Monilethrix

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Abstract
Monilethrix is an autosomal dominant hair shaft dysplasia. This rare genodermatosis is characterized by periodic constrictions of the hair shaft giving the appearance of beaded hair. It is accompanied by fragility, local follicular hyperkeratosis and pronounced hypotrichosis. Patients may also present with nail dystrophy. Examination of this hair under light microscopy shows the periodic alternation of constrictions (defect) and nodes (real hair diameter). Electron microscopy reveals that the hair has broken off at the constriction areas. Monilethrix is due to a mutation in the type II keratins: hHb1 and hHb6. Treatment consists in topical retinoic and glycolic acid or minoxidil in postpuberal patients, although this condition improves over the years.

Keywords: Monilethrix, beaded hair, hHb1 keratin and hHb6 keratin

Disease name and synonyms

- Monilethrix
- Beaded hair

Excluded diseases

- Pseudomonilethrix
- Iatrogenic pseudomonilethrix

Diagnosis criteria / definition
This defect of the hair shaft is transmitted as a dominant trait. The hair appears short and beaded and is characterized by the presence of periodic constrictions of the hair shaft which are even observed at intrafollicular level (1). Local hyperkeratosis follicularis and pronounced hypotrichosis are also observed.

Differential diagnosis

- Pseudomonilethrix, although it may be associated to monilethrix; in the first condition no follicular hyperkeratosis is observed and the hair shaft defect is in the nodes.
- Iatrogenic pseudomonilethrix that is caused by excessive pressure of the hairs when taking the samples for microscopic examination.

Clinical description
Patients present with diffuse hypotrichosis with short, beaded and fragile hair. Strong follicular hyperkeratosis is also observed, especially in the occipital region where the defect may be more
evident. In some families, nail defects are associated (2). Additional non dermatological manifestations have been described: bilateral cataract, found blepharitis and corneal changes (pannus) (3). Clinical expression is variable, ranging from dystrophic hair confined to the occiput (mild cases) to nearly total alopecia (severe cases) (4).

**Epidemiology**

No data on prevalence or incidence are available. Most known cases are of European origin though an Indian and a Arab pedigree have been reported (5).

**Management**

This condition improves over the years, especially if hyperkeratosis is treated with topical retinoic and/or glycolic acids. Topical minoxidil could be tested in postpuberal patients (6).

**Etiology**

Monilethrix is an autosomic dominant hereditary defect due to a mutation in the type II keratins: hHb1 and hHb6. They were mapped to chromosome 12q13 (4). It usually affects several generations of the same family (7). The hHb1 mutation is associated with a less severe phenotype and with nail dystrophy (8).

**Diagnostic methods**

Monilethrix may be diagnosed by:

- light microscopy: examination of this hair allows one to determine the periodic alternation of constrictions (defect) and nodes (real hair diameter) causing the characteristic beaded aspect (6).
- scanning electron microscopy: similar image, proving that the hair has broken off at the constriction areas, where the longitudinal crests have been described (9).

**Unresolved questions**

No consensus about genetic and antenatal diagnosis in monilethrix has been reached yet. Perhaps, for some of the more severe phenotypes, prenatal diagnosis may be requested and this can now be performed from chorionic villus samples at an early stage of the pregnancy (10).

**References**