcated hair endings (trichoptilosis) as a result of external factors are also seen.

Trichorrhexis nodosa may also be observed in other hair dysplasias, mainly in Menkes syndrome.

Electron microscopy. Absence or altered cuticle is observed along with typical images of trichorrhexis

Figure 4.18 **Fracture of the hair shaft at the nodule, with both sides of the nodule showing open spalling of the cortex.**

nodosa and trichoptilosis. The cuticle and cortex show small spalling areas close to the fracture zone at the nodule in both electron and scanning electron microscopy.[67]

Trichoscopy. Nodules where the fracture takes place are easily seen. At higher magnification, cortex fibers are also seen at the fracture point. At lower magnification, lighter nodules and slits along the hair shaft are observed. Hair endings look like brushes. Dermoscopy is a useful and fast tool to diagnose trichorrhexis nodosa.[14,62]

Treatment. It is mandatory to avoid compulsive hair treatments such as obsessive combing and brushing, as well as exposure to heat, dust, wind, salt, or other physical–chemical repetitive and intense cosmetic treatments. Finally, haircut, just some centimeters long, is advisable.

Proximal trichorrhexis nodosa

The morphological defect is the same as that observed in the distal variant; however, it is a more complex condition because trichorrhexis nodosa and trichoptilosis repetitively alternate in the same hair. It is more frequently observed in black people, who may show hypotrichosis in the scalp, beard, mustache, and other anatomical regions. It may be a hereditary hair disorder.[68,69] Proximal trichorrhexis nodosa can be a hair marker of congenital arginosuccinuria and the trichohepatoenteric syndrome.

Genetics. Trichorrhexis nodosa associated with the syndrome of Pollitt is a variant of trichothiodystrophy without photosensitivity. Gene mutations have been reported in *C7orf11* and *TFIIH*.

Clinical diagnosis. There may be alopecic areas in the scalp, beard, mustache, axillae, and mons pubis. Hairs look short and broken, with endings that show little balls appearing like dust spots (Figure 4.19). It may be easily confused with trichotillomania and "bubble hair."

Optical microscopy. Typical images of trichorrhexis nodosa and trichoptilosis are observed (Figure 4.20).

Figure 4.19 **Proximal trichorrhexis nodosa is seen as a hypotrichotic area with short, broken hairs.**

Figure 4.20 **Trichorrhexis nodosa and hair shaft fracture at the nodule with spalling of the cortex distally is seen.**

Electron microscopy. Complex images of trichorrhexis nodosa and trichoptilosis are observed in the same area and associated with cuticle defects in both electron microscopy (EM) and scanning electron microscopy (SEM) (Figure 4.21).

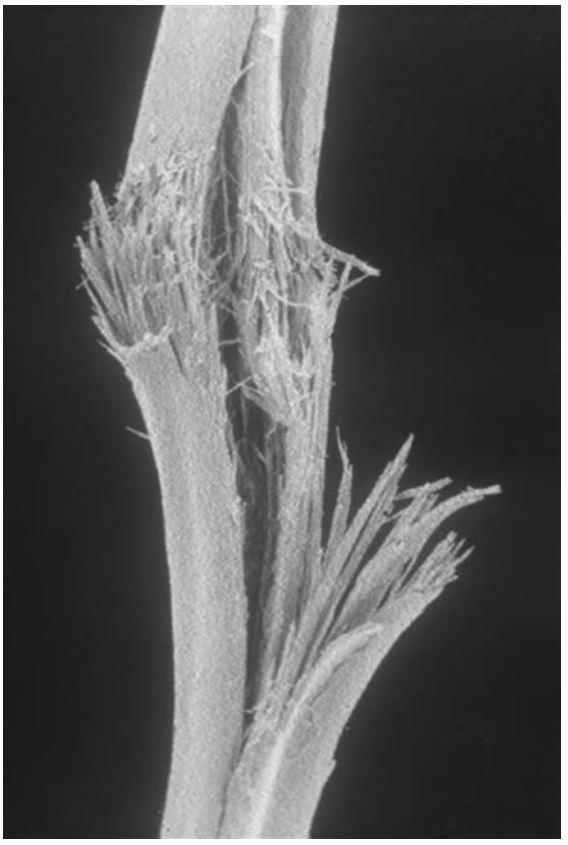

Figure 4.21 **Complex images of trichorrhexis nodosa and trichoptilosis are frequently seen.**

Trichoscopy. As stated in the previous section, dermoscopy shows complex images of some nodules and fractures in the same hair shaft, but always proximally.

