

## Monilethrix. Case report.

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### Summary

Monilethrix or beading of hair is an inherited defect of the hair shaft. It precociously starts during the first months of life and then persists throughout life with little or, mostly, no improvement at all. We present a 4-year-old boy with monilethrix and discuss the different aspects of the disease.

### Key words

Monilethrix, hair shaft defect.

**M**onilethrix is a rare dysplastic disorder of the hair shaft that is inherited as an autosomal dominant trait with high penetrance but variable expressivity (4). It usually starts after the second month of life, but its onset can be delayed until childhood or later. It is characterized by hair fragility and small follicular papules (1).

### Case report

A 4-year-old boy was brought to our department for evaluation of his hair problem. The hair, especially at the occipital and temporal areas of the head, was dry, thin, brittle and very fragile. Moreover, the length of the hairs in the affected areas was not more than 2 centimeters (Fig. 1, 2).

His parents informed us that the problem had started 2 years before and that since then the hair did never reach a normal length.

The physical and psychological development of the boy was within normal limits. Both parents had normal hair but their family history revealed that the mother's father had very dry and short hair, especially at the occipital area of his head.

Microscopic examination of hair showed symmetrical and identical in size elliptical nodes of the hair shaft that were separated by narrow internodes (Fig. 3, 4). This picture was characteristic and diagnostic for monilethrix.

We did not start any treatment but only advised the parents to avoid any trauma of the hair. At the follow-up, 6 months later, the boy's hair had the same length and showed no improvement.

### Discussion

Monilethrix, or beading of hair, is a rare inherited defect of the hair shaft. Inheritance is auto-

somal dominant. The gene appears to have high penetrance but variable expressivity. According to some reports, the disease can be also inherited as an autosomal recessive trait (1).

Monilethrix usually starts in the first months of life, although, in some cases, it does not become apparent until childhood or even later. At birth, hair may have normal appearance. Soon in the second or third month of life it is replaced by abnormal, dry and brittle hair that fractures easily and remains short -1 or 2 centimeters- because of its fragility. Follicular papules are also seen sometimes, especially on the nape of the neck.

The most common affected areas in this disorder are the nape and occiput. However, the entire scalp may be involved. Eyebrows, eyelashes, pubic, axillary and body hair may also be affected (1).

Diagnosis can be easily made by microscopic examination of the hair shaft. Elliptical nodes form along the hair shaft at intervals of 0.7 to 1mm and are separated by narrower internodes. Level with the latter breakage of hair occurs. The general structure of the follicle is otherwise normal (2).

Studies on scanning and transmission electron microscope demonstrated in some hair shaft the alterations as follows:

- a) considerable degeneration of root sheath cells;
- b) signs of breakage or invagination of cuticle cells;
- c) structural disorder of the cortex microfibrils, as well as axis deviation;
- d) decreased number of cortex cells (5, 3).

These signs can explain the thinning and fragility of the hair shaft (2).

Chromosomal studies of the hair shaft demonstrated mutations of the type 2 keratin gene cluster at chromosome 12q13, favoring the hypothesis of a defect in the hard keratins of hair and nails (5, 6).

The considerable variation in the severity of this condition in heterozygotic patients and the reported improvement during pregnancy support the hypothesis that other possible genetic or environmental factors may alter the expression of keratin genes (4).

In our case we assumed that the mother's father was affected by monilethrix, according to the family history, although we could not visit



Fig. 1



Fig. 2

Fig. 1, 2: Dry, thin, short hair at the occipital and temporal areas of the head.

