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## Letter

### Hypotrichosis with juvenile macular dystrophy: Portuguese case

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## Abstract

Hypotrichosis with juvenile macular dystrophy is a rare congenital disease mainly found in the Druze population of Northern Israel. This disorder is caused by the CDH3 mutation encoding P-cadherin, which is expressed in retinal pigment epithelium and hair follicles. An 11-year-old girl who was born to related Portuguese parents, had hypotrichosis since birth and macular dystrophy diagnosed at age 5. Fundus examination and fluorescein angiography revealed located macular pigmentary abnormalities. No molecular analysis was done. A fundus examination should be considered mandatory in the assessment of congenital hypotrichosis.

**Keywords:** hypotrichosis; macular dystrophy; P-cadherin; CDH3 mutation

## Background

Hypotrichosis with juvenile macular dystrophy is a rare autosomal recessive disorder, mainly described in Israeli families of Arab Muslim origin. We report a case of this disease in a Portuguese family.

## Case synopsis

Our patient, an 11-year-old girl, was the second daughter of third-degree consanguineous parents who were from Portugal. She was seen in our department for short and slowly growing hair present since birth; she did not have a sweating abnormality. Her past medical history included macular dystrophy diagnosed at age 5 without any visual impact. There was no family history of hair or visual impairment and her brother was unaffected.

Physical examination revealed hypotrichosis, sparse short hairs over the entire scalp without alopecia or erythema. The hair pull test was negative. The eyebrows and eyelashes were normal and no abnormalities of skin, teeth, or nails were noted. General examination of the respiratory, cardiovascular, and central nervous systems did not show abnormalities. A complete blood count was normal. The blood iron, ferritin, vitamin B12, and thyroid hormone levels were all within normal ranges.

Trichogram analysis demonstrated a reduced diameter of hair and polarized light examination showed non-specific hair dystrophy. Fundus examination and fluorescein angiography revealed localized macular pigmentary abnormalities. Visual acuity and ERG testing were normal. Molecular analysis of the CDH3 gene was not done.



**Figure 1.** Hypotrichosis with sparse short hairs over the entire scalp



**Figure 2.** Fundus examination: located macular pigmentary abnormalities

## Discussion

Hypotrichosis with juvenile macular dystrophy is a rare autosomal recessive disorder. It was first described in 1935 by Wagner in two German sisters [1]. It is characterized by hypotrichosis and progressive macular dystrophy leading to blindness. Hair abnormalities include short and sparse scalp hair, which apparently results from an increased proportion of involuting hair follicles, thus suggesting a cell cycle defect [2]. Various forms of macular dystrophies have been reported, including hypopigmentation, mild pigmentary changes, and severe hyperpigmentation [2]. Although hair changes are stable, the retinal disorder is progressive, affecting cone-mediated as well as rod-mediated vision, and causing decreased visual acuity and early blindness between the second and the fourth decades of life [3]. Hypotrichosis with juvenile macular dystrophy is caused by a CDH3 mutation. The CDH3 gene locus encodes P-cadherin, a classical cadherin molecule and a major component of the adherens junction in several tissues including retinal pigment epithelium and hair follicles [4]. The disease was shown in families of various origins and mainly found in Israeli families of Arab Muslim origin. Those families belonged to the Druze population who had lived in mountainous areas of the middle east as a closed society almost from their inception in Cairo around 1017 A.D.[3, 5]. Several CDH3 mutations have been described causing this disease; the missense R503H mutation was reported to be the most common CDH3 mutation in the Muslim Arab Israeli population [3].

Our patient was born to related Portuguese parents. Two other Portuguese cases had been previously described in two siblings in 1995. The parents of these patients were not consanguineous, but were native of the same village. They had abnormally sparse hair and at the age of 25 years had still never cut their hair. Ophthalmologic examination revealed reduced visual acuity and macular pigmentary degenerative changes in both of them. No molecular analysis was done [6].

Earlier identification of hypotrichosis with juvenile macular dystrophy patients allows for educational measures, appropriate counselling, and visual rehabilitation to be provided. Fundus examination should be considered as mandatory in the assessment of congenital hypotrichosis.

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