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Case Series

Hair shaft disorders: a rare case series

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ABSTRACT

The hair shaft is composed of multiple layers, which include the medulla, cortex and cuticle. Genetic disorders, diseases or environmental factors can lead to changes in the properties of these structures, resulting in fragility or alterations in hair shaft texture, appearance or manageability giving rise to various hair shaft disorders. Depending on the fragility of the hair the hair shaft disorders are divided into those with increased fragility and those without increased fragility. A clear detailed history and accompanying microscopic, trichoscopic evaluation help in establishing a diagnosis. In this case series we presented a series of seven hair disorders which included pili torti, trichorehexis nodosa, monelithrix, plica palonica, white piedra, trichoptilosis and Tay's syndrome. Most of the hair shaft disorders have underlying metabolic disorders or genetic disorders that are left unnoticed and are commonly treated as alopecias which do not improve on treatment. Thereby creating a need of diagnosing these conditions appropriately. The treatment of hair shaft disorders depends on treating the underlying cause. In addition patients must be advised to reduce the traumatic effects on the hair like ironing, chemical treatments, curling.

Keywords: Hair shaft disorders, Pili torti, Trichorrhexis nodosa, Trichothiodystrophy, Tay's syndrome

INTRODUCTION

The hair shaft is composed of multiple layers, which include the medulla, cortex, and cuticle.

Genetic disorders, diseases or environmental factors can lead to changes in the properties of these structures, resulting in fragility or alterations in hair shaft texture, appearance or manageability.[1]

Most hair shaft disorders are not curable. Some may improve over time or with treatment of the underlying cause. In general, patients with hair fragility should avoid physical or chemical trauma to the hair to reduce additional hair breakage and loss. Hair shaft disorders can be localized or generalized. Hair shaft diseases are separated into those with and those without increased hair fragility.[1]

Table 1: Classification of hairshaft disorders.

Increased fragility	No increase in fragility
Trichorrhexis nodosa	Pili anulati
Trichorrhexis invaginta	Wooly hair
Monilethrix	Acquired progressive kinking of hair
Pseudomonelithrix	Uncombable hair syndrome
Pili torti	
Pili bifurcati	
Trichothiodystrophy	

CASE SERIES

Case 1

A 23 years old female presented to our OPD with complaints of dry, rough, brittle hair associated with pain

while combing over the crown area since 5 years. On examination patient had a localized patch of rough, brittle hair over the crown region. Microscopic examination showed 180° twist in 40× magnification. Patient was evaluated for presence of pyschomotor defects and subsequently was found to be normal. Neurological evaluation, audiometry and serum zinc levels were tested and were found normal. With the above features a diagnosis of pili torti was made.

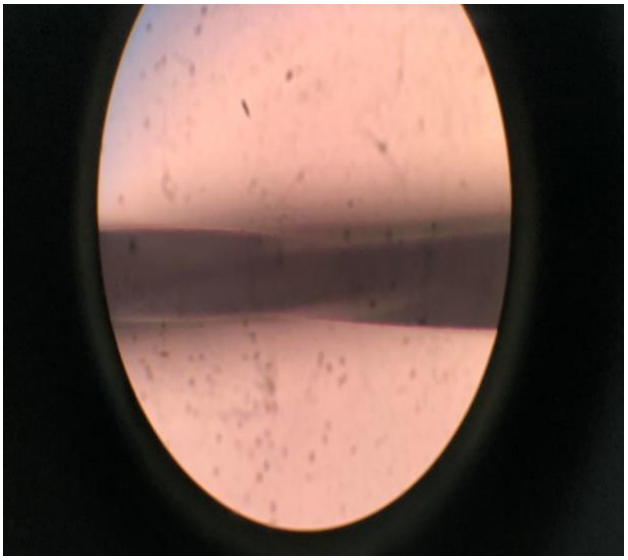

Figure 1: Light microscopy in 40× magnification of slide shows 1800 twist over the shaft.

Figure 2: Light microscopy in 40× magnification showing thrust paint brush appearance.

