Community Dermatology

# The value of diagnosis in the clinic or by telemedicine. Genotrichology siné genetic study: “Nine approaches to creative problem solving”

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In the context of thousands of skin conditions many of which are of genetic aetiology, in a world in which poverty and an abundance of children predominates,the skills of diagnosis that the dermatology profession demonstrates are an important earliest first phase of good management. I have chosen the simple procedure of examining a child’s hair to illustrate this significant benefit from visiting a skin clinic or, as described by Schmidt in this CD by recent advances in telemedicine. A plucked hair will; survive long distance travel by post. It is a low cost maneuver that can benefit when a child presents with serious undiagnosed systemic illness.

Genotrichology implies study of hair disorders with genetic etiology. These disorders are not very rare and molecular diagnostic methods are required for definitive diagnosis. However, in resource poor populations of developing countries, facilities for genetic studies are either non-existent or beyond the affordability of patients. In such situations the clinical acumen of the treating physician, aided by easily available side laboratory procedures is the mainstay of diagnosis of these disorders. It steers the physician to examine for underlying and multiple signs of other genetic defects not just in the skin.

While confronting the diagnosis of a child with genetic hair disorder, a systematic approach is required. A novel way to do this would be to follow the “nine approaches to creative problem solving”. In order to reach the solution in a problem-shooting case, the following strategies may be followed:

- Rethink
- Visualize
- Produce
- Combine
- Form
- Opposite
- Metaphor / simile
- Patience
- Failure

Following these strategies helps the clinician to think in a productive manner and thus reach a conclusion in a situation of diagnostic dilemma.

In the following sections, problem-oriented clinical scenarios in children with genetic hair disorders will be discussed.

**Case scenario 1:** A child of non-African ancestry presented with tightly curled hair with curl diameter of 0.5cm (Figure 1). The morphological appearance of hair made us "rethink" the case and investigate to find out a possible underlying disorder. Flow Chart 1 presents possible causes of wooly hair and other associated organ involvement. Fatal cardiac involvement is an important accompaniment of some disorders with wooly hair. Hence all cases with wooly hair should undergo cardiac screening. It is important to distinguish curly hair from wooly hair; the former is manifested by large, loose, spiral locks and in association with other organ involvement, may constitute various syndromes (Flow Chart 2). Rethinking broadens diagnostic possibilities.

**Case scenario 2:** A 5-year-old girl presented with an asymptomatic patch of alopecia on the scalp, present since birth. On examination, a triangular-shaped area of alopecia was present on the left temporal area of the scalp with the base of the triangle located towards the forehead (Figure 2). The history of its presence since birth, the location and shape of the lesion helped to make the diagnosis of congenital triangular alopecia.

**Case scenario 3:** Children may present with altered hair color, which is a pointer to the diagnosis of disorders like oculo-cutaneous albinism (tyrosinase positive / negative) (Figures 3a and 3b), Hermansky-Pudlak syndrome, Chediak-Higashi syndrome. White forelock is indicative of piebaldism or Waardenburg syndrome and may lead to early diagnosis of associated deafness.

In the above two situations imageries (triangular shape of alopecia / various hair colors) had been used as diagnostic tools. "Visualization" is an important diagnostic mode and helps in the application of diagrams and imageries to analyze a dilemma.

**Case scenario 4:** A 3-year-old child presented with absence of hair (scalp, eyebrows, eyelashes, body hair), present since birth. He was diagnosed as a case of alopecia universalis congenita (Figure 4).

**Case scenario 5:** A 5-year-old boy presented with total alopecia of scalp, sparse eyelashes and eyebrows. At birth he had normal-looking scalp hair, which was shed by 1 year of age. On close examination, there were numerous papular lesions on the scalp skin. A provisional diagnosis of 'atrichia with papular lesions' was made which was confirmed by histopathology (Figure 5).

Constant application of the thought process and utilization of appropriate tools help to produce a solution.

**Case scenario 6:** Consultation with a Dermatologist was asked for because of the abnormal appearance of the hair in a 1-year-old male baby admitted in to a neonatal intensive care unit for repeated episodes of convulsion followed by severe central nervous system deterioration. On examination, the child was deeply comatose, with pale complexion and short, sparse, ivory-colored hair. Light microscopy of plucked hair showed pili torti.