Bubble hair

This acquired hair dysplasia is caused by air bubbles trapped within the hair—a situation that may be due to intense hair drying in predisposed individuals.[70–72] It is more common in women who show a localized hypotrichotic plaque. Dry heating may widen hair medulla and cause cortex dilatations, which appear as bubbles in the hair shaft surface. The hair shaft may break off in its widest ballooning portion.

Clinical diagnosis. This condition may be suspected in women with a sudden hypotrichotic plaque with short, broken hair without little balls in the hair endings. Predisposed individuals usually admit intense and close hair drying.

Optical microscopy. The typical image is that of a ballooning hair shaft urface with trapped air bubbles.

Scanning electron microscopy. A swollen or ballooning aspect of the hair shaft due to cortex dilatation is seen. Superficial orifices and fissures may also be present. The hair ending shows an image of "Gruyère cheese" due to bubbles or cavitations. Bubbles may also be found between cuticle layers.[73]

Trichoscopy. Either confluent or ballooning at different points of the hair shaft s observed (Figure 4.22).

Treatment. It is necessary for patients to avoid the cause—intense and frequent hair drying.

Figure 4.22 **In *bubble hair*, either confluent or ballooning at different points of the hair shaft is observed.**

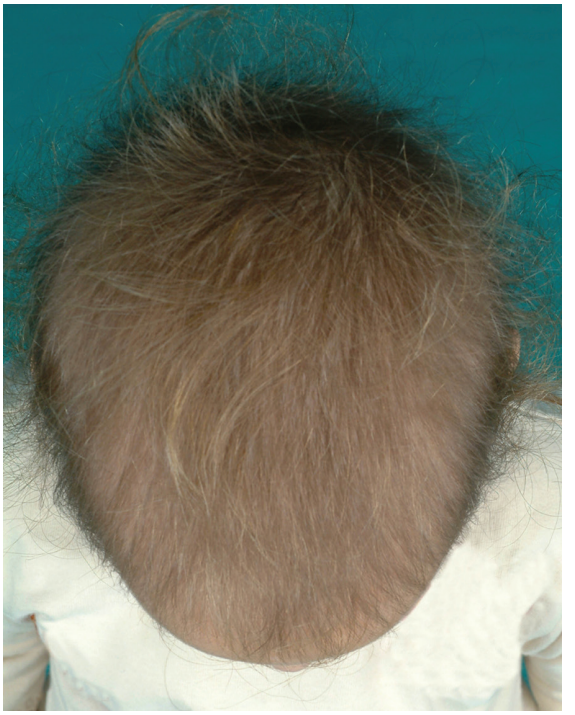

Figure 4.23 **This hair dysplasia is observed in 3- to 6-year-old blonde girls who show spontaneous, easy, and painless hair loss, especially when minimally pulled. The condition improves with time.**

Loose anagen hair

Loose anagen hair is a hair dysplasia due to lack of adhesion of the hair shaft to the hair follicle. It is more frequently observed in young blonde girls in whom hair is lost easily and with no pain when pulled.[74–76] Affected hair shows a ruffling aspect in its intrafollicular portion.

Genetics. Gene mutations have been reported in *SHOC2* and *KRT75*.[77,78] The former encodes leucine-rich repeat-containing protein, which participates in protein–protein interaction during cascade activation of kinases (RAS/ERK MAP). This mutation is responsible for Noonan-like syndrome with loose anagen hair. *KRT75* encodes a family of type 2 keratins that participate in the formation of hair and nails. Its mutation causes *pseudofolliculitis barbae* and loose anagen hair. Genes have been identified in 10q.25 and 12q.13, respectively.

Clinical diagnosis. It is more frequently observed in 3- to 6-year-old blonde girls. Hair is lost easily and without pain when pulling. Parents describe spontaneous, diffuse hair loss, especially when it is minimally pulled, and a haircut is not necessary because hair seems not to be growing farther than a certain length (Figure 4.23). Familiar cases have been seldom reported and may be associated to ectodermic dysplasias.[79]

Optical microscopy. Twisted anagen roots and proximal cuticle ruffling are observed (Figure 4.24).

