Monilethrix

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Monilethrix is a condition that affects hair growth. Its most characteristic feature is that individual strands of hair have a beaded appearance like the beads of a necklace. The name monilethrix comes from the Latin word for necklace (monile) and the Greek word for hair (thrix). Noticeable when viewed under a microscope, the beaded appearance is due to periodic narrowing of the hair shaft. People with monilethrix also have sparse hair growth (hypotrichosis) and short, brittle hair that breaks easily.

Keywords: genetic conditions

1. Introduction

Affected individuals usually have normal hair at birth, but the hair abnormalities develop within the first few months of life. In mild cases of monilethrix, only hair on the back of the head (occiput) or nape of the neck is affected. In more severe cases, hair over the whole scalp can be affected, as well as pubic hair, underarm hair, eyebrows, eyelashes, or hair on the arms and legs.

Occasionally, the skin and nails are involved in monilethrix. Some affected individuals have a skin condition called keratosis pilaris, which causes small bumps on the skin, especially on the scalp, neck, and arms. Affected individuals may also have abnormal fingernails or toenails.

2. Frequency

The prevalence of monilethrix is unknown.

3. Causes

Monilethrix is caused by mutations in one of several genes. Mutations in the *KRT81* gene, the *KRT83* gene, the *KRT86* gene, or the *DSG4* gene account for most cases of monilethrix. These genes provide instructions for making proteins that give structure and strength to strands of hair.

Hair growth occurs in the hair follicle, a specialized structure in the skin. As the cells of the hair follicle mature to take on specialized functions (differentiate), they produce particular proteins and form the different compartments of the hair follicle and the hair shaft. As the cells in the hair follicle divide, the hair shaft is pushed upward and extends beyond the skin.

The *KRT81*, *KRT83*, and *KRT86* genes provide instructions for making proteins known as keratins. Keratins are a group of tough, fibrous proteins that form the structural framework of cells that make up the hair, skin, and nails. The *KRT81* gene provides instructions for making the type II hair keratin K81 protein (K81); the *KRT83* gene provides instruction for making the type II hair keratin K83 protein (K83); and the *KRT86* gene provides instructions for making the type II hair keratin K86 protein (K86). The K81, K83, and K86 proteins are found in cells of the inner compartment of the hair shaft known as the cortex. These proteins give hair its strength and elasticity.

The *DSG4* gene provides instructions for making a protein called desmoglein 4 (DSG4). This protein is found in specialized structures called desmosomes that are located in the membrane surrounding certain cells. These structures help attach cells to one another and play a role in communication between cells. The DSG4 protein is found in particular regions of the hair follicle, including the hair shaft cortex. Desmosomes in these regions provide strength to the hair and are thought to play a role in communicating the signals for cells to differentiate to form the hair shaft.

In people with monilethrix, the cortex of the affected hair shaft appears abnormal. However, it is unclear how mutations in the *KRT81*, *KRT83*, *KRT86*, or *DSG4* genes are related to the abnormality in the cortex or the beaded appearance of the hair.

Some people with monilethrix do not have a mutation in one of these genes. These individuals may have a genetic change in another gene, or the cause of the condition may be unknown.

3.1. The Genes Associated with Monilethrix

- DSG4
- KRT81
- KRT83
- KRT86

4. Inheritance

Monilethrix can have multiple patterns of inheritance. When the condition is caused by a mutation in one of the keratin genes, it is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In rare cases, the condition results from a new mutation in the gene and is not inherited.

When the condition is caused by mutations in the *DSG4* gene, it is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- beaded hair

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