**Case scenario 7:** A 6-month-old child presented with sparse, broken hair on scalp, thin eyebrows, mild ichthyosis and intolerance to sunlight (Figure 6). Light microscopic examination of plucked scalp hair showed clean transverse fracture of the hair shaft (trichoschisis) and wavy distribution of melanin. Presenting features of photosensitivity, ichthyosis and brittle hair helped us to consider the clinical diagnosis of PIBIDS in this case, which was supported by hair microscopy.

The "Combination" of the background knowledge of the clinician with available clinical features in a given patient and related simple investigations are the greatest of assets in prompt diagnosis of multisystem disorders. Clinical features and light microscopic morphology of the patients' hairs helped us to reach the provisional diagnosis in above two cases. Pili torti is the common denominator for various syndromes in different combination with other systemic features (Flow Chart 3). Light microscopic finding of pili torti may be a pointer towards a serious systemic disorder like Menke's kinky hair syndrome.

**Case scenario 8:** A 1.5-year-old male child presented with scalp alopecia since birth and lost other body hairs subsequently. He had a widely open anterior fontanel till this age and radiological evidence of rickets which was refractory to treatment with vitamin D. Laboratory investigations revealed hypocalcemia and high serum alkaline phosphatase level. A provisional diagnosis of "vitamin D resistant rickets type II" was made.

"Forming" relationship and making connection among apparently non-related symptoms may help to reach a diagnosis.

**Case scenario 9:** A 7-year-old girl was admitted to a pediatric intensive care unit in a deep comatose state and profound hypotonia. Cerebrospinal fluid analysis was non-contributory and CT scan of brain revealed cerebral edema. Dermatological consultation was asked for unusual color of her hair. On examination, the child's scalp hair showed a silvery shine and photo-exposed body parts showed a bronze-tan. Light microscopic examination of plucked hairs showed small and large irregular clumps of melanin (Figure 7). The patient was labeled as a case of 'silvery hair syndrome' and worked up further. Her elder sister had similar skin and hair color and died at young age due to some undiagnosed neurological disorder. There was no history of recurrent infections suggestive of immunodeficiency. Neutrophil count was within normal range and peripheral blood smear did not show giant abnormal granules in leucocytes. Skin biopsy from photo-exposed area showed irregular-sized melanin granules dispersed in the basal layer. History and laboratory tests helped in establishing the diagnosis of Elejalde disease by differentiating from other disorders with silvery hair, i.e. Griscelli syndrome and Chediak Higashi syndrome.

Some disorders may share a basic similarity, though different pathogenetically. Clinicians should not always stick to an obvious diagnosis, but think "opposite" to pin-point the actual diagnosis out of common symptoms.

While diagnosing some hair disorders the clinician must think "metaphorically", bringing "simile" to other objects. Trichorrhexis invaginata or "bamboo hair" (Figure 8a and 8b) and trichorrhexis nodosa may be the feature of various syndromes and methodical systemic analysis may help in diagnosing the underlying disorder (Flow Charts 4 and 5).

While diagnosing a genetic hair disorder in a resource poor set up the clinician must use his clinical acumen and utilize easily available simple investigative procedures with "patience" to derive the

best possible diagnosis out of his hard work. However, “failure” may come on the way, but it stimulates the human brain to think and work further; practically, all successful diagnostic attempts are based on failure.

