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HAIR CAN SPEAK – A CASE SERIES OF RARE AUTOSOMAL RECESSIVE GENODERMATOSES

Dermatology

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ABSTRACT

Background: Hair shaft anomalies serve as vital diagnostic indicators in various genodermatoses. Microscopic examination of hair can reveal characteristic patterns that aid in the identification of underlying genetic disorders. **Aim:** To evaluate the diagnostic utility of hair shaft microscopy in identifying rare autosomal recessive genodermatoses. **Methods:** This case series encompasses 11 patients presenting with hair abnormalities suggestive of genodermatoses. Each patient underwent detailed clinical evaluation followed by light microscopy of hair samples. The cases included: 2 cases of Griscelli syndrome, characterized by silvery-gray hair with large, clumped melanosomes on the hair shaft. 5 cases of Monilethrix, exhibiting beaded hair appearance due to periodic constrictions along the shaft. 2 cases of Trichothiodystrophy. A case of Trichorrhexis invaginata, displaying the "bamboo hair" pattern and a case of Trichorrhexis nodosa, identified by nodes along the hair shaft leading to breakage. **Results:** Hair microscopy provided definitive diagnostic clues in all cases. The distinctive microscopic features observed correlated well with clinical findings, facilitating accurate diagnosis without the immediate need for genetic testing. **Conclusion:** Hair shaft microscopy is a non-invasive, cost-effective diagnostic tool that can reveal hallmark features of rare autosomal recessive genodermatoses. Its application can expedite diagnosis and management, especially in resource-limited settings.

KEYWORDS

Hair microscopy, Genodermatoses, Griscelli syndrome, Monilethrix, Trichothiodystrophy, Trichorrhexis invaginata, Trichorrhexis nodosa.

INTRODUCTION

Hair shaft anomalies often serve as diagnostic indicators in various genodermatoses. Microscopic examination of hair can reveal characteristic patterns that aid in the identification of underlying genetic disorders. This non-invasive, cost-effective diagnostic tool is particularly valuable in resource-limited settings where advanced genetic testing may not be readily available.[1]

Several rare autosomal recessive genodermatoses exhibit distinctive hair shaft abnormalities observable under light microscopy. For instance, Griscelli syndrome presents with large, irregular melanin granules in the hair shaft, while Monilethrix is characterized by a beaded appearance due to periodic constrictions along the shaft. Trichothiodystrophy shows brittle hair with alternating light and dark bands (tiger-tail banding) under polarized light, and Trichorrhexis invaginata displays the "bamboo hair" pattern. Trichorrhexis nodosa is identified by nodes along the hair shaft leading to breakage.[2]

Despite the diagnostic significance of hair microscopy, it remains underutilized in clinical practice. A lack of awareness and limited training in hair microscopy techniques contribute to missed or delayed diagnoses of these genodermatoses.

This case series aims to evaluate the diagnostic utility of hair shaft microscopy in identifying rare autosomal recessive genodermatoses. By analyzing characteristic microscopic hair findings in a series of clinically suspected cases, we seek to highlight the importance of hair microscopy as a valuable diagnostic tool in dermatology.

MATERIALS AND METHODS

This case series was conducted in the Department of Dermatology at Kurnool medical college (KMC), Kurnool, Andra Pradesh; following Institutional Ethics Committee approval. A total of 11 patients presenting with hair abnormalities and clinical suspicion of rare autosomal recessive genodermatoses were included over a period of a period of 12 months (March 2024 - February 2025).

Inclusion Criteria:

Patients with hair shaft abnormalities suggestive of genodermatoses and clinical features consistent with pedigree analysis showing inherited autosomal recessive dermatologic disorders.

Exclusion Criteria:

Patients with acquired hair shaft disorders.

Clinical Evaluation:

A thorough history was taken, including consanguinity, family history of similar disorders, associated skin/systemic findings, and age of onset. Each patient underwent a detailed dermatological examination with emphasis on scalp, eyebrows, eyelashes, and body hair.

Hair Microscopy:

Hair samples were collected by gentle plucking from the scalp and other involved areas. Samples were mounted on glass slides with normal saline and examined under light microscopy at 10x and 40x magnifications.

Diagnostic Features:

- **Griscelli Syndrome:** Presence of large, irregular melanin granules clumped along the medulla of hair shafts.
- **Monilethrix:** Beaded appearance with elliptical nodes and constrictions.
- **Trichothiodystrophy:** Brittle hair with typical clinical features and alternating dark and light bands (tiger-tail banding) under polarized light.
- **Trichorrhexis Invaginata:** Telescoping of distal hair shaft into proximal portion ("ball-and-socket" or bamboo hair).
- **Trichorrhexis Nodosa:** Frayed, brush-like fractures of the shaft with nodular swellings.

