

The Role of Genetics in Hair Shaft Abnormalities and their Psychosocial Impact

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DESCRIPTION

Hair shaft abnormalities encompass a wide range of conditions that affect the structure, shape, and integrity of the hair shaft. These abnormalities can lead to brittle, fragile hair that breaks easily, and they often manifest as part of broader genetic syndromes or as isolated defects. Understanding the types, causes, clinical presentations, and management strategies for hair shaft abnormalities is important for dermatologists and healthcare providers in order to offer appropriate treatments and improve patient outcomes.

Types of hair shaft abnormalities

Hair shaft abnormalities encompass a range of structural defects that affect the appearance, strength, and texture of hair. These abnormalities can be congenital or acquired and often result in hair that is brittle, easily broken, or has an unusual texture or appearance. Here are some common types of hair shaft abnormalities:

Trichorrhexis nodosa: Trichorrhexis nodosa is characterized by the presence of nodes or swellings along the hair shaft, leading to breakage at these weak points. This condition can be either acquired, due to physical or chemical trauma to the hair, or congenital, resulting from genetic defects in the hair structure. Clinically, affected hair appears frayed or broom-like at the ends.

Monilethrix: Monilethrix is a hereditary disorder where the hair shaft exhibits a beaded appearance due to periodic narrowing and swelling. This condition is often noticeable from birth or early childhood and results in fragile hair that breaks easily. It is typically inherited in an autosomal dominant pattern, with mutations in genes such as *KRT81*, *KRT83*, and *KRT86* being implicated.

Pili torti: Pili torti involves the twisting of the hair shaft along its axis, giving the hair a corkscrew-like appearance. The condition is frequently linked to genetics and can be acquired with other systemic conditions, such as Menkes disease, Björnstad syndrome, and Bazex-Dupr -Christol syndrome. The twisted hair is prone to breakage and may lead to patchy alopecia.

Pili annulati: Pili annulati, or "ringed hair," is a condition where the hair shaft exhibits alternating light and dark bands when viewed under transmitted light. This condition is typically benign and does not usually cause hair fragility or breakage. It can be inherited in an autosomal dominant pattern, though the exact genetic cause remains unclear.

Bubble hair: Bubble hair is a condition characterized by the presence of air-filled cavities within the hair shaft, leading to weak and brittle hair. This abnormality is usually acquired and is often associated with excessive heat exposure from styling tools, such as hairdryers or curling irons.

Trichoptilosis: Commonly known as split ends, trichoptilosis involves the splitting of the distal hair shaft into multiple segments. This condition is primarily acquired due to mechanical damage, chemical exposure, or environmental factors. Regular trimming and protective hair care practices can help manage split ends.

Psychosocial impact and support

Hair shaft abnormalities can have a significant psychosocial impact on affected individuals. The visible changes in hair texture and integrity can lead to self-esteem issues, social anxiety, and emotional distress. Providing psychological support and counseling can be beneficial in helping patients cope with the emotional aspects of their condition.

Hair shaft abnormalities encompass a diverse range of conditions that affect the structure and integrity of the hair. These abnormalities can be congenital or acquired and may result from genetic mutations, environmental factors, or systemic conditions. Understanding the types, causes, clinical presentations, and management strategies for hair shaft abnormalities is important for providing effective care and improving the quality of life for affected individuals. Early diagnosis, appropriate treatment, and psychosocial support play key roles in managing these challenging conditions. Ongoing research and advances in genetic and molecular therapies hold promise for future development in the treatment of hair shaft abnormalities.

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