## References


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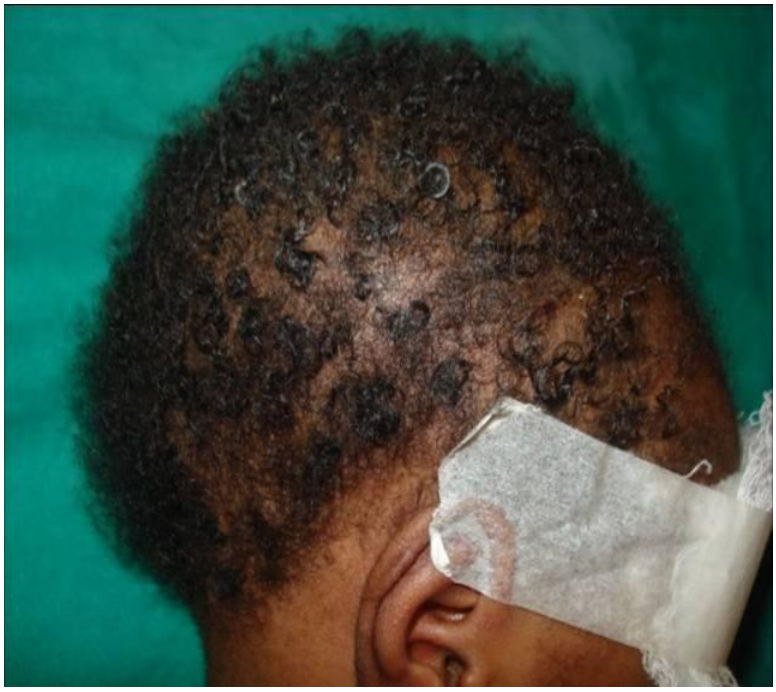