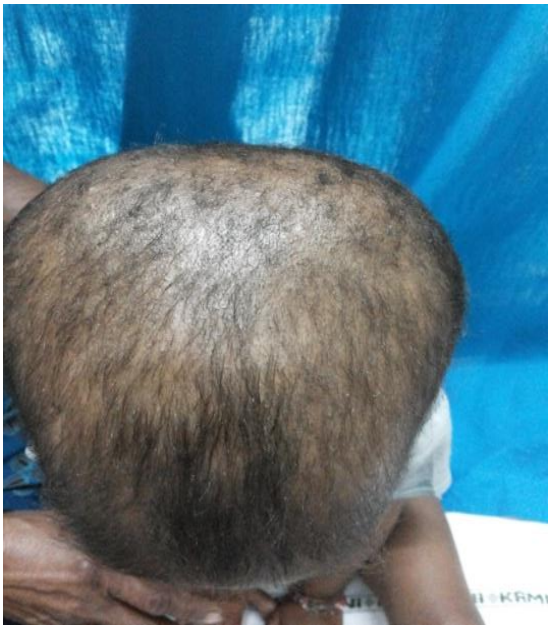

Figure 3: Sparse hair over the scalp which shows rough texture.

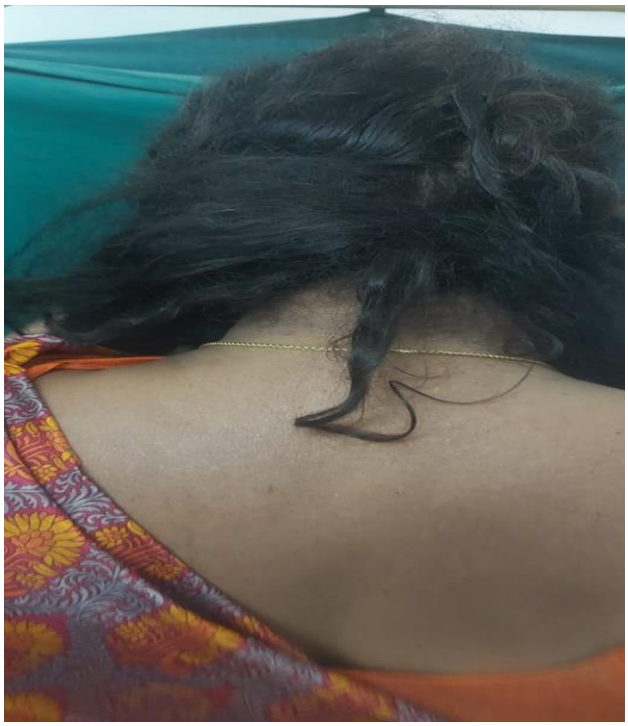

Figure 4: Matting and tufting of hair over the occipital region of scalp.

Case 2

A 29 year old male presented with dry and white spotty appearance on hair over his beard. There was pain and no itching. There was no history of trauma, itching or any topical application. Patient denied history of any manipulation. Clinical examination showed short stubs of hair with glistening tips over beard. No similar findings

were found in other hairy areas of the body. Systemic examination was unremarkable. The damaged hair sample was taken for light microscopy which showed a breakage of hair shaft with fraying ends giving the characteristic paint brush appearance. With the above history, clinical and microscopic findings a diagnosis of trichorrhexis nodosa was made.

Case 3

A 3 year old male child presented with brownish scanty hair over the scalp since 1 year of age. History of repeated tonsuring was done by parents in order to obtain stronger and thicker hair. There were no teeth, nail abnormalities noted. Examination showed short, sparse, brown, rough hair over bitemporal, frontal, vertex, occipital areas. Multiple hyperkeratotic papules were noted over the occipital region of scalp. Light microscopy showed beaded appearance of hair suggesting monelithrix.

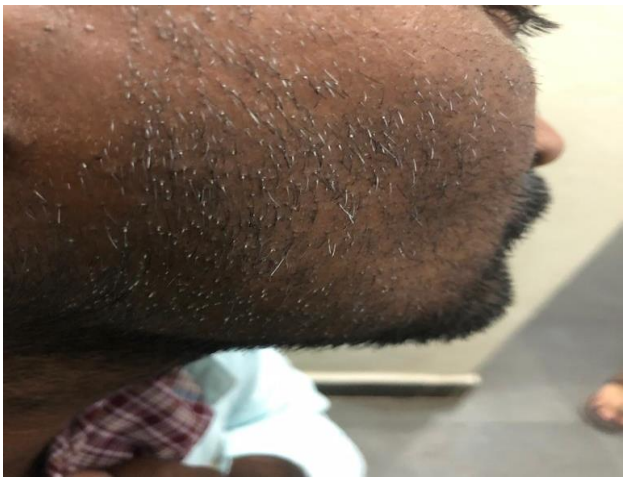

Figure 5: Multiple white glistening nodules noted over hair on the beard.