Data Collection And Analysis:

Demographic details, clinical features, and microscopic findings were documented in a structured format. Correlation between clinical suspicion and hair microscopy was analyzed descriptively. Images were captured from a light microscope.

Case Report 1: -

A 1-year-old girl, born to second-degree consanguineous parents, presented with silver-gray hair since birth (Figure 1) and hypopigmented macules over the face and abdomen for the past 6 months. She had a history of recurrent fevers, abdominal distension, and delayed developmental milestones. There was no history of seizures, bleeding tendencies, or similar complaints in family members. Clinical examination revealed pallor, silver-gray hair involving the scalp, eyebrows, and eyelashes, multiple hypopigmented macules on the face and abdomen, and marked hepatosplenomegaly

Differential diagnoses considered included Griscelli Syndrome (GS), Chediak-Higashi Syndrome (CHS), Oculocutaneous Albinism (OCA), Phenylketonuria (PKU), and Menkes Disease. Complete blood count revealed pancytopenia, and peripheral smear confirmed pancytopenia with normal neutrophil morphology. Abdominal ultrasonography showed hepatosplenomegaly. Bone marrow aspiration and biopsy demonstrated hemophagocytosis. Tandem mass spectrometry results were within normal limits. Hair shaft microscopy revealed large, irregular melanin clumps along the medulla (Figure 1), which is diagnostic of Griscelli Syndrome.

Based on the combination of pigmentary dilution, immune dysfunction manifested as pancytopenia and hemophagocytic lymphohistiocytosis, along with characteristic findings on hair shaft microscopy, a diagnosis of Griscelli Syndrome Type 2 was established.

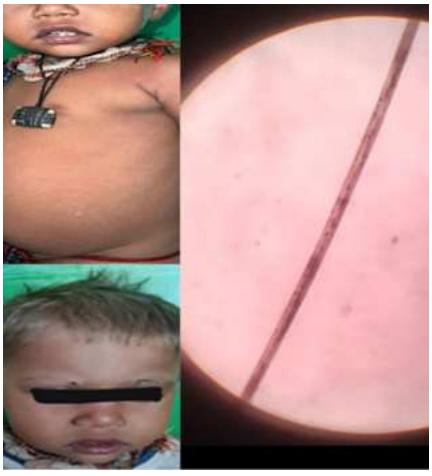

Figure 1. Silvery grey scalp hair, hypopigmented macules over protruded abdomen and hair shaft showing large irregular melanin granules. (10 X)

Case Report 2: -

A 2-year-old male child, born of a third-degree consanguineous marriage, was brought to the dermatology OPD with complaints of silver-gray hair on the scalp and eyebrows since birth, along with hypopigmented macules over the face and abdomen for the past three months. The child also exhibited significant developmental delays, including inability to roll over, poor neck control, and failure to grasp objects. Multiple history of seizure episodes was present. No similar findings were reported in other family members.

On examination, the child appeared anemic and had silver-gray hair over the scalp, eyebrows, and eyelashes. Multiple hypopigmented macules were noted on the face and abdomen (Figure 2). Clinical features included microcephaly, generalized hypotonia, and global developmental delay; the child was unable to sit or roll over. Deep tendon reflexes, however, were preserved.

Differential diagnoses considered included Griscelli Syndrome (GS), Chediak-Higashi Syndrome, Oculocutaneous Albinism (OCA), and Phenylketonuria (PKU). Complete blood count revealed anemia, and peripheral smear showed hypochromic microcytic anemia with normal neutrophils lacking abnormal granules. Abdominal ultrasonography, tandem mass spectrometry, EEG, and MRI brain was normal. Hair shaft microscopy demonstrated large, irregular melanin clumps (Figure 2) a hallmark diagnostic feature of Griscelli Syndrome. Based on the combination of pigmentary dilution and sever neurological involvement with characteristic findings on hair shaft microscopy, a diagnosis of Griscelli Syndrome Type 1 was established.