Figure 1: Wooly hair in a child

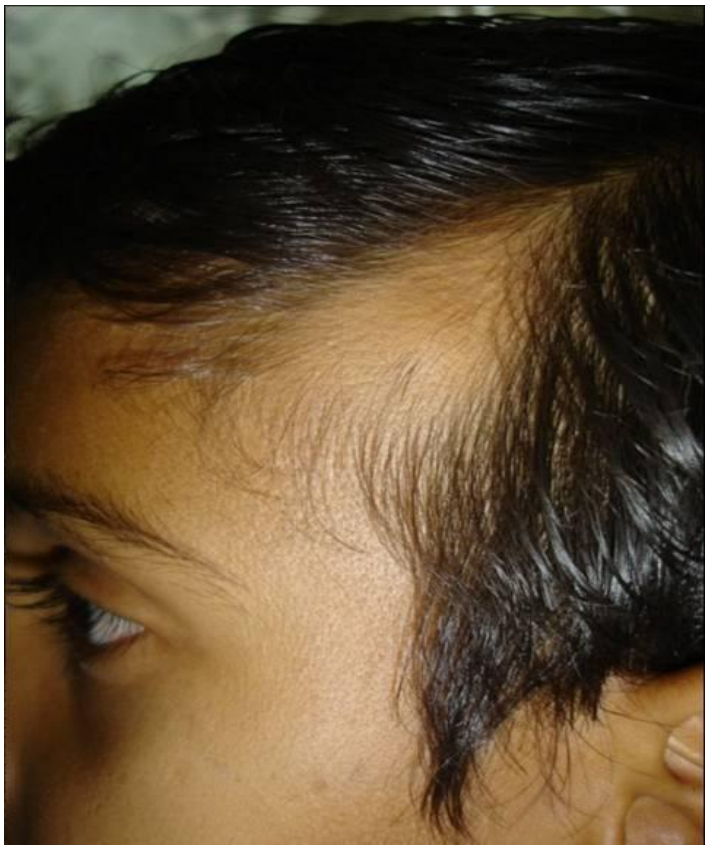


Figure 2: Congenital triangular alopecia

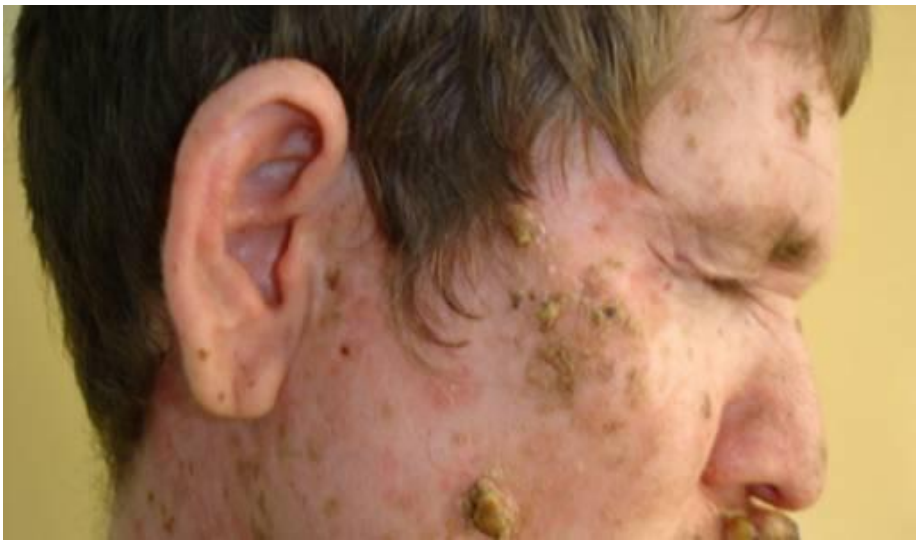


Figure 3a: Tyrosinase positive albinism with photophobia and squamous cell carcinonma

Figure 3b: Rufus albinism

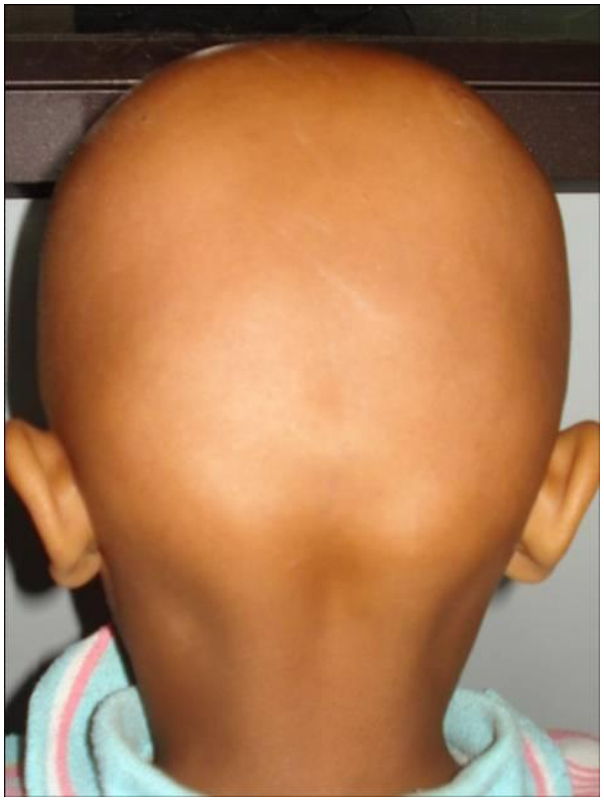


Figure 4: Alopecia universalis congenita.