Scanning electron microscopy. Twisted anagen roots with cuticle ruffling limited to the intrafollicular portion are observed. *Pili canaliculi* may also be seen.

Trichoscopy. Twisted anagen roots with cuticle ruffling may be observed.

Prognosis. No treatment has been reported. However, the condition may improve spontaneously with time.

Pili annulati (ringed hair)

Pili annulati or "morse alphabet" hair is perhaps the only condition that affects the hair medulla, which shows alternating dilatations that give the hair shaft a ringed appearance. Alternating dark and bright bands are observed in the hair shaft due to trapped air in the cortex and medulla (dark bands) and keratin distortions that diffuse incident light (bright bands).[1,4] This is either an autosomal dominant trait or a sporadic condition that affects individuals

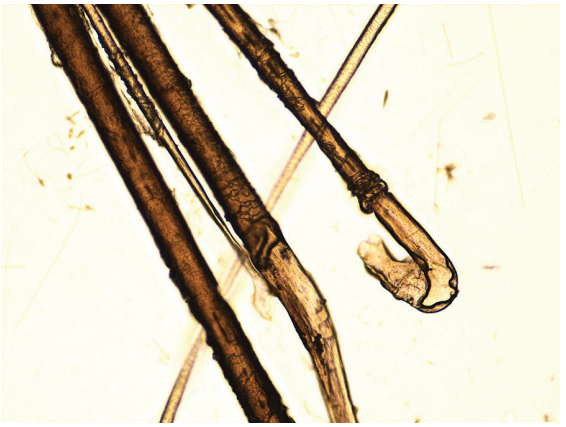

Figure 4.24 **Twisted anagen hair with cuticle ruffling is observed.**

with thick and apparently normal hair. In either case, this is an esthetic condition that may be well accepted by the affected individual. A delay in hair growth and a low cystine content that may lead to an alteration of the formation of the microfibrils complex have been reported.

Genetics. The gene responsible for this hair dysplasia has been reported to be in the region 9.2 cM of chromosome arm 12q between D12S367 and D12S1723.[80]

Clinical diagnosis. This hair condition affects young females with thick, normal appearing hair that shows alternating dark and bright bands that may appear as sequins, like in pili torti, but without using any light source (Figure 4.25).

Optical microscopy. Regular alternating dark (cavitations with trapped air in the medulla) and bright bands are seen. Polarized light clearly reveals these bands (Figure 4.26).

Figure 4.25 **Alternating dark and light bands are seen in ringed hair in females with thick and bright normal appearing hair.**

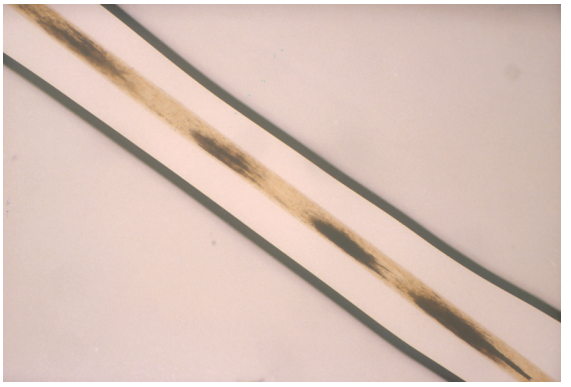

Figure 4.26 **Regular alternating dark and light bands are clearly seen.**

Electron microscopy. Alternating dark and bright bands are observed in electron microscopy and scanning electron microscopy. As in optical microscopy, dark bands correspond to cavitations in the medulla and a thinner cortex. The cuticle shows little cavitations that lead to an increased permeability and trapped air within the hair.[81–83]

Trichoscopy. Light bands correspond to trapped air in cavitations in contrast to what is seen in optical microscopy in which they appear as dark bands. Individuals with normal, thick hair may show light bands, pseudopili annulati, that correspond to less than 50% of the hair width.[14]

Pseudopili annulati

This is considered a variation of normal hair in which an optical/physical defect leads to the presence of bands, as those observed in *pili annulati*, but with no hair fragility. Bands are the consequence of light that is reflected in a straight angle by flat and twisted segments of the hair shaft hat act as little mirrors.[40,84]

Clinical diagnosis. Dark- or golden-haired children show alternating dark and bright bands. So far no hereditary cases have been reported.

