BRIEF ARTICLES

Geometric Facial Erosions on a Newborn

Shelby Stribling, BA[1], Matthew LaCour, BS[1], Lindy Ross, MD[1], Sharon Raimer, MD[1], Brent Kelly, MD[1]

[1]Department of Dermatology, University of Texas Medical Branch, Galveston, TX

ABSTRACT

When a newborn exhibits dermal aplasia and linear erosions on the face and neck, especially if there are ocular anomalies, further investigation is needed to determine if he or she has MiDAS (microphthalmia, dermal aplasia, and sclerocornea) or other syndromes with associated skin findings. MiDAS syndrome or MLS (microphthalmia with linear skin defect) is an X-linked dominant genodematosis characterized by cutaneous, ocular, central nervous system, and cardiac defects. It is essential to diagnose MiDAS syndrome early in life to allow for a thorough workup. This workup is to determine if there are any associated abnormalities in the child that require a multidisciplinary approach for diagnosis and treatment. While the skin lesions of MiDAS syndrome heal spontaneously, other associated abnormalities require early intervention and can be life threatening. This case report describes the work up, diagnosis, etiology, potential complications, and necessary follow up of MiDAS syndrome in a new born African American female.

CASE REPORT

A two-day old African American female presented with abnormal skin findings present at birth. She was born at 38 weeks and 1 day by spontaneous vaginal delivery with APGARs of 7 and 8 at one and five minutes. The infant's mother had a past medical history of type II diabetes mellitus, AV block, eczema, schizoaffective/bipolar disorder, possible first trimester illicit drug use, and syphilis treated in 2006. There was no family history of congenital defects. Upon examination of the patient, sharply demarcated erosions were present on the bilateral cheeks with scalloped, geometric borders (Figure 1), as well as erosions on the intertriginous areas of the neck and a small

Figure 1: Bilateral geometric facial erosions on the cheeks of a newborn present at the time of birth.

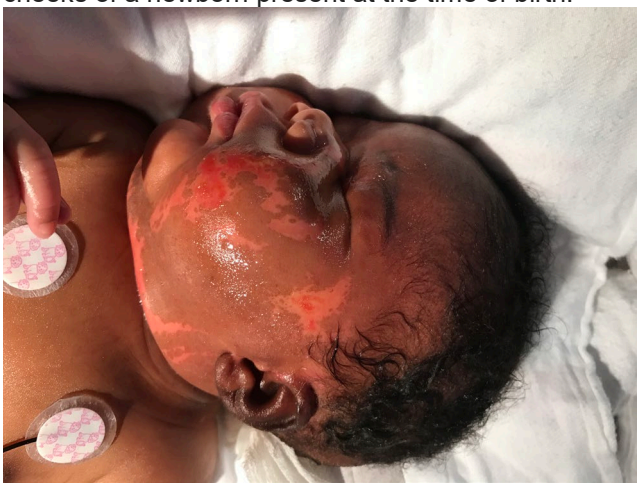


Figure 2: Right sided micropthalmia and corneal opacity.

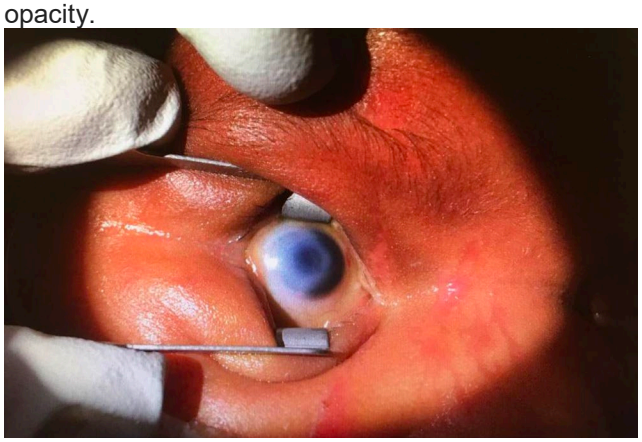
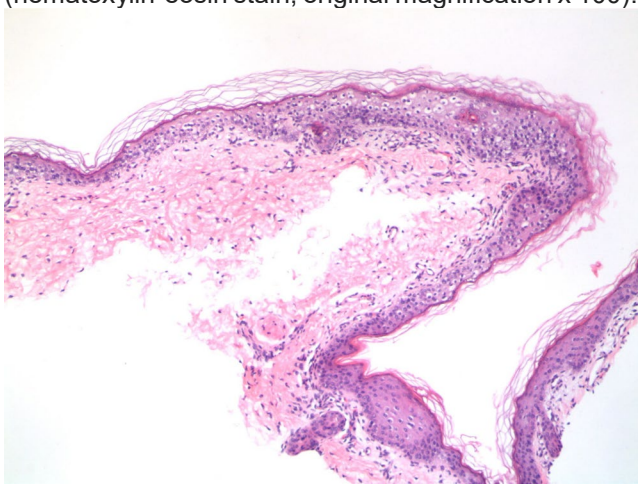


Figure 3: Skin biopsy of an eroded plaque (hematoxylin-eosin stain, original magnification x 100).



eroded macule on the left labia majora. A wound culture of the skin lesions was negative, and bacitracin was applied by the pediatric team. Other notable findings on exam included left micropthalmia and bilateral corneal opacities (Figure 2). Also, on the first day of life, a 3 X 3 cm occipital encephalocele containing a small tongue of cerebellar tissue and abnormal venous structures was repaired.

The biopsy specimen showed upper epidermal pallor and minimal perivascular lymphocytic infiltration with no evidence of lupus or primary bullous disorder (Figure 3). Epidermