Case reports

Hair microscopy – a bed side test effectively used for diagnosing genodermatosis

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Introduction

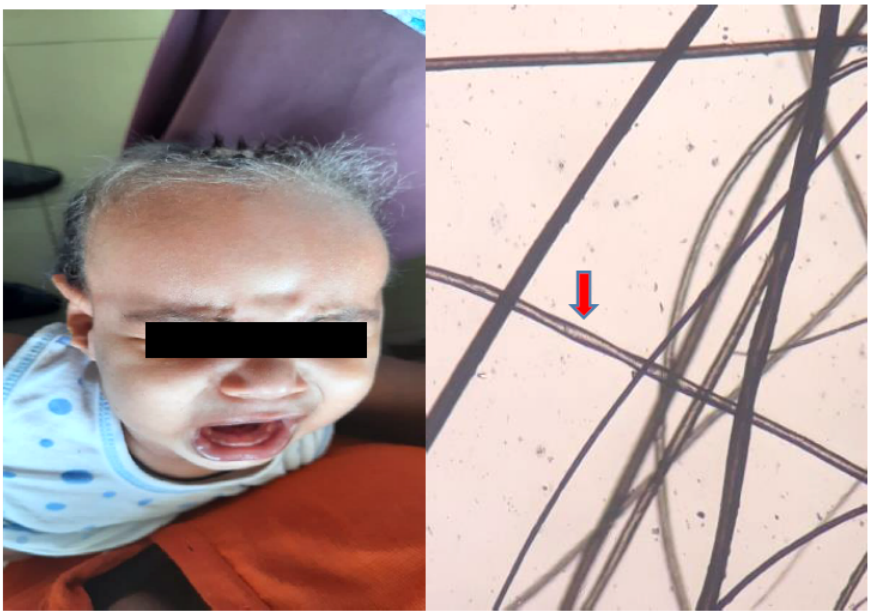
Genodermatoses are a large group of inherited disorders with skin manifestations. Early recognition of these diseases are important for detection of other associated abnormalities, commencement of therapeutic intervention, prevention of complications and genetic counselling. Diagnosis of genodermatosis is a challenge due to disease diversity, overlapping or heterogeneous phenotypes and huge amount of new information. Diagnosis of these rare diseases enable genetic analysis which are expensive and unavailable.

Hair microscopy form an important bedside clinical tool for the diagnosis of various disorders affecting the hair and the adjacent scalp. It is inexpensive and widely available. Herein we describe the clinical experience of using hair microscopy which helps in diagnosis of rare genodermatosis.

Case 1

One year old baby boy presented with early onset seizures, hypotonia and global developmental delay. He was referred to dermatology department due to pigmentary dilution of skin and hair.

He had fair skin compared to his parents. He had spangled hair with hypotrichosis. Microscopic examination of hair showed abnormal torsion of the hair strands along their own axis (Pilli torti) when compared to normal hair strands.



Diagnosis of Menkes disease was made clinically, later confirmed by reduced serum levels of both copper and ceruloplasmin.

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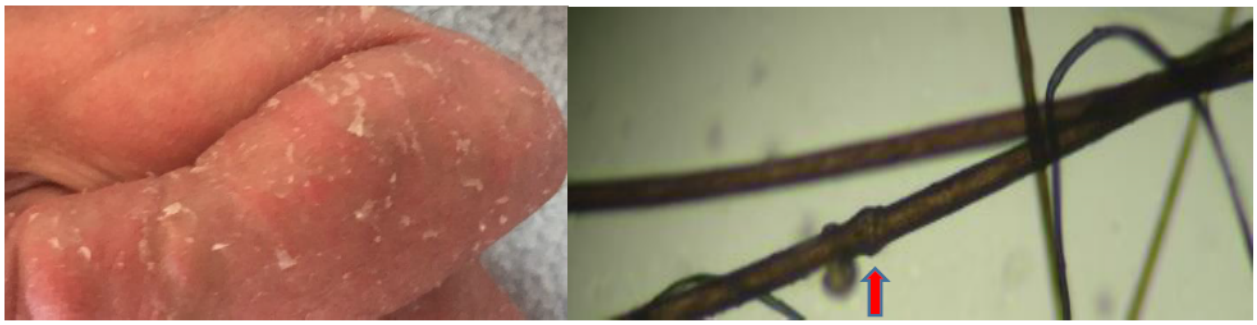
Menkes disease is an X-linked recessive disorder of generalized copper deficiency. It is characterized by psychomotor deterioration, refractory epilepsy, thin and brittle hypopigmented hair, skin pallor and hypothermia. Hair shaft abnormality is one of the striking features of this disease. Clinically the scalp hairs appear sparse, coarse, short, and lightly pigmented. They are fragile and break easily, resulting in apparent alopecia. Hair shaft microscopy most commonly reveals Pilli torti (180º twisted on axis). Other hair shaft abnormalities that occasionally seen are trichorrhexis nodosa and monilethrix.

Case 2

Four weeks old neonate was referred with generalized scaling and redness of the skin. He was product of consanguineous marriage and elder sibling had similar condition died during neonatal period. Dermatological examination revealed erythroderma and few hair in the scalp. Hair microscopy showed trichorrexis invaginata. Characteristic double edged serpeginous scale (ichthyosis linearis circumflexa) was appeared after few weeks.



Netherton syndrome diagnosis made clinically.

Netherton syndrome is a rare genodermatosis catheterized by neonatal erythroderma, trichorrhexis invaginata and an atopic dermatitis.

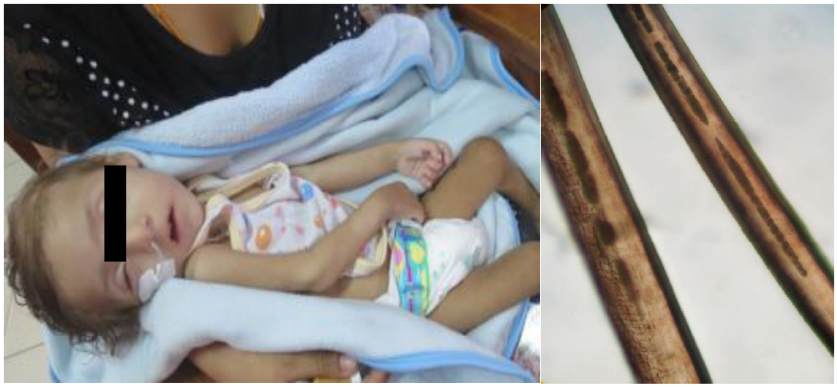
It is caused by mutations in the serine protease inhibitor of Kazal type 5 *(SPINK5)* gene which encodes lymphoepithelial Kazal-type inhibitor (LEKTI), expressed in mucosal and epithelial surface. These mutations result in an unopposed activity of epidermal proteases leading to premature desquamation of the stratum corneum and impairment of the skin barrier.

Trichorrhexis invaginata of hair and eyebrows due to invagination of the distal portion of the hair shaft into the proximal portion is pathognomonic for Netherton syndrome. When hair breaks at the point of invagination, the appearance simulates a “golf tee”. Some patients may have trichorrhexis nodosa and pili torti.

Case 3

Five year old child presented with seizure disorders and developmental delay. There was no history of recurrent infections.

Dermatological examination revealed fair skin with silvery hair. Hair microscopy showed irregular large clumps of melanin in the hair shaft which help to diagnose Griscelli syndrome clinically.



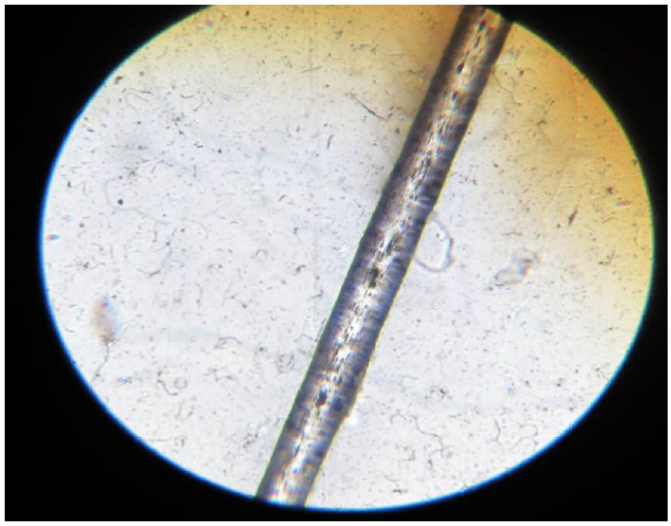
Griscelli syndrome (GS) is a rare autosomal recessive disorder that results in pigmentary dilution of the skin and the hair (silver hair). Defective transfer of melanin into keratinocytes is the underlying defect. Depending on the affected protein, three clinical subtypes has been described.