Optical microscopy. Bands are observed only if light shines perpendicularly on the longitudinal axis of the hair.

Polarized light microscopy. This tool reveals only variability in hair shaft diameter without alternating bands, a finding that contrasts with true *pili annulati*.

Scanning electron microscopy. The hair shaft cross section is elliptical, while the long axis shows wider and narrower segments. Moreover, alternating 30°–40° twisting in two different directions is observed.[85]

Trichoscopy. A variation in hair shaft diameter is observed. No alternating bands are present as in true *pili annulati*.

Generalized woolly hair

Woolly hair is defined as the presence of curly, flat, and thinner than normal hair in Caucasians in contrast to black individuals in whom hair is normally flat and curly. Three different groups of woolly hair are recognized: (a) generalized or diffuse woolly hair (congenital and AD or AR), (b) woolly hair nevus (congenital and sporadic, localized or multifocal, associated or not), and (c) acquired woolly hair (progressive curly hair, partial and diffuse, mainly). The generalized or diffuse variant rarely occurs and may affect the entire scalp. It may be associated with alteration of keratinization, and eye, teeth, and bone defects, among others.[86–88]

Genetics. Autosomal dominant variants are due to a mutation of *KTR75* (12q13, 20.91 MB between D12S1301 and D12S1610).[89] Autosomal recessive variants are caused by mutations in *P2RY5* (13q14.2-q14.3), *LIPH* (3q27-q28), and *LAH2* (3q27-q28).[90] In Naxos disease, woolly hair is associated with cardiomyopathy and keratoderma of the palms and soles and is caused by the mutation of the plakoglobin gene (17q.21).[91,92]

Clinical diagnosis. Children normally present with very thin, soft, and curly hair that resembles wool. Hair density is low, and the scalp can be seen very easily (Figure 4.27). Hair fragility may lead to some hypotrichotic areas. This condition should be differentiated from uncombable hair, a distinctive hair dysplasia with which this has been confused in the medical literature.

Optical microscopy. Thin and curly hairs form small balls.

Histopathology. Groups of miniaturized anagen hair follicles that appear in normal quantity are observed.

Scanning electron microscopy. Very flat, tagliatelle-like, curly hair with an oval cross section is revealed by SEM (Figure 4.28). It may also been found isolated twisting and *pili canaliculi*.

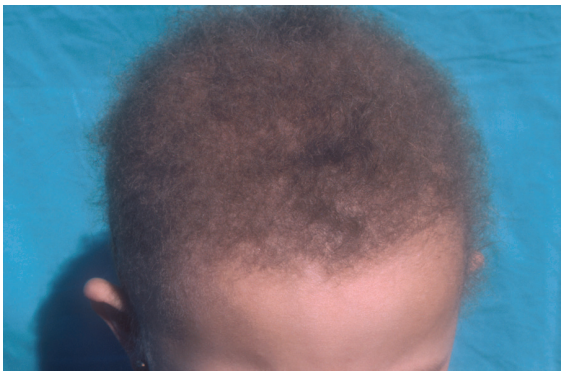

Figure 4.27 **Characteristically diffuse woolly hair is presented in a child with soft, curly hair that leads to scarce density that enables the examiner to easily see the scalp.**

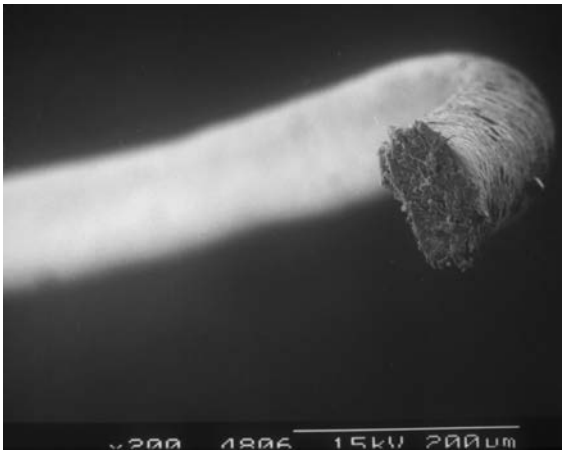

Figure 4.28 **In woolly hair the hair shaft is very flat, tagliatelle-like, with an oval cross section.**

Trichoscopy. Rudnicka et al.[62] have reported cyclical short, wavy hair that recalls a characteristic "moving snake" image. Broken hairs are also seen due to fragility caused by longitudinal twisting (Figure 4.29).