Case 4

A 44 year old female presented with tufted and entangled hair over scalp since 1 month. Patient gave history of usage of unknown herbal oil for about 6 months prior to this presentation. Patient denied history of any lice infestation. There was no history of any psychiatric illness in the patient as well as her family. On examination there was localized dry, rough, tufted hair over few regions of the occipital area. There were no scalp erosions, no infestation of lice, no nits, no discharge. On potassium hydroxide examination, no fungal hyphae were seen. With the above features a diagnosis of plica polonica.

Case 5

A 26 years old male patient came to our OPD with complaints of whitish lesions over the hair of beard since 3 months. There was no associated itching and pain. Other hairy parts of body were normal. On clinical examination, hair on the scalp was normal without evidence of any sparseness. Hair over beard showed barely visible but well-palpable whitish to cream-colored nodules which were easily detachable. On potassium hydroxide (KOH 10%) wet mount of the affected hairs showed septate hyaline hyphae arranged perpendicular to the hair shaft. The patient was advised to use topical terbinafine twice daily for 8 weeks, after which there was complete resolution.

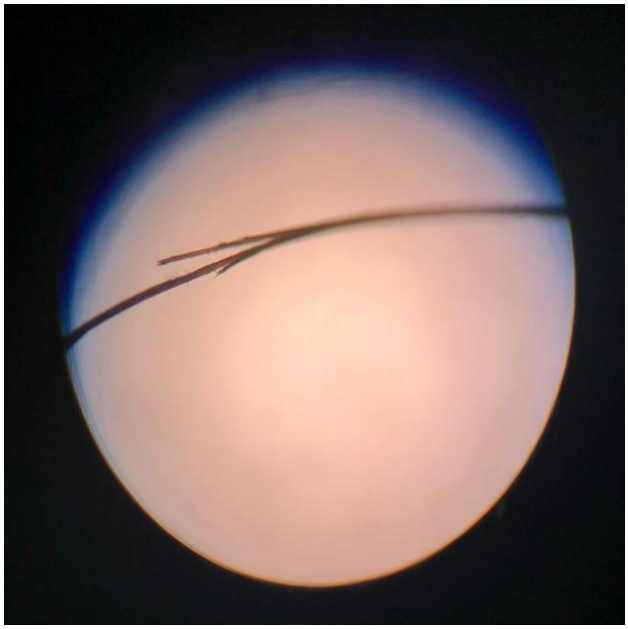

Figure 6: Light mircoscopy of hair at 10× magnification showing splitting of hair shaft at the ends.

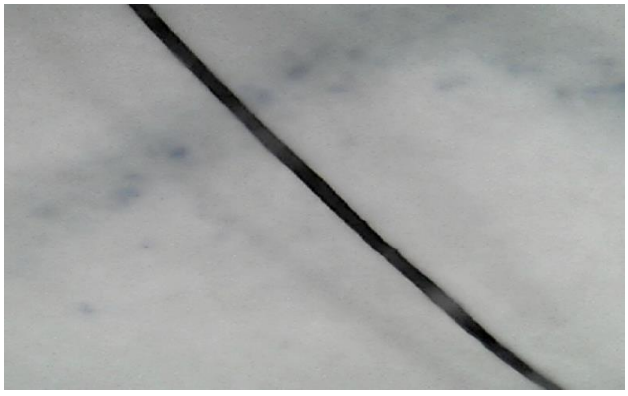

Figure 7: Hair shaft showing tiger tail appearance on trichoscopy.

Case 6

A 20 year old female patient came to OPD with complaints of loss of hair and coarse brittle hair over the scalp sine 3 months. Patient gave history of usage of curling irons and chemical treatment of hair 2 months

prior. Other hairy areas over the body were normal. On examination there was splitting of multiple hair shafts. On microscopic examination splitting of hair shaft was observed suggesting the diagnosis of trichoptilosis.

Case 7

A 8 year old girl was brought to the OPD by his mother, who was born out of second degree consanguineous marriage with complaints of dry scaly lesions over the entire body, diffuse sparse and rough hair noted over the scalp. History of colloidan baby present. There was a history of delayed developmental milestones compared to the other sibling. Other sibling had no similar complaints. On examination, there were diffuse fine white-colored scales all over the body and to a lesser extent on the face. Hair over the scalp and eyebrows was thin, short and sparse. The ophthalmic evaluation was normal. There was sparseness of hair over the lower eyelids and eyelashes. Fingernails and toenails showed longitudinal ridging. The patient showed height less than 3rd centile. Routine investigations were normal. Histopathological examination revealed features of ichthyosis. Trichoscopic examination of hair revealed alternate light and dark bands giving the appearance of tiger tail. CT and MRI were done and found to be normal. Further genetic testing revealed that the individual harbors two copies of pathogenic variant of GTF2H5 gene which was further shown to be associated with trichothiodystrophy. With the above findings and history a diagnosis of Tay's+Ibids syndrome which was a subtype, type E trichothiodystrophy was made.

