The Skin: another organ to be considered in Myotonic dystrophy type 1 diagnosis

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BACKGROUND

Myotonic dystrophy type 1 (DM1 or Steinert's disease) is the most frequent muscular dystrophy in adults, caused by expansion of a CTG trinucleotide repeat in the non-coding region of dystrophia myotonica protein kinase (DMPK) gene, located on chromosome 19 and characterized by progressive myopathy, myotonia and multiorgan involvement that affects skeletal and smooth muscle as well as the eye, heart, endocrine system, and central nervous system [1,2]. To date, few studies have been performed to evaluate skin features in MD1 patients compared to control subjects, showing controversial features: a significantly higher numbers of nevi, dysplastic nevi, melanomas and pilomatrixoma have been reported [3], whereas others authors do not rule out an increased prevalence of pre-neoplastic and neoplastic skin lesions [4].

Aim of this study is to evaluate the prevalence of neoplastic, proliferative, functional and inflammatory skin lesions in a largest group of MD1 patients, compared to controls, by complete clinical examination of their skin and mucosae.

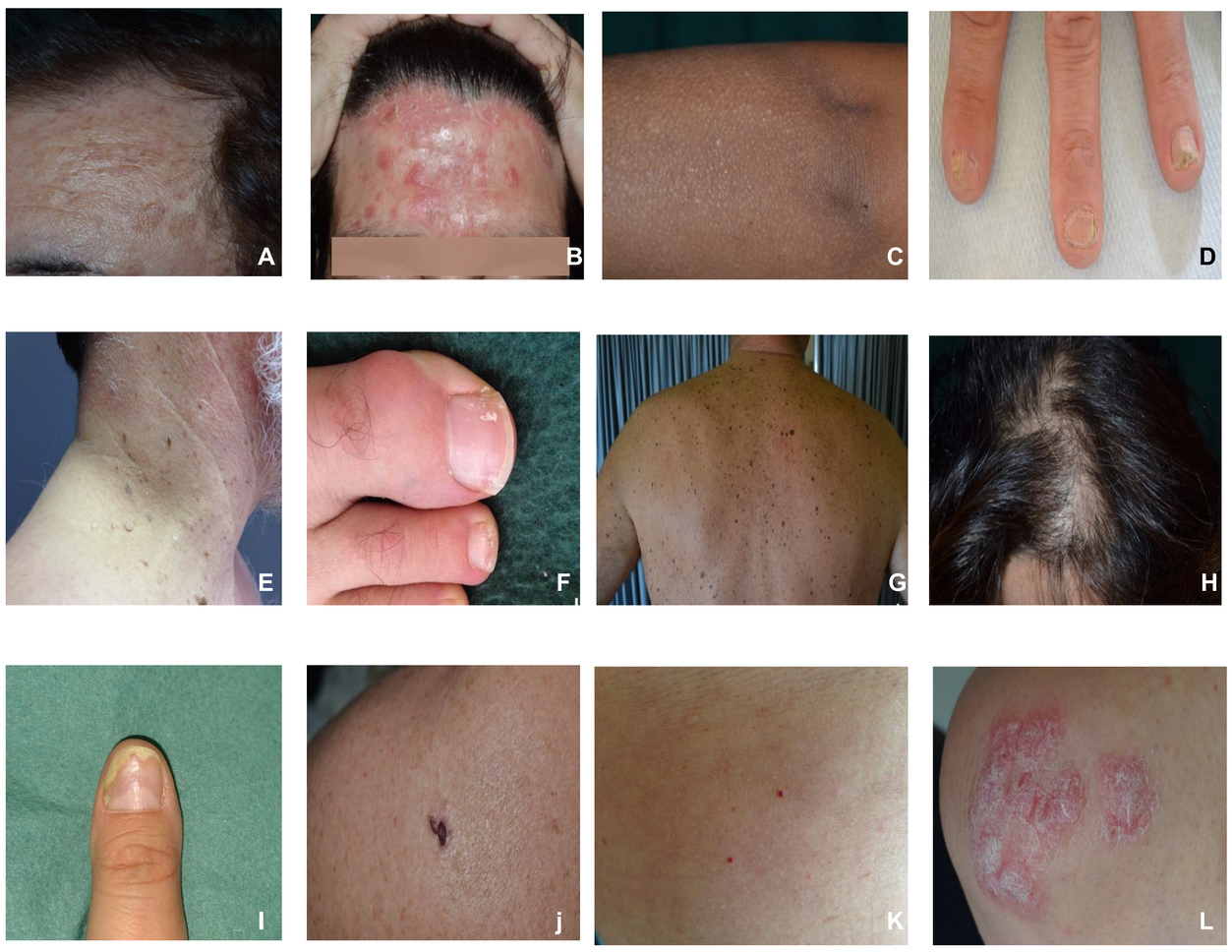
Materials and methods

103 Caucasian patients, affected by genetically confirmed MD1, with a different disease severity, were referred to the same dermatologist, between January and December 2015.