Woolly hair nevus

This is a localized, congenital, nonfamilial variant of woolly hair. It can be isolated or multifocal. This is the most frequently seen clinical form of curly/kinky hair syndromes.[93] Approximately 50% of cases are associated with melanocytic nevus or epidermal nevus in the same affected area or in the head.[94] Other alterations may include ocular (cataracts, retinal dysplasia, persistent pupillary membrane), and teeth defects, as well

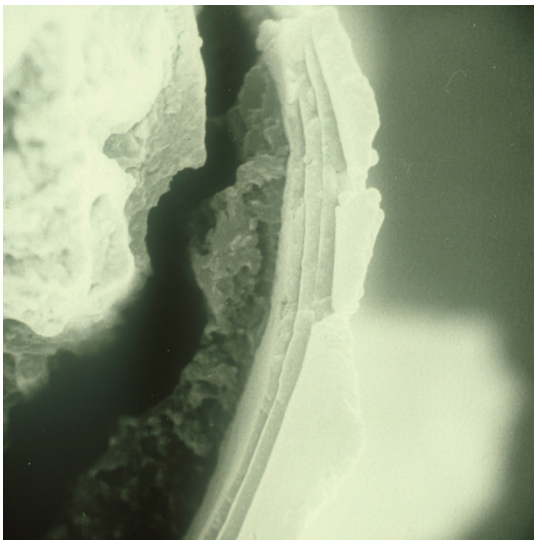

Figure 4.29 **Woolly hair at the SEM present also a thinkiness of hair cuticle.**

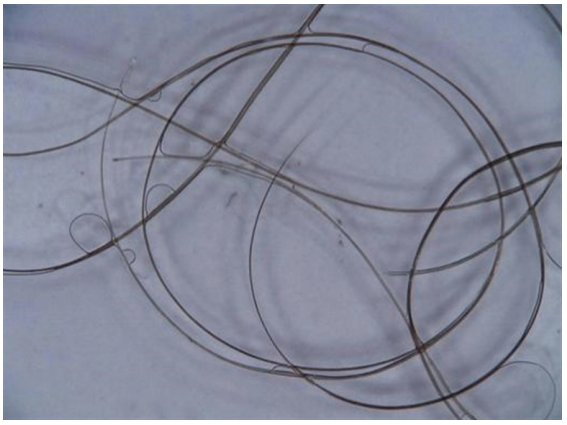

Figure 4.30 **At the trichoscopy, woolly hair is observed as a cyclical short, wavy hair that recalls a characteristic "moving snake" image; longitudinal twisting increases hair fragility and leads to broken hair.**

as gingivitis, growth and speaking delay, and congenital triangular alopecia.[95] No genetic findings have been reported.

Clinical diagnosis. One or some areas of woolly, curly, thin, and light hair are observed, surrounded by normal hair. An isolated plaque is the most frequent presentation, but multifocality is occasionally reported (Figure 4.30). Melanocytic and epidermal nevi may be present in the same affected area or other areas of the head.

Optical microscopy. Under optical microscopy, hair looks very thin, flat, light, and as forming a ball. The cross section is oval.

Histopathology. An area of grouped miniaturized anagen follicles are observed.

Scanning electron microscopy. The hair appears flat and curly, and the cross section is oval. Longitudinal twisting and channels along the axis are also observed. Fewer cells in cuticle layers may be found.

Trichoscopy. Similar findings to those found in generalized woolly hair are observed, but in a circumscribed area.

Treatment. Being a localized hair dysplasia, surgery is a therapeutic alternative to be considered.