DISCUSSION

The normal hair shaft showed a consistent diameter all along its length, with cross sectional shape being oval. Significant variations existed particularly in different racial groups from straight to woolly hair as well as in thickness of the hair shaft. The medulla of the hair shaft was characterized by a central cavity, but was only present in some individuals. It can be either present as an continous entitiy or intermittent. The medulla can be pigmented or remain clear on light microscopy analysis.

Hair-shaft disorders were categorized into those associated with hair fragility and those that did not affect the integrity of the hair shaft. Each specific hair abnormality consideration should be given to whether the hair-shaft disorder was occurring as isolated or in association with other cutaneous, non-cutaneous abnormalities or as a syndrome.

Hair-shaft disorders can vary from barely noticeable even subclinical anomalies to severe effects hair-shaft. They can be present throughout the scalp or as patchy areas.

Patients with these disorders usually presented with short hair that broke easily.

Pili torti

In pili torti or corkscrew hair, there was irregular thickening of the outer root sheath and the flattened hairs rotate completely through 180 degrees at irregular intervals. The twists can at times resemble beads on light microscopy.[2] Occasional twists of less than 180 degrees did not qualify as true pili torti. These incomplete twists may occasionally occur in normal hair (seen in African hair and in the pubic/axillary hairs of other races).

Affected hairs were brittle, fracture easily and did not grow to any considerable length. Patients presented with a sparse and short coarse stubble over the entire scalp and may have a few circumscribed bald patches. Longer hairs may be seen in areas subject to less trauma. Minimizing trauma was the key aim. Occasionally the hairs were unruly, resembling uncombable hair syndrome. A late-onset variant of isolated pili torti that first presented after puberty with patchy alopecia had also been described.

Menke's kinky-hair syndrome was an X-linked recessive syndrome.[13] The defective gene, MKN or ATP7A, encoded for a copper-translocating membrane protein ATPase that prevented copper transport and led to the accumulation of intracellular copper in some tissues.[3] The affected child typically had pili torti, pale lax skin and intellectual or neurological impairment secondary to degeneration of cerebral, cerebellar and connective tissue. It was unknown why the abnormality in copper metabolism made the hair twist and defects in copper metabolism have not been demonstrated in other forms of pili torti. As copper was a cofactor for tyrosinase, affected hairs were lighter in color. Pili torti can also be associated with Bjornstad syndrome, Conradi-Hunerman syndrome.[4]

Trichorrhexis nodosa

Trichorrhexis nodosa was a localized thickening of the hair shaft due to fractures and fraying of the cortex to resemble a node.[12] A small paintbrush like appearance of the shaft was noted if there was a transverse fracture of the shaft.[5]

This was known as trichoclasis and the clinical correlate was a split end. African hair often withstood trauma poorly and nodes were common in long hair. Vigorous attempts to straighten curly hair may cause formation of nodes. When severe, this was described as acquired trichorrhexis nodosa. Trichorrhexis nodosa was the most common defect of the hair shaft leading to hair breakage.[5]

Treatment of trichorrhexis nodosa either congenital or acquired involved the avoidance of mechanical or chemical injury to hair. Strict avoidance of usage of staightners, curling irons, hair dryers were to be maintained.

Monilethrix

Monilethrix was a rare autosomal dominant hair-shaft disorder characterized by beading of hairs and hair fragility and might be associated with multiple papules over scalp also called as keratoses pilaris.[6] These varied from barely detectable effects to severe fragility. In severe cases the entire scalp was affected and patients were totally bald or more often have a sparse covering of short, twisted, broken and lusterless hairs. At time it was confused with confusion with congenital hypotrichosis. Perifollicular erythema was occasionally seen. Hairs of the eyebrows, eyelashes, face, pubis and legs may be involved but was rare. Fragile and splitting of the nails noted. Follicular keratosis and abnormal hairs were found most frequently on the nape and occiput but may affect the entire scalp. Monilethrix can be caused by mutations on chromosome 12q136. Diagnosis was by hair microscopy. Intermittently placed nodes form and the internodes tend to be the site of transverse hair fracture. Nodes range between 0.7-1.0 mm distance but there may be variation in internodal distance and nodal thickness.[6] They also might be associated with physical and mental retardation, koilonychias, cataract, syndactyly. This should be differentiated from pseudomonilethrix which resembled monelithrix but was due to optical illusion on viewing the slide.