58 males and 45 females, aged from 10 to 78 years (average: 48 years).

103 subjects, paired for age and gender with the MD1 patients, were matched as controls.

All the subjects involved in the study received a complete clinical examination of skin and mucosae and an intra-vital digital videodermoscopy through the Fotofinder Dermoscope by a trained dermatologist to check for the presence of skin lesions.



Significant skin features of DM1 patients: A.) Hyperpigmentation, B.) Seborrheic dermatitis, C.) Follicular hyperkeratosis, D.) Linear nail dystrophy, E.) Mollusca fibrosa, F.) Leukonychia, G.) Melanocytic nevi, H.) Early androgenic alopecia, I.) Nail 'pitting', J.) Basal cell carcinomas, K.) Ruby angiomas, L.) Psoriasis.

SKIN LESIONS	DM1 PATIENTS N (%)	CONTROLS N (%)	X^2/F test (df = 1)
Hyperpigmentation	68 (66.0%)	3 (2.9%)	88.0 ****
Seborrheic dermatitis	67 (65.0%)	12 (11.6%)	59.8 ****
Follicular hyperkeratosis	50 (48.5%)	13 (12.6%)	29.6 ****
Nail distrophy	27 (26.2%)	0 (0.0%)	28.8 ****
Mollusca fibrosa	66 (64.1%)	27 (26.2%)	28.3 ****
Leukonychia	38 (36.9%)	7 (6.8%)	25.6 ****
Melanocytic nevi (>50)	66 (64.1%)	31 (30.1%)	22.5 ****
Early androgenic alopecia	35 (34.0 %)	8 (7.8%)	19.9 ****
Nail pitting	25 (24.3%)	4 (3.9%)	16.1 ****
Basal cell Carcinoma	24 (23.3%)	4 (3.9%)	14.9 ***
Ruby angiomas	42 (40.1%)	18 (17.5%)	12.4 ***
Psoriasis	20 (19.4%)	6 (5.8%)	7.4 **
Guttate hypomelanosis	14 (13.6%)	3 (2.9%)	6.4 *
Pilomatricomas	5 (4.8%)	0 (0.0%)	3.3 n.s.
Angiomas of the oral cavity	5 (4.8%)	0 (0,0%)	3.3 n.s.
Melanoma	1 (1.0%)	7 (6.8%)	3.2 n.s.
Acne	1 (1.0%)	7 (6.8%)	3.2 n.s.
Eczema	1 (1.0%)	7 (6.8%)	3.2 n.s.
Warts	1 (1.0%)	7 (6.8%)	3.2 n.s.
Skin xerosis	28 (27.2%)	17 (16.5%)	2.8 n.s.
Seborrheic keratosis	48 (46.6%)	42 (40.1%)	0.5 n.s.
Dysplastic nevi	19 (18.4%)	23 (22.3%)	0.3 n.s.
Fibrous histicytomas	19 (18.4%)	23 (22.3%)	0.3 n.s.
Actinic keratosis	19 (18.4%)	15 (14.6%)	0.3 n.s.
Hyperhidrosis	1 (1,0%)	1 (1.0%)	0.01 n.s.
Hidradenitis suppurativa	1 (1.0%)	1 (1.0%)	0.01 n.s.
Squamous cell carcinoma	1 (1,0%)	1 (1.0%)	0.01 n.s.
Vitiligo	1 (1.0%)	1 (1.0%)	0.01 n.s.
Hyperkeratosis of the nipple and areola	1 (1.0%)	1 (1.0%)	0.01 n.s.
Terra firma-forme dermatosis	1 (1.0%)	1 (1.0%)	0.01 n.s.
Pityriasis versicolor	1 (1.0%)	1 (1.0%)	0.01 n.s.
Onychomycoses	1 (1.0%)	1 (1.0%)	0.01 n.s.

DISCUSSION

The attention to skin involvement in MD1 is an already reported data in literature, and several studies have been performed in the last decade to better define a possible cutaneous 'phenotype' of the disease. Our study failed to demonstrate any significant prevalence of neoplastic skin lesions, except for basal cell carcinoma. Although a great percentage (64.1%) of MD1 patients presented with more than 50 melanocytic nevi, only 20 underwent surgical excision, histologically reporting 19 dysplastic nevi and one superficial spreading melanoma. Furthermore, similar with the results showed by Campanati et al. [4], we showed a highest prevalence of adnexal manifestations, like follicular hyperkeratosis and early androgenic alopecia. In our study, we underlined prevalence of hyperpigmentation, seborrheic dermatitis, mollusca fibrosa and nail disorders. These latest seem to represent a possible hallmark of the disease.

CONCLUSIONS

Finding an explanation for these skin alterations needs to understand better the disease pathogenesis**.** You could hypothesize that ribonuclear inclusions can play an important role.

In conclusion, we suggest to consider skin and nail alterations as part of MD1 picture and, so, to evaluate their presence in disease assessment.

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