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Alopecia as a prominent feature of myotonic dystrophy type 1

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Alopecia is a common finding in myotonic dystrophy type 1 (MD1) but it is usually restricted to the fronto-temporal areas (frontal baldness).[1-3] Alopecia also of other areas of the scalp has not been reported as a dominant feature of the disease.

The patient is a 54yo Caucasian female with a history of myotonia of all toes bilaterally since childhood, recurrent falls since at least 15a, disturbed fine motor skills since at least 15a, and progressive gait disturbance from distal weakness with a foot drop and abortive stepping gait such that walking over steps was possible only with holding the handrail. She also reported multiple atheromas on the scalp since 15-20y, dysphagia since 10y, cholecystectomy, easy tiring, and exercise intolerance. She had undergone cataract surgery bilaterally some years ago. Since 2-3 years prior to the last visit she had developed extra-fronto-temporal alopecia to such a degree that she was wearing a wig since age 53y. Her family history was positive for MD1 in her oldest daughter and her son, her two sisters and her brother. Two of her children were unaffected.

Clinical neurologic examination revealed a myopathic face, atheroma over the scalp, alopecia over the entire scalp (Figure 1), slight bilateral ptosis, weakness for lid closure, slight weakness for finger straddling on the upper limbs, diffuse weakness of the lower limbs with distal and right-sided predominance, reduced tendon reflexes on the upper as well as lower limbs, and discrete distal wasting on the lower limbs. She was unable to walk on her heels but also toe-walking was slightly impaired. There was recurrent mild hyper-CK-emia with a maximal value of 279 U/L (n, < 146 U/L). Serum testosterone was 0.13 ng/mL (n, 0.06-0.8 ng/mL). Other hormones, such as follicle stimulating hormone, luteinizing hormone, prolactin, estradiol, progesterone, and thyroidea stimulating hormone were also within normal limits. Needle electromyography revealed the typical myotonic and pseudomyotonic discharges in all muscles investigated. Molecular genetic investigations revealed an abnormal, expansion of 500 CTG-repeats within the DMPK gene on chromosome 19q13.3. 24h-ECG showed sinusrhythm throughout the entire recording, one pause of 2.4ms, and only occasionally ventricular or supraventricular ectopic beats. Abdominal ultrasound showed mild steatosis hepatis. Echocardiography was normal. Videocinematography at age 52y was normal.

MD1 is an autosomal dominant trinucleotide disease, caused by an unstable CTG-repeat expansion of > 50 repeats in an untranslated region of the DMPK gene on chromosome 19q13.3.[3] The expansion on the DNA level results in impaired RNA-processing not only of the DMPK-RNA but also of other RNAs,[3] why MD1 presents as a pleiotropic multisystem disease, affecting the eyes (cataract), muscles (myopathy with ptosis, myopathic face, myotonia and wasting), heart (conduction block, cardiomyopathy), endocrine organs (insulin resistance, erectile dysfunction, hypogonadism), and the cerebrum (dementia, reduced alertness).[2] There is no causal therapy available but conservative measures, such as surgical ptosis correction, diabetes therapy, pacemaker implantation, implantation of an implantable cardioverter defibrillator, or administration of mexiletine tocainide, or flecainide for myotonia may alleviate the clinical manifestations.[4]

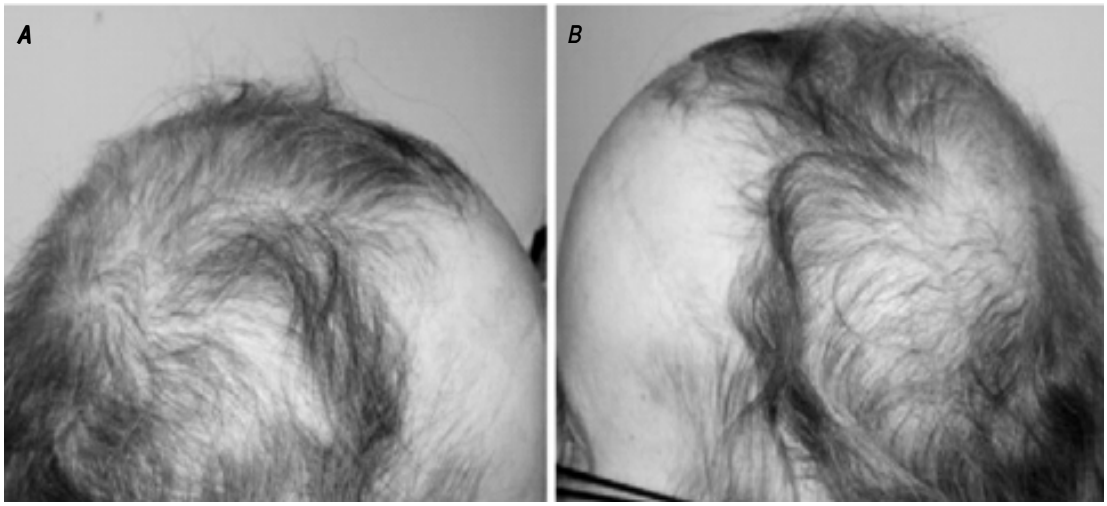

Figure 1. Patchy alopecia of the scalp in a patient with myotonic dystrophy type 1 developing since age 51y.

Dermal abnormalities are a frequent manifestation of MD1 and include pilomatricomas, basaliomas, acne, hidradenitis supportiva, keratosis piloris, pigmental basal cell cancer, and alopecia.[5,6] Alopecia is defined as non-physiologic pronounced loss of hair due to various abnormalities, such as fungal infections (tinea capitis), lichen planopilaris, pseudopelade of Bricq, tuffed folliculitis, dissecting cellulitis, keratosis follicularis, or alopecia mucinosa), secondary syphilis, iron deficiency, hereditary disorders of the hair shaft, hormonal impairment (increased testosterone levels, hypothyroidism), chemotherapy, frequent use of chemical relaxer, compulsive hair pulling like in *trichotillomania*, discoid lupus erythematosus or chronic cutaneous lupus erythematosus, chronic exposure to traction on hair, repeated hot comb use, radiation therapy, or physical or psychological stress.[7]

Alopecia has been repeatedly reported as a clinical feature of MD1,[2,8,9] but in the majority of the cases loss of hair is restricted to the frontal regions (frontal baldness).[2,3] In a 27yo female with the association of MD1 and celiac disease, alopecia was initially attributed to the enterologic disorder but after resolution of the gastro-intestinal tract disorder was lastly interpreted as a manifestation of MD1.[9] Alopecia in MD1 is attributed to disturbances of the endocrine glands, in particular of the androgen metabolism with either an increase in the serum androgen levels or due to an increased sensitivity of the androgen receptors.[6] More likely it is not the absolute circulating amount of androgen but rather the peripheral response to the hormone which causes alopecia.[6] How oversensitivity of the androgen receptors could be related to the reduced DMPK levels, however, remains speculative. Alopecia in the presented case was also attributed to MD1 since the above mentioned other causes were excluded.

This case shows that alopecia in a non-frontotemporal distribution may be a rare phenotypic feature in MD1. Whether this feature is predominantly found in female patients or unrelated to sex is unknown. Extensive alopecia of the scalp in MD1 may be attributed to oversensitivity of the androgen receptors since serum levels of testosterone were normal.

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