Acquired and progressive kinking hair

Acquired and progressive kinking hair is an acquired nonfamilial variant of woolly hair. It frequently affects male teenagers. Locks of hair become curly and kinky, a condition that slowly but progressively affects the entire scalp. No external factors have been identified, and the condition slowly progresses to androgenetic alopecia.[96–98] A localized variant has also been reported; it is whisker hair nevus or circumscribed symmetric allotrichia. Systemic retinoids have been reported as a possible cause

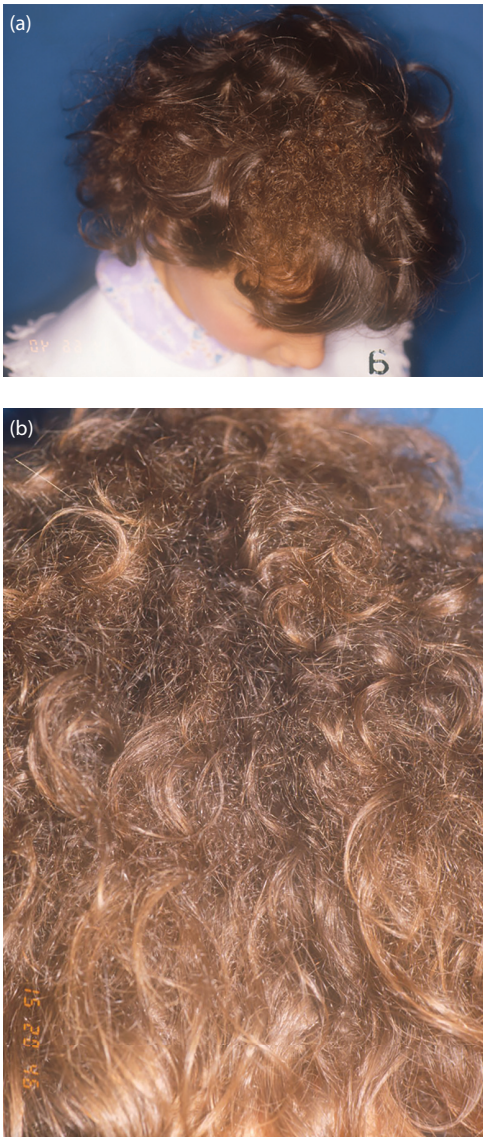

Figure 4.31 **(a) In woolly hair nevus an area of woolly, fine, thin, and curly hair surrounded by normal appearing hair is observed. (b) In acquired progressive kinking of hair multiple locks of hair acquire a curly and kinky features.**

of acquired and progressive kinky hair. No genetic findings have yet been reported.

Clinical diagnosis. Typically it is a male teenager that shows locks of acquired thin, kinky hair (Figure 4.31).

Optical microscopy. Curly, flat hair that appears oval in the cross section is observed.

Histopathology. The cross section of the specimen shows oval hair follicles (Figure 4.32).

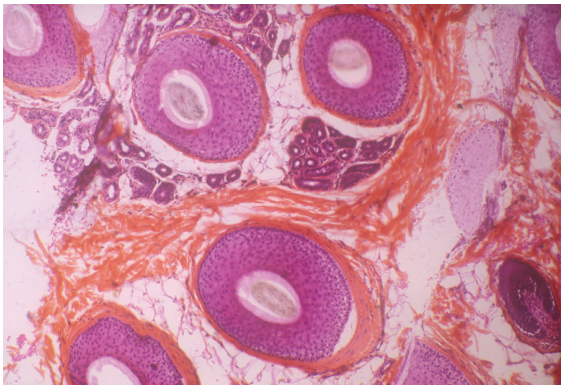

Figure 4.32 **In those cases, a cross section shows oval hair follicles.**

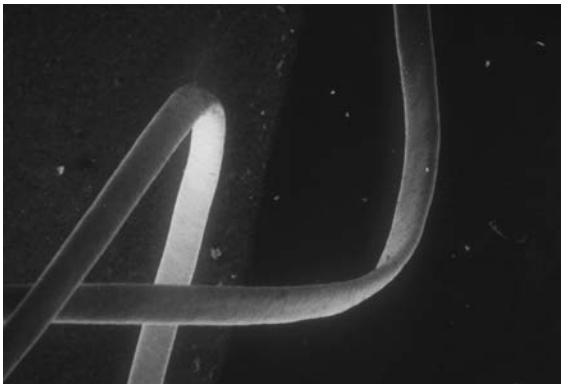

Figure 4.33 **Affected hairs are flat, like a tagliatelle, thinner, curly, and oval in the cross section.**

Scanning electron microscopy. Affected hair looks very flat like tagliatelles, thinner, and curly; the cross section reveals an oval hair as in congenital woolly hair, either diffuse or localized (Figure 4.33).