Peripilar casts

Hair casts also known as peripilar keratin casts or pseudonits which were keratinous material that were present over the shaft seen in children and adults.[7] Disorders of keratinisation and inflammation of the follicular ostium produced the casts. Peripilar casts were more commonly seen in disorders like psoriasis and seborrhoeic dermatitis, lichen planopilaris or traction alopecia.[7] The casts can also be of a non-keratin type; derived from chemicals (e.g. haircare products) or micro-organisms bacteria or fungi attaching to the hair e.g. white piedra.

White piedra is an infection caused by superficial fungi which presented as asymptomatic whitish nodules over the shaft of hair.[11] They can be present on scalp or on any other hairy areas of the body. It was caused by yeast-like fungus, *Trichosporon beigelii*, now known as *T. asahii*. The disease can be controlled by usage of topical and systemic antifungals.[8]

Pilca polonica

The term plica neuropathica or pilca polonica had been used to describe an extremely rare condition in which the hair became compacted into numerous, irregularly twisted, irreversibly entangled plaits.[9] Focal matting of the hair was probably a common occurrence and was easily remedied by cutting or untangling the hair. The exact cause of this condition was unknown. It was thought to be caused due to usage of harsh shampoos, oils, usage of herbal medication, friction. It was also seen in certain religious groups who applied certain lotions or oil as and don't wash their hair or comb their hair for a long periods. Treatment involved cutting of the matted hair and prevention of usage of any harsh chemicals over hair.[9]

Trichoptilosis

This was longitudinal splitting of hair shaft starting from the distal end. This was usually due to repeated chemical or physical trauma caused to the hair. Treatments including avoidance of further usage of chemical and physical trauma and cutting of existing spilts.

Trichothiodystrophy (TTD)

TTD was a group of autosomal recessive disorders that occured due to deficiency of sulphur content.[10] The characteristic finding was alternating light and dark bands also called as the tiger tail pattern.[10] Other associated neuro and ectodermal abnormalities included short stature, ichthyosis, mental retardation, nail dystrophy, cataracts, photosensitivity and neurological deficits, impairment, decreased fertility and short stature. Some patients only exhibited hair changes. Sparse, brittle hair of different lengths may be seen. The nails were brittle and dystrophic. Follicular keratosis with or without erythroderma can also be present. Hair microscopy reveals brittle, weathered hair. Flattened and twisted ribbon-like hairs can also be seen. Light microscopy demonstrated the characteristic tiger-tail pattern of alternating dark and light diagonal bands.[10] The cause of this pattern was unknown. The diagnosis of TTD was by measuring the cysteine content, which paralleled total sulphur content. Genetic testing for GTH2H5 gene can also be done to rule out trichothiodystrophy. Tay's syndrome was a rare disorder which included ichthyosis, brittle hair, presence of absence of nail involvement, mental retardation, short stature, decreased gonadal function, progeria like features, microcephaly, presence or absence of lenticular opacities, presence or absence of ataxia, presence or absence of calcifications in basal ganglia, erythroderma and scales.

Advices

These were the following advices: avoid wetting hair when showering or taking a bath; do not wash the hair more than once a week; when shampooing, always use a conditioner and leave it on the scalp for at least five minutes before rinsing; use a conditioner without shampoo if the hair was clean; after wetting the hair, only pat it lightly dry and do not rub it with a towel; do not blow-dry the hair or use hot combs; comb the hair with a wide-toothed comb no more than once a day; do not brush the hair; avoid all hairdressing procedures in particular did not have hair bleached, dyed, straightened, permanent waved; avoid tight hats and bathing caps; do not tie the hair back tightly in a ponytail or in braids;

consider a satin pillowcase to reduce friction while sleeping.

Limitations

The number of cases were limited and genetic testing was done for one case.

CONCLUSION

An understanding of the normal variations of the hair shaft is essential to be able to tell whether one is dealing with a hair-shaft disorder. The normal hair shaft has a consistent diameter throughout its length, with the most common shape in cross section being oval. Significant variations exist particularly in different racial groups from straight to woolly hair as well as in thickness of the hair shaft. The diagnosis of structural abnormalities of the hair shaft is of clinical relevance, since such abnormalities may indicate underlying metabolic disorders, or may be associated with other diseases. Many of the hair shaft abnormalities present themselves as alopecia, in such cases in spite of giving treatment there might not be much improvement. In patients whose hair texture is not consistent with their racial background a diagnosis of hair shaft disorders is to be considered.

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