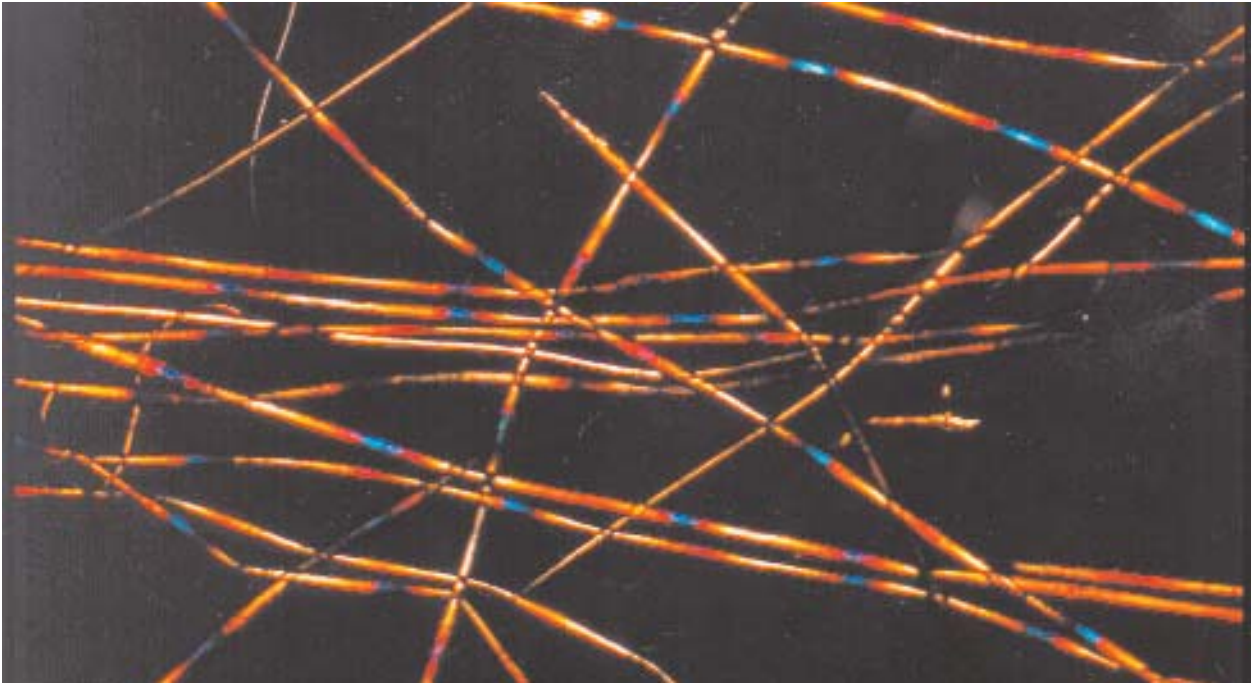


Fig. 3

Fig. 3, 4: Nodes and internodes of the hair shaft in microscopic examination under polar light microscope (Fig. 3->25x and Fig. 4->100x).

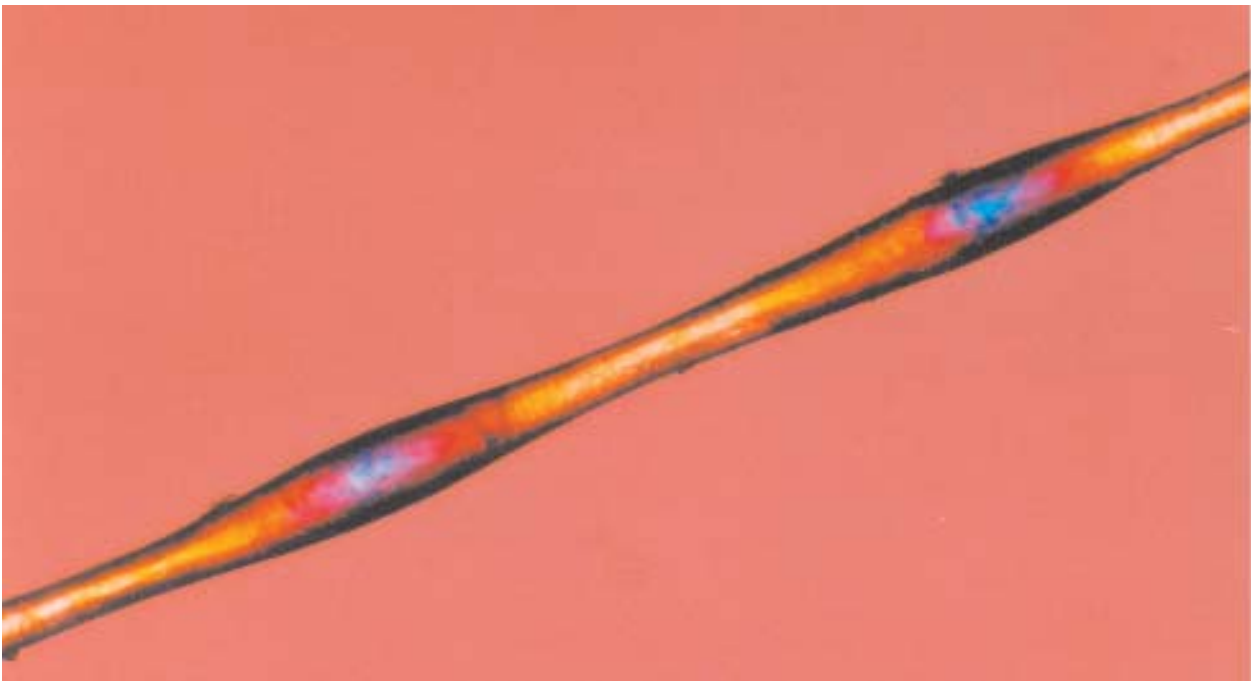


Fig. 4

him. We also assumed that the patient's mother had apparently normal hair, because of the variable expression of the gene.

The disease persists throughout life with little or no changes at all.

It is possible that there is association between monilethrix and other defects such as central

nervous system abnormalities, dentition problems, aminoaciduria. However, this association has not been defined.

There is no effective treatment for this disorder. Reduction of trauma may decrease hair fragility. There are reports that oral retinoids may induce a favorable response (1).

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