Trichoscopy. The same findings as in generalized or diffuse woolly hair are observed.

Diffuse partial *woolly hair*

This is a new variant of acquired kinky hair. In this condition, individual and isolated kinking occurs; consequently, only one curly kinky hair is observed amid 10–12 normal hairs. The disorder may be sporadic or familial. No external factors have been identified, and most affected individuals are children or teenagers.[99–100]

Genetics. Autosomal dominance may be related to mutation of *KRT74* (12q.13) as discussed previously.

Clinical diagnosis. It is observed in children or teenagers with isolated curly and kinky hair that stands out from normal hair.

Optical microscopy. Affected hair looks flat, curly, and oval in the cross section.

Histopathology. Anagen curved hair follicles are observed amid normal appearing follicles.

Scanning electron microscopy. Affected hair is thinner and flatter than normal hair, and oval at cross section. Twisting and longitudinal channels may also be present.

Trichoscopy. Affected hair is less curly, but findings parallel those described above for woolly hair nevus.

Acquired partial curly hair

This is a partial and acquired variant of kinky hair that affects isolated hair in the scalp and may be confused with diffuse partial woolly hair. This is a nonfamilial condition that affects only the distal part of the hair shaft; it improves with time and may be a response to external factors, either environmental factors or cosmetic procedures.[101]

Clinical diagnosis. The typical case is a young woman with isolated curly hairs surrounded by normal hair (Figure 4.34). This is a sporadic and acquired condition, which differentiates it from diffuse partial woolly hair.

Figure 4.34 **A child or teenager is affected by acquired partial curly hair.**

Optical microscopy. Affected hair is thinner, flatter, and curly only distally.

Scanning electron microscopy. Affected hair is flatter, thinner, and oval in cross section. Characteristically, hair is normal in its proximal portion, but acquires a thinner and curly aspect in the distal half of the hair shaft.

Trichoscopy. The same findings as seen in woolly hair nevus are observed, but only in isolated affected hairs, which appear less curly.

Treatment. Hair conditioners may help. Avoidance of weathering and aggressive cosmetic treatments is mandatory.

Straight hair nevus

Similar to woolly hair nevus, straight hair nevus has been described, which corresponds to the inverse image of the former. It is the presence of a delimited area with straight hair locks surrounded by curly hairs.[102] In the few cases that have been reported, the condition is linked to a local keratinization disorder associated with ichthyosis or epidermal nevus,[103] and it has also been reported in a patient with straight hair.[104] No genetic changes have been reported.

Clinical diagnosis. One or more locks of straight hair are observed, and characteristically they are surrounded by curly hair. However, some cases have been reported to occur in individuals who have straight hair but variable hair diameter, and associated local epidermal defects.

Histopathology. In the only case that we reported, hyperplasia of the sebaceous glands was reported, which gave a yellowish discoloration to the affected skin in an 8-year-old child.

Optical and electron microscopy. We observed minimal channels along the longitudinal axis.

X-ray microanalysis. No findings have been reported.

Trichoscopy. It is the inverse image of woolly hair; affected hair in this case appears straight.

Comment. Straight hair nevus has been reported to be associated with *ichthyosis hystrix* and warty plaques of the affected skin, independent of ethnic origin. Another plausible theory is that this condition corresponds to either hair mosaicism or a hair follicle disorder induced by the affected skin, instead of an individual and distinctive hair disease.

Uncombable hair (pili canaliculi)

This syndrome is characterized by dry, light, and slurred hair in children or teenagers with thick hair but with locks of hair in different directions that makes it impossible to comb or style.[104,105] It is also known as "fiberglass hair." This is a sporadic or familial autosomal dominant or recessive condition.[106] It is generally described in a child with straight uncombable hair, but it has also been reported to occur in curly haired young individuals. Localized variants have also been reported. In all cases, the common finding of this hair dysplasia is the presence of *pili canaliculi*. However, *pili canaliculi* may also be found in ectodermal dysplasias, other hair dysplasias, and in almost all cases of loose anagen hair.