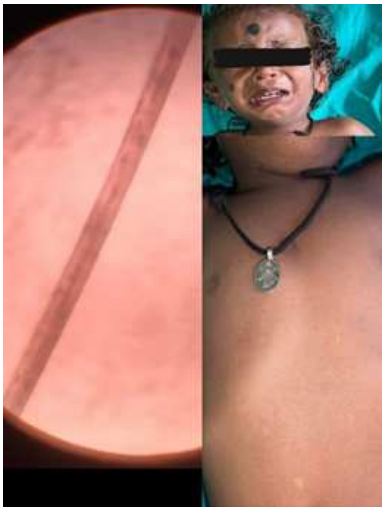

Figure 2. hair shaft showing large irregular melanin granules. (40 X), Silvery grey scalp hair with hypopigmented macules over abdomen

Case Report 3,4 & 5

A 6-year-old girl, born of a second-degree consanguineous marriage with complaints of scalp hair thinning and abnormal hair texture since infancy. The parents reported no history of psychomotor delay. A positive family history was noted, with two other cousin siblings presenting similar complaints. Their parents also had history of second-degree consanguineous marriage. Clinical examination revealed short, sparse, and brittle scalp hair. No abnormalities were noted in the eyebrows, eyelashes, or body hair. Systemic examination was unremarkable. Pedigree analysis demonstrated an autosomal recessive (AR) inheritance pattern, with all affected children born to consanguineous parents.

Hair shaft microscopy showed the classical “pearl necklace” or “beaded” appearance (Figure 3), characterized by moniliform constrictions at regular intervals, confirming the diagnosis of monilethrix. These constrictions represent points of weakness due to abnormal keratinization, rendering the hair susceptible to breakage. Differential diagnoses included pseudo-monilethrix, monilethrix-like hair, Menkes disease, and pili torti. As there is currently no definitive cure for monilethrix, management remains supportive. Treatment focused on minimizing hair trauma through gentle hair care, along with the use of topical 2% minoxidil, keratin and biotin supplementation to potentially enhance hair strength

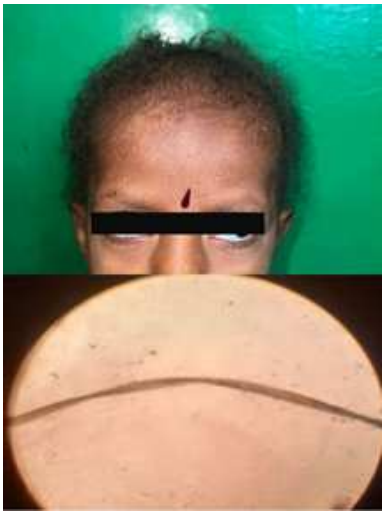

Figure 3: 6 years old girl with short, sparse, and brittle scalp hair. And hair shaft showing “beaded” appearance. (10 X)

Case Report 6 & 7

Two male siblings, aged 8 and 5 years, born to second degree consanguineously married parents, were brought to the dermatology outpatient department by their mother with complaints of abnormal scalp hair since early childhood. There was no history of psychomotor

delay or other systemic abnormalities. On examination, both children had short, dry, lusterless, sparse, and brittle scalp hair. No abnormalities were noted in the skin, nails, or teeth. Systemic examination was within normal limits. Pedigree evaluation revealed two affected siblings born to second-degree consanguineous parents, suggesting an autosomal recessive (AR) inheritance pattern.

Differential Diagnosis Considered Pseudo-monilethrix, Monilethrix-like hair, Pili torti, Menkes kinky hair disease and Trichorrhexis invaginate. Light microscopy of the hair shafts revealed a characteristic "pearl necklace" or beaded appearance (Figure 4), with regularly spaced elliptical nodes and constrictions diagnostic of Monilethrix. The patients were advised topical 2% minoxidil, oral retinoids for hair strengthening (used cautiously, considering potential side effects).

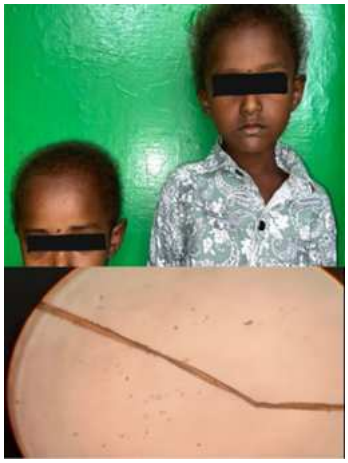

Figure 4: Kids with short, sparse, and brittle scalp hair. And hair shaft showing "beaded" appearance. (10 X)

Case Report 8 & 9

An 8-year-old male child, born of a second degree consanguineous marriage, presented with rough, brittle scalp hair and intellectual delay. A younger 3-year-old female sibling in the family exhibited similar symptoms.

Clinical Findings In The Index Case: (IBIDS Syndrome)

- **Hair:** Dry, short, brittle hair with easy breakage
- **Skin:** Generalized ichthyosis
- **Growth & Development:** Short stature, intellectual impairment, delayed developmental milestones

Hair shaft light microscopy shows "through-and-through fractures" (trichoschisis) and disruption of overlying cuticle (Figure 5), characteristic of trichothiodystrophy. The typical clinical features and light microscopy pointed the diagnosis toward the TDD – IBIDS. Further advised plarizing microscopy to demonstrate "tiger-tail banding", Hair sulfur content analysis (low sulfur confirms diagnosis).