Type 1 – associated with neurological abnormalities (Mutation occur in the *MYO5A* gene)

Type 2 – severe immunodeficiency and lymphohistiocytotic hemophagocytosis (mutation in the *RAB27A* gene)

Type 3 – only limited to the skin (mutation in the melanophilin gene)

Chédiak-Higashi syndrome (CHS) is an autosomal recessive immunodeficiency disorder that shows pigmentary dilution with silvery hair similar to Griscelli syndrome. Hair microscopy aids to diagnose as well as differentiate these two conditions. In chediak – Higashi syndrome, melanin clumps are small and regularly spaced within the hair shaft.



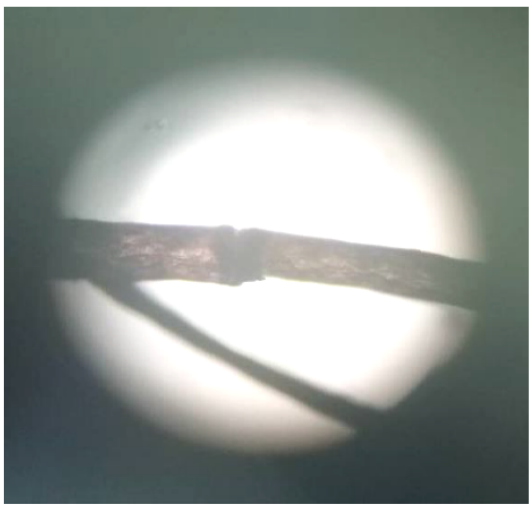
Giant cytoplasmic granules can be seen in the leukocytes.

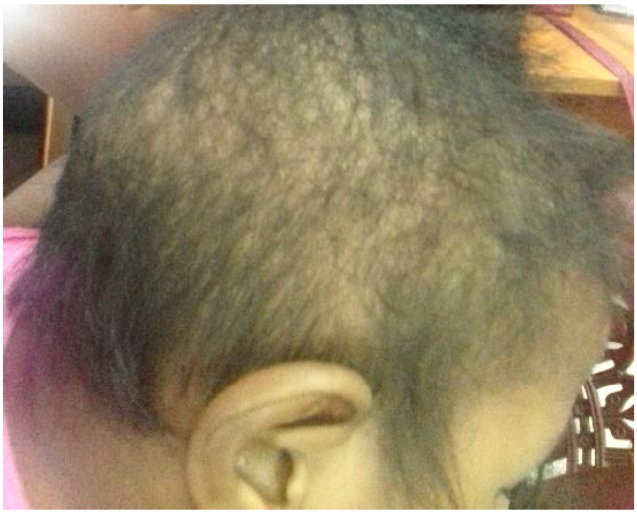
Case 4

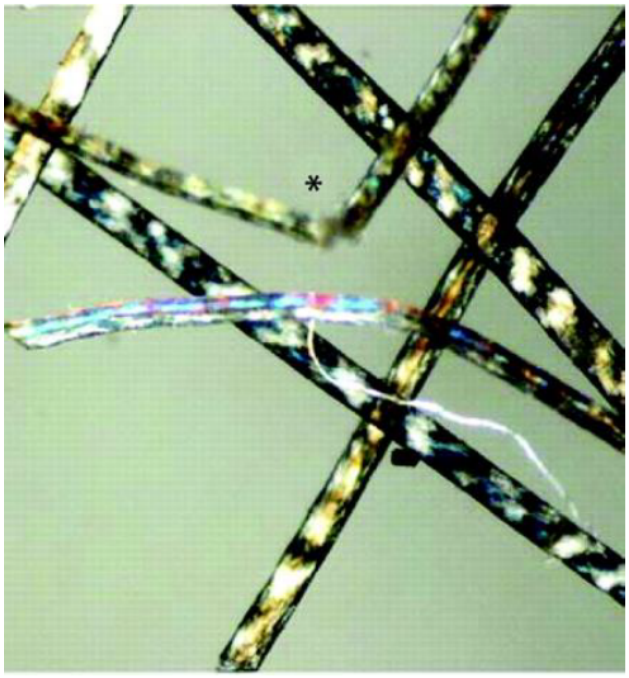
Two year old girl presented to our clinic with abnormal hair since birth along with dry skin. According to mother, baby was wrapped with a membrane (collodian baby) at birth and later appeared dry icthyotic skin. She had global developmental delay. No photosensitivity.

