COMMUNICATION

Dermatoscopic aspects of the Microphthalmia with Linear Skin Defects (MLS) Syndrome*

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Abstract: The association of microphthalmia and linear skin defects was named microphthalmia with linear skin defects syndrome (MLS) or MIDAS syndrome (microphthalmia, dermal aplasia, and sclerocornea), an X-chromosomal disorder manifesting mainly in females. We examined a female newborn with facial linear skin defects following the Blaschko lines. Computer tomography and ophthalmological examination confirmed bilateral microphthalmia. An interstitial microdeletion at Xp22.2, encompassing the entire HCCS gene, was identified. Dermatoscopic examination showed erythematous linear areas with telangectasias and absence of sebaceous glands, which appear as brilliant white dots. Vellus hairs were also absent in the red areas. Dermatoscopy could help to establish the diagnosis of MLS/MIDAS syndrome by confirming the aplastic nature of the lesions.
Keywords: Congenital abnormalities; Dermoscopy; Genetic diseases, X-linked; Skin diseases, genetic

The association of microphthalmia and linear skin defects was described in the early 1990s. This condition was named microphthalmia with linear skin defects syndrome (MLS) or MIDAS syndrome (microphthalmia, dermal aplasia, and sclerocornea).[1-4] It can be genetically and clinically distinguished from focal dermal hypoplasia, a condition also associated with linear aplastic defects.[1,4]

We examined a female newborn, who presented congenital facial linear skin defects following the Blaschko lines, some of them covered with hemorrhagic crusts (Figure 1). Computer tomography showed normal central nervous system and small eyes. Ophthalmological examination revealed sclerocornea and confirmed bilateral microphthalmia. A microdeletion of a minimum size of 3 Mb which encompasses one of the two causative genes for MLS syndrome, HCCS at Xp22.2[5] was detected in the patient but was absent in her mother.

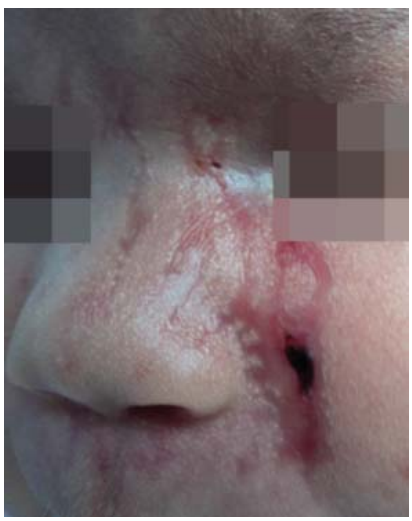

Figure 1: Linear facial defects following the Blaschko lines.

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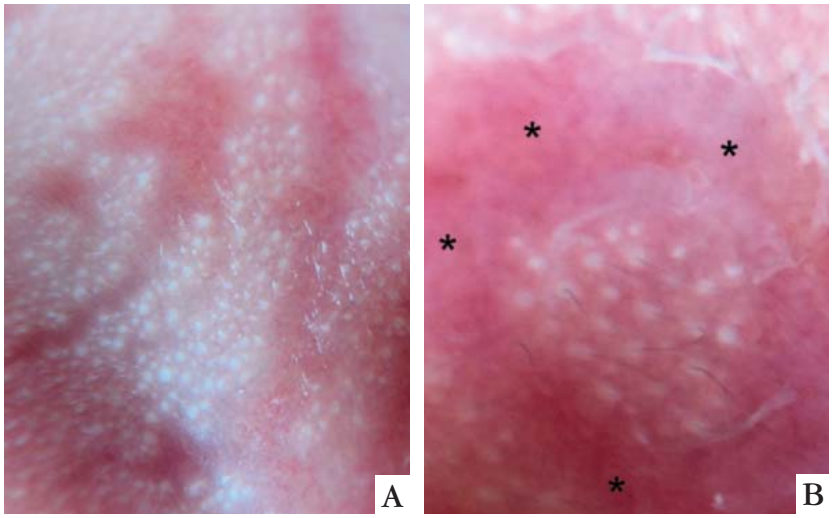

FIGURE 2: Dermatoscopic findings **A.** linear absence of sebaceous glands (NO brilliant white dots). **B.** absence of vellus hairs and sebaceous glands in the affected area (asteriscs).

Dermatoscopic examination showed erythematous linear areas with telangectasias and absence of sebaceous glands, which appear as brilliant white dots (Figure 2A). Vellus hairs were also absent in the red areas (Figure 2B).

The skin defects of this peculiar genodermatosis are described as dermal aplasia. It has been linked to segmental aneuploidy of the Xp22.2 region or intragenic mutations in the HCCS gene, which encodes the mitochondrial holocytochrome c-type synthase.[5-7] To date most cases described have been female and a X-linked male-lethal trait was suggested.[4,6] Similar to other X-linked genetic conditions, rare male cases have been described.[8]

Dermatoscopy is widely used in the diagnosis of melanocytic lesions and could also help to establish the diagnosis of MLS syndrome by confirming the aplastic nature of the lesions, with the absence of cutaneous adnexes, such as sebaceous glands and vellus hairs, in a linear distribution.[9] ❑

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