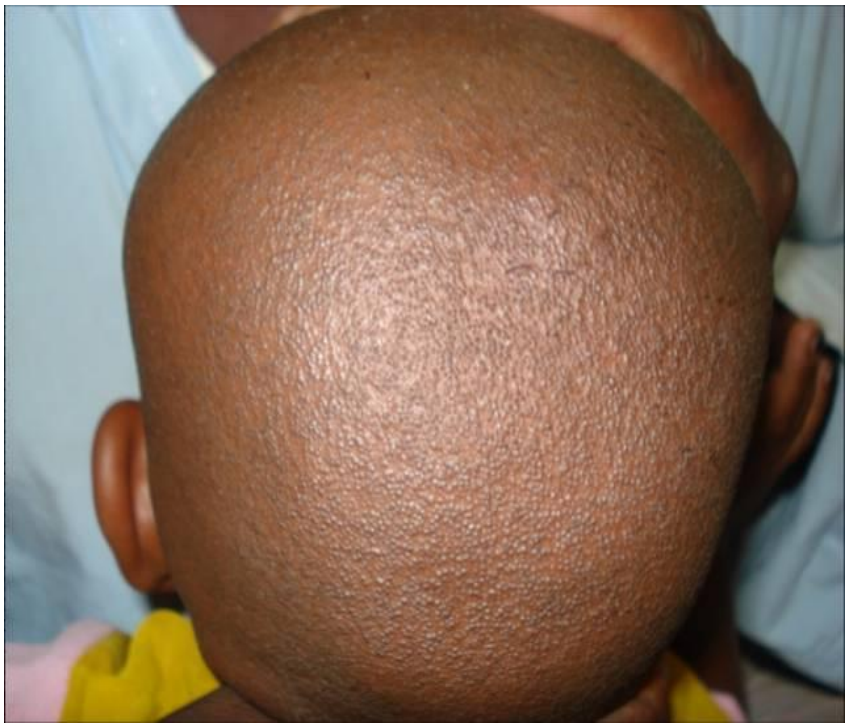


Figure 5: Atrichia with papular lesions

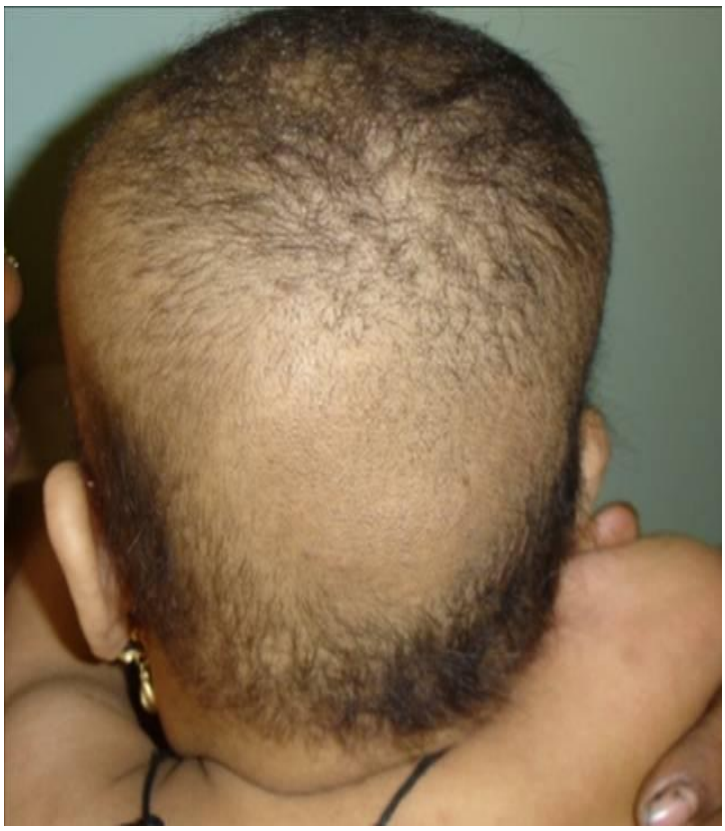


Figure 6: Sparse, broken hair of PIBIDS syndrome

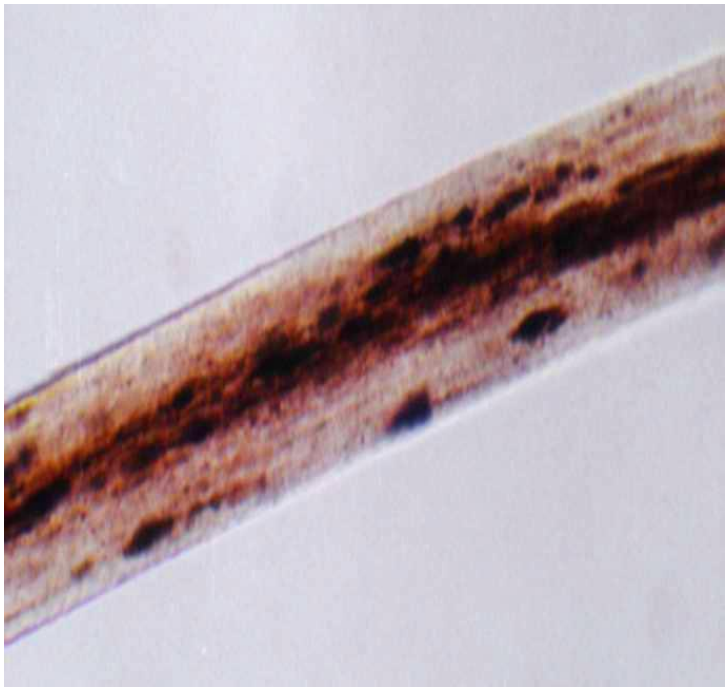


Figure 7: Photomicrograph of hair showing large and small clumps of melanin in irregular pattern; Elejalde disease (x10).