Figure 5: Kids with rough and brittle scalp hair; hair microscopy showing trichoschisis and few alternative dark and light bands.

Case Report 10

An 8-year-old female child, born of a second degree consanguineous marriage, was brought with complaints of rough, brittle scalp hair and generalized dry skin since infancy. Although there was no family history of similar hair abnormality, a positive family history of atopic dermatitis was noted.

Clinical examination revealed ichthyosis linearis circumflexa with generalized dry, scaly skin with Brittle, fragile scalp hair with areas of hair loss. Serum IgE was elevated and Hair shaft microscopy showed classic trichorrhexis invaginata ball and socket appearance / bamboo hair appearance (Figure 6), consistent with bamboo hair. The case was diagnosed as Netherton Syndrome, managed with supportive care and including the use of emollients.

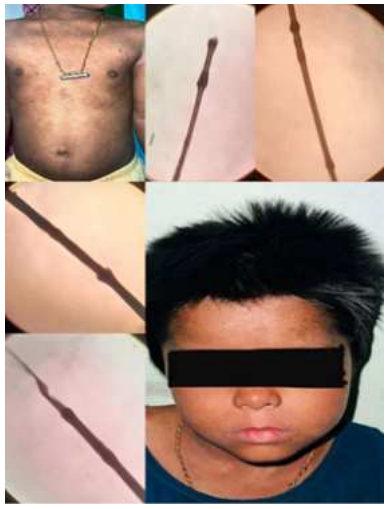

Figure 6: 8 years old girl with ichthyosis linearis circumflexa, classic trichorrhexis invaginata (ball and socket appearance) in hair microscopy and powdery appearance over hair.

Case Report 11

13 years old boy born to second degree consanguineously married parents; came with complaints of easily breakable hair, examination showed dry and rough strands. Although there was no similar hair abnormality in family.

Microscopy showed brush-like fraying and nodes along the hair shaft suggestive of thrichorrhexis nodosa (Figure 7). The case was managed with supportive care, including gentle hair care and biotin supplementation.

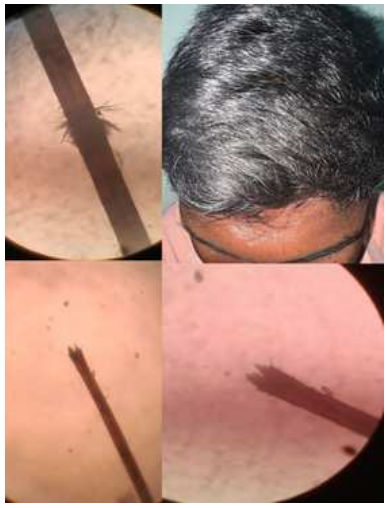

Figure 7: 13 years old boy with powdery apperance of scalp hair with hair shaft microscopy showing thrichorrhexis nodosa (brush-like fraying and nodes along the hair shaft).

DISCUSSION:

Hair microscopy findings in all patients were consistent with the clinical diagnosis and significantly aided in confirming the suspected genodermatoses. No adverse effects were reported during or after

sample collection.

Autosomal recessive genodermatoses associated with hair shaft abnormalities include Griscelli syndrome, Chediak-Higashi syndrome, Monilethrix, Trichothiodystrophy (TTD), Netherton syndrome, congenital Trichorrhexis nodosa and Pili torti.

Hair shaft microscopy is a time-tested but often underutilized tool that provides crucial diagnostic clues in genodermatoses. In our series, microscopy confirmed clinical suspicion in all 11 patients, underlining its diagnostic value.