Genetics. No genetic changes have been reported. However, in Rapp–Hodgkin syndrome (anhydrotic ectodermal dysplasia and cleft lip/palate) the mutation has been identified in *TP63* (3q.27).[107]

Clinical diagnosis. It is seen in children or teenagers who present with dry, lusterless hair that cannot be combed. This disorder can be familial (Figure 4.35).

Optical microscopy. A channel alongside the hair shaft is easily observed when adjusting the micrometer (Figure 4.36). If the hair has medulla, it is impossible to see the channel.

Histopathology. The biopsy is seldom required. Inner sheaths adhesions are observed as well as hair shaft twisting.

Scanning electron microscopy. One or more channels are observed along the longitudinal axis of the hair shaft, and this is definitive for the diagnosis (Figure 4.37).[108] Depending on the number of channels, the cross section

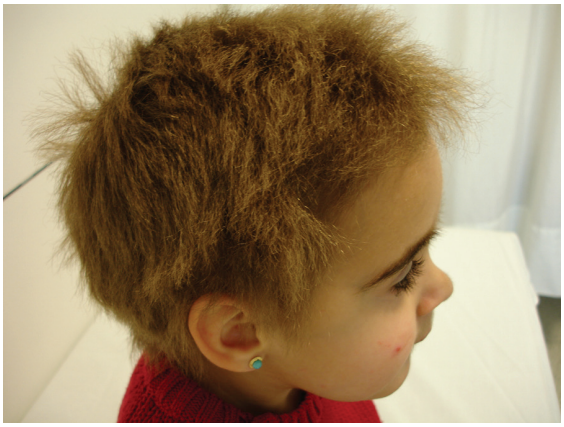

Figure 4.35 **Pili canaliculi affects a child or young individual with dry, lusterless, and uncombable hair.**

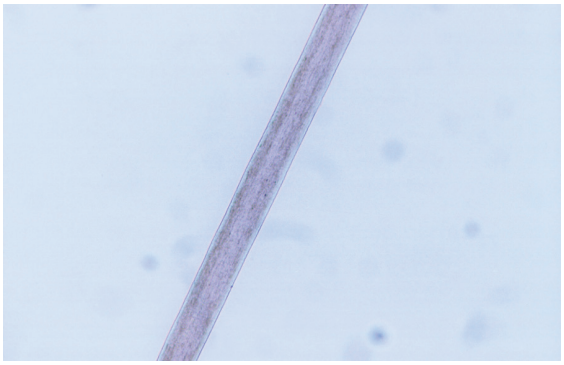

Figure 4.36 **Characteristically, a channel along the longitudinal axis of the hair shaft is observed.**

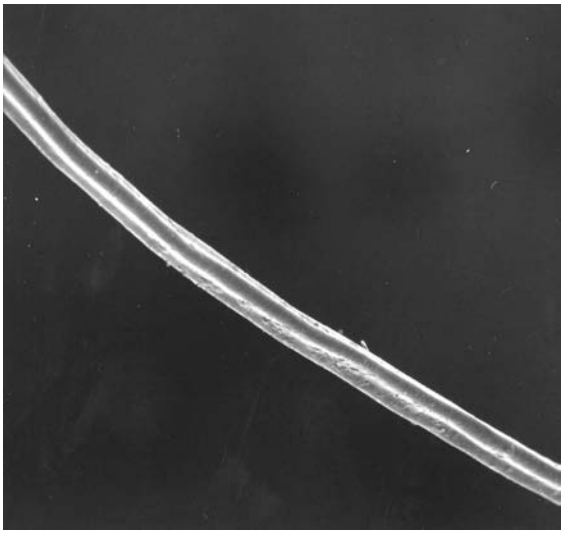

Figure 4.37 **Deep channels are clearly observed along the hair shaft.**

of the hair shaft may show a kidney-like, triangular, square, or irregular shape.[109]

Trichoscopy. More recently, this dermoscopy was demonstrated to be a useful, simple, and fast diagnostic tool. Flattening and longitudinal channels are observed along the hair shaft axis, while the cross section of the hair shows a kidney-like or triangular shape.

Treatment. Spontaneous improvement with time has been reported. Zinc pyrithione shampoo may improve hair due to the rebound fat effect.

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