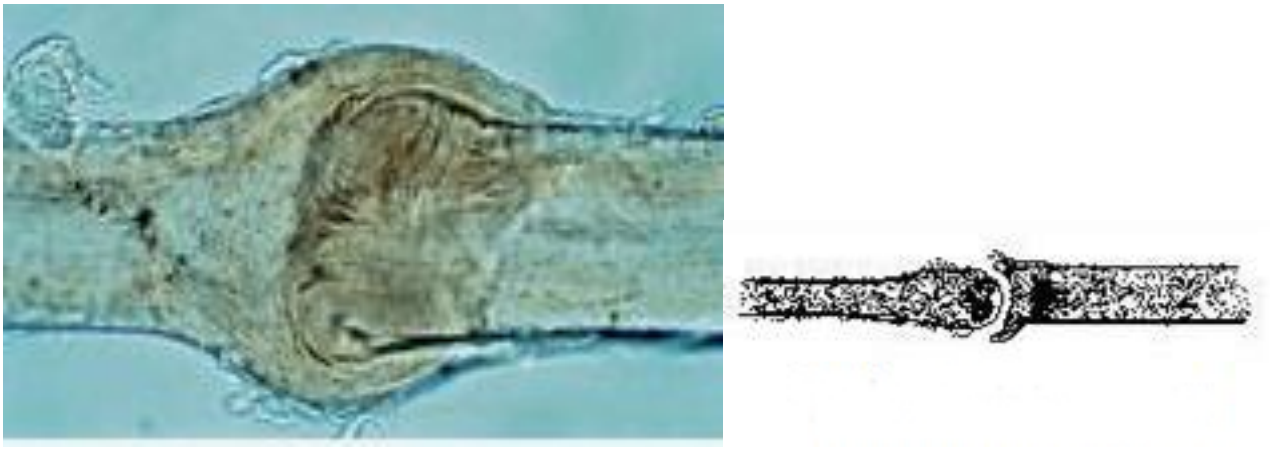
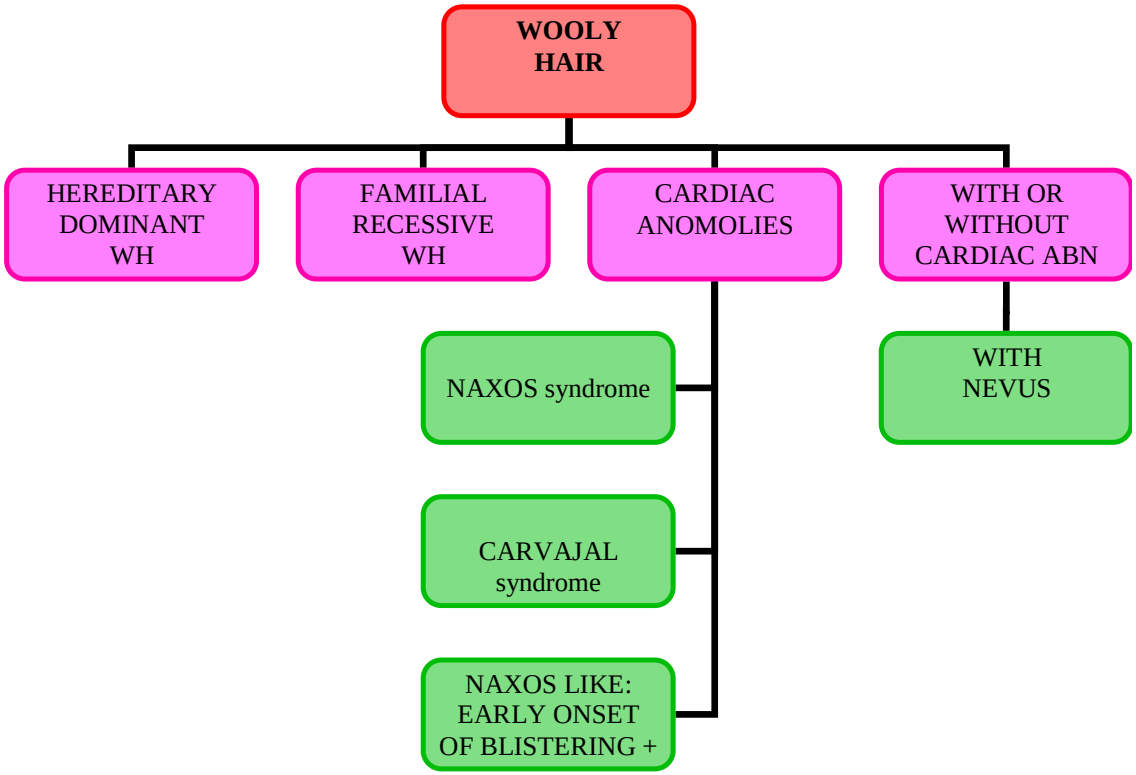