Table 1

Disease	Gene & inheritance	Clinical features	Microscopic features	Differential diagnosis	Management
Griscelli Syndrome (GS).[3,4,5,6,7.] AR pattern of inheritance	Type 1 GS - MYO5A on chromosome 15q21 Type 2 GS- RAB27A on chromosome 15q21.3 Type 3 GS - MLPH on chromosome 2q37.3	Silvery-gray hair and hypopigmented skin. Type 1 GS with severe neurological impairments. Type 2 GS with hematological abnormalities. Type 3 GS with skin manifestation.	Large, irregular melanin granules in hair shafts.	Chediak-Higashi syndrome, Elejalde syndrome.	Type 1 GS – neurological care. Type 2 GS- Immunosuppressive therapy, bone marrow transplant.
Monilethrix.[8,9.] Both AR & AD of pattern of inheritance	AD type: - KRT81, KRT83, KRT86 on chromosome 12q13 AR type: - DSG4 on chromosome 18q12	short, sparse, and brittle hair	Beaded hair shafts with periodic constrictions	Pili torti, Trichorrhexis nodosa	Minoxidil, hair care, keratin supplements
Trichothiodystrophy-TTD.[10,11,12.] AR pattern of inheritance	ERCC2, MPLKIP, GTF2H5 (AR)	PIBIDS – Photosensitivity, Ichthyosis, Brittle hair, Intellectual impairment, Decreased fertility & Short stature.	Tiger-tail banding under polarized light	Menkes disease, Xeroderma Pigmentosum	Supportive, sun protection, nutrition
Trichorrhexis Invaginata (Netherton syndrome).[13,14,15.] AR pattern of inheritance	SPINK5 (AR)	Ichthyosis linearis circumflexa Trichorrhexis invaginate :- rough & brittle scalp hair. Atopic diathesis	Ball-and-socket deformity of the hair shaft (Bamboo Hair)	Trichorrhexis Nodosa	Supportive, emollients, avoid trauma
Trichorrhexis Nodosa.[16.17.]	Congenital:- associated with Menkes disease, Argininosuccinic aciduria or Netherton syndrome. Acquired:- physical/chemical trauma or nutritional deficiencies (e.g., iron, biotin, zinc).	Dry and rough strands with easy breakable hair.	Frayed/broken nodes along shaft (bamboo-like appearance)	Netherton syndrome, Hair shaft trauma	Gentle hair care, biotin, emollients

Griscelli Syndrome (GS) is a rare autosomal recessive disorder marked by skin and hair hypopigmentation, with subtype dependent neurological and immunological involvement.[3] GS is classified into three distinct subtypes based on the underlying gene mutation.[456] It is an uncommon condition, with only around 160 cases documented globally.[7] Genetic testing confirms the diagnosis and distinguishes among subtypes.

Monilethrix is a rare hereditary hair shaft disorder marked by a beaded appearance, caused by periodic constrictions (internodes) alternating with normal segments (nodes). Light microscopy reveals the characteristic beaded pattern elliptical nodes alternating with narrow, fragile internodes.[8,9.]

Trichothiodystrophy (TTD) is a rare AR neuroectodermal disorder, the diagnosis is often guided by characteristic findings on polarized light microscopy, where the hair shaft displays a "tiger-tail" banding pattern, reflecting alternating light and dark transverse bands due to the uneven distribution of sulfur-rich proteins.[10,11] A systematic review encompassing 112 published cases of trichothiodystrophy was reported up to the year 2008.[12]

Netherton syndrome is marked by ichthyosis linearis circumflexa, allergic tendencies and a characteristic hair shaft defect called trichorrhexis invaginate also known as bamboo hair, which is considered diagnostic of the condition.[13,14,15.]

Trichorrhexis Nodosa (TN) is a common hair shaft defect marked by weak points that fracture, appearing as white or translucent nodes under light microscopy. These nodes resemble "brooms pushed end-to-end" due to cortical fiber splaying.[16,17]

TN Can Be:

- **Congenital –** sometimes associated with syndromes like Menkes disease or Netherton syndrome.
- **Acquired –** more common, caused by physical/chemical trauma (e.g., styling, harsh products) or nutritional deficiencies (e.g., iron, biotin, zinc).

All the patients of our cases (100%) had a history of consanguinity, which reflects the autosomal recessive inheritance pattern and is in line with global data on genodermatoses in consanguineous populations.[1]

Overall, hair shaft microscopy proved to be a highly valuable, non-invasive, and low-cost diagnostic tool, especially in pediatric dermatology where genetic tests may be unaffordable or inaccessible. Studies have reiterated its importance, including Bhattarai D et al. (2021) who emphasized its utility in systemic pediatric disorders.[1]

Our findings support the integration of routine hair microscopy into dermatologic evaluations of suspected genodermatoses. Early recognition enables appropriate counseling, targeted investigations, and preventive strategies for affected families.

CONCLUSION:

Hair shaft microscopy is a simple, rapid, and non-invasive diagnostic tool with significant utility in identifying rare autosomal recessive genodermatoses. In this case series, characteristic microscopic patterns closely matched clinical features, enabling accurate diagnosis even without genetic testing. Its cost-effectiveness and accessibility make it particularly valuable in resource-limited settings. Routine integration of hair microscopy in dermatological practice especially for pediatric and inherited disorders can expedite diagnosis and improve patient care. Greater awareness and training are essential to fully harness its diagnostic potential.

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Conflicts Of Interest: - There is no conflict of interest.

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