Dermatological examination revealed spangled hair with hypotrichosis and icthyotic skin.

Hair examination under light microscope showed trichoschisis. (Broken or split hairs) Alternating light and dark bands (tiger-tail banding) in the hair shaft, found at polarized light microscopy.





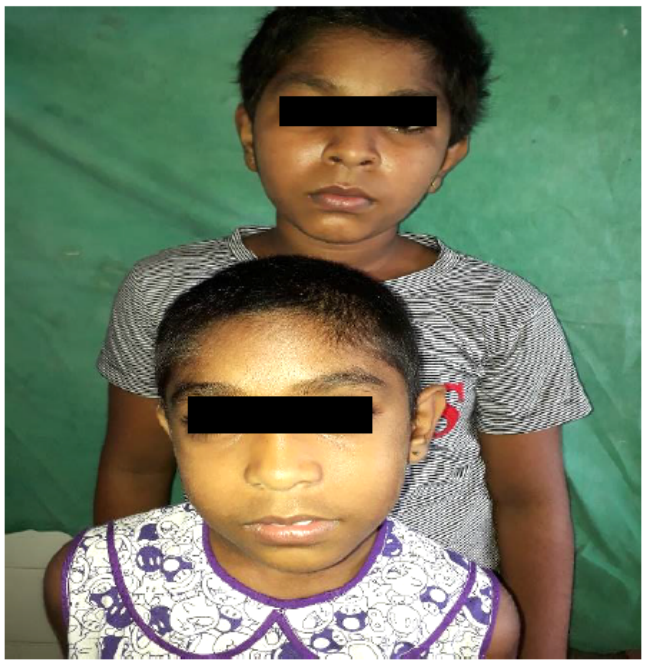


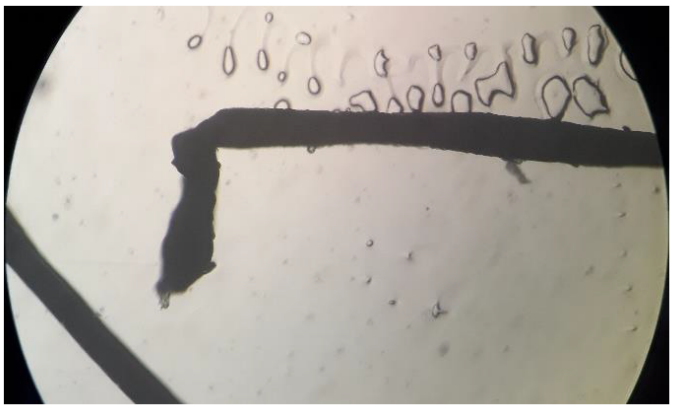
Trichothiodystrophy clinical diagnosis was made clinically.

Trichothiodystrophy is caused by defective DNA repair and transcription and is inherited in an autosomal recessive pattern. Additional cutaneous features other than spangled hair are ichthyosis and brittle nails. Other clinical features are photosensitivity, intellectual impairment and short stature which can be highly variable in expression.

Case 5

Two sisters, age of 13 and 7 presented to our clinic with poor hair growth and abnormal hair since birth. On examination hair density was normal without any patches of alopecia. Hair was lusterless and can be easily pull out without pain. Microscopic examination of extracted hair showed appearance of ruffled cuticle distal to the hair bulb resembling crumpled sock. The hair bulb is oriented at an acute angle to the hair shaft.





Loose anagen syndrome diagnosis made clinically.

Loose anagen hair syndrome is a rare congenital hair disorder that is caused by premature keratinization of the inner root sheath which causes impaired adhesion between the cuticle, the sheath and the hair shaft. It is confirmed by positive hair pull test and characteristic findings on hair microscopy.

References

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