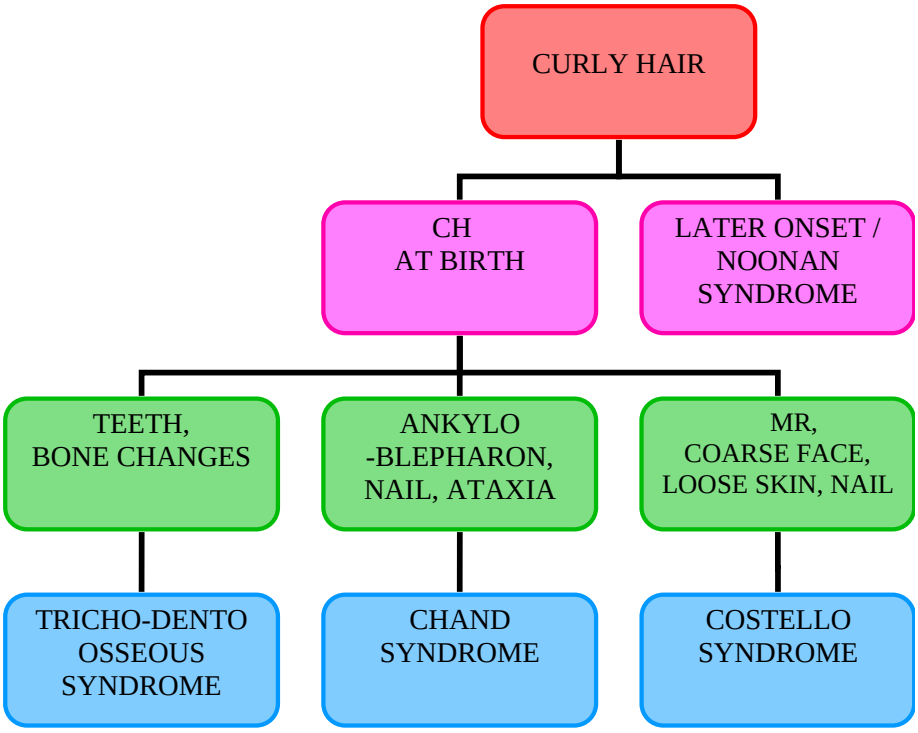


