Hair Shaft Disorders

Elena Nogales M.D., and Sheila M. Valentín Nogueras M.D., F.A.A.D.

	Clinical findings	Microscopic findings	Associated syndrome(s)	Inheritance/ Genetic defect	Other findings	Comment
With increased hair fragility						
Trichorrhexis nodosa (TN)		Frayed cortical fibers pushed up against each other.	Argininosuccinuria	AR/ Argininosuccinate lyase deficiency	Organ toxicity, seizures, hyperammonemic coma, neurologic damage, growth retardation.	Arginine supplementation can be beneficial in patients with less severe deficiencies.
			Citrullinemia	AR/ Argininosuccinic acid synthetase deficiency	PT, acrodermatitis enteropathica-like dermatitis.	
Trichoschisis	Eyebrows and eyelashes also affected	Clean transverse fracture. Alternating light and dark bands ("tiger-tail") seen with polarized light.	Trichothiodystrophy	AR/Nuceotide excision repair genes (XPD, XPB, TTD-A) TTDN1 (non-photosensitive TTD)	Photosensitivity (without increased risk of skin cancer), intellectual impairment, decreased fertility, short stature, progeria-like features.	Low cystine (sulfur) content of hair is postulated to account for cuticular and cortical weakness.
Trichorrhexis invaginata (TI)	Eyebrows may be affected	Distal hair shaft invaginating into the proximal hair shaft ("bamboo hair").	Netherton syndrome	AR/SPINK5 gene: Absent LEKTI (serine protease inhibitor)	PT, TN, ichthyosis linearis circumflexa, atopic diathesis, FTT, recurrent skin infections, dehydration.	Hair breakage may improve with age. Any topical medication should be used with caution due to skin barrier dysfunction.
Pili torti (PT)		Flattened and twisted at 180° angles. Fractures occur at twists.	Menkes syndrome	XLR/ATP7A deficiency	TN, Skin/hair hypopigmentation, "doughy" skin, lethargy, seizures, FTT, MR, joint laxity, vascular abnormalities.	Low serum copper and ceruloplasmin. Parenteral copper histidine in the first 8 weeks of life may be of benefit; most do not survive beyond the first decade of life.
			Björnstad syndrome	AR/BCS1-like protein deficiency	Congenital sensorineural hearing loss.	
			Crandall syndrome	AR	Congenital sensorineural hearing loss and hypogonadism.	
Monilethrix	Hair rarely grows beyond 1-2cm in length because of breakage	Nodes/ internodes at regular intervals. Breaks at internodes.		AD/hHB6, hHB1, AR/Desmoglein 4	TN, keratotic follicular papules at the nape of the neck, upper back, and arms.	Topical minoxidil and oral etretinate may improve hair growth.
Without increased hair fragility						
Pili annulati (PA)	Alternating light and dark bands; only detectable in lightly pigmented hair	Alternating light and dark bands.		AD, sporadic		Thought to be caused by formation of abnormal air cavities in the hair shaft.
Loose anagen syndrome (LAS)	Hair is easily pulled from the scalp	Anagen hairs without IRS, ruffled cuticle.		AD,sporadic/ Keratin defect (K6hf)		Most cases are blonde girls >2 years of age. Gentle hair care recommended.

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Hair Shaft Disorders (cont.)

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Without increased hair fragility						
Uncombable hair syndrome (UHS)	Dry, shiny ("spun-glass"), cannot be combed flat. Not as noticeable in dark hair	Longitudinal grooving (Pili trianguli et canaliculi) on EM.		AD		Hair tends to be more manageable with age. A positive response to biotin has been reported in a few cases.
Other						
Woolly hair (WH)	Tightly curled hair in persons of non-African ancestry		Naxos disease	AR/Plakoglobin	TN, trichoschisis, PA, PPK, arrhythmogenic right ventricular cardiomyopathy (ARVC).	
			Carvajal syndrome	AR/Desmoplakin	TN, trichoschisis, PA, striated PPK, biventricular dilated cardiomyopathy.	
			Naxos-like disease	AR/Desmoplakin	TN, trichoschisis, PA, ARVC, early-onset blistering, xerosis.	
			Woolly hair and skin fragility syndrome	AR/Desmoplakin	TN, trichoschisis, PA, early-onset blistering, PPK, dystrophic nails, alopecia.	No cardiac abnormalities.
			Diffuse partial woolly hair	AD	TN, trichoschisis, PA, cataracts, papillary membranes.	
	Hair is lighter and thinner when compared to adjacent, normal hair.		Woolly hair nevus	Sporadic	TN, trichoschisis, PA, ipsilateral epidermal or congenital nevus (50%), precocious puberty, bone/speech/dental anomalies.	
Curly hair	Large loose spiral locks		CHAND syndrome	AR	Ankyloblepharon, nail dysplasia, ataxia.	
			Trichodentosseous syndrome	AD/Homeobox protein (DLX3)	Enamel hypoplasia, otosclerosis, dolychocephaly, frontal bossing.	
			Costello syndrome	AR, AD/GTP binding protein in MAPK pathway (Hras)	Growth deficiency, MR, coarse facies, perioricial papillomas, solid tumors.	Increased risk of rhabdomyosarcoma, neuroblastoma, and transitional cell carcinoma.
			Noonan syndrome	AD/SHP-2 tyrosine phosphatase (PTPN11)	WH, dysmorphic facies, ear/ocular/cardiovascular anomalies, multiple nevi, short stature, webbed neck, keratosis pilaris atrophicans.	

**FTT= failure to thrive, MR= mental retardation*

References

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