Figure 8: Trichorrhexis invaginata (a), simile to bamboo(b).

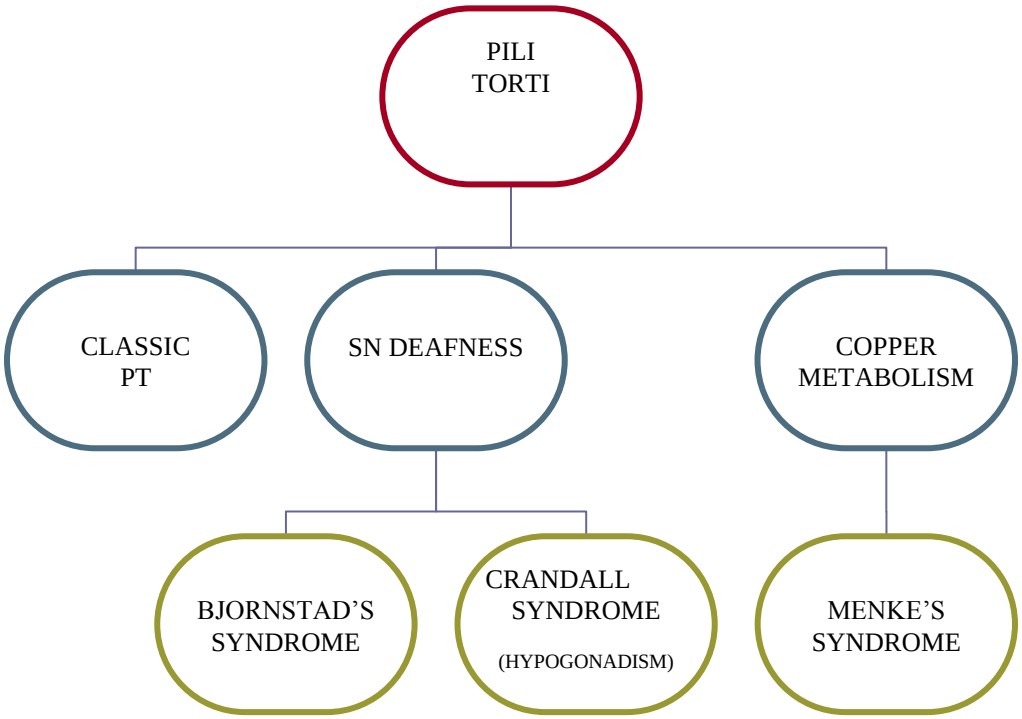
Flow Chart 1: Causes of wooly hair (WH) and other associated organ involvement



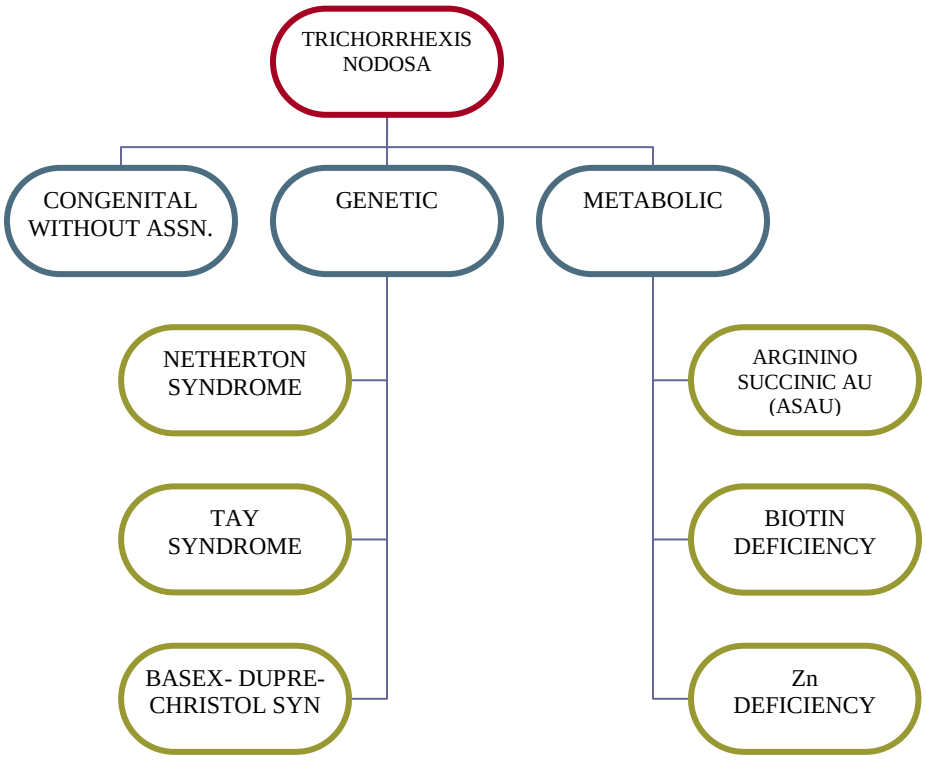

Flow Chart 2: Conditions associated with curly hair (CH)



Flow Chart 3: Syndromes associated with pili torti (PT)




Flow Chart 4: Trichorrhexis nodosa (TN) and associations



Flow Chart 5: Trichorrhexis nodosa (TN) as a marker for inborn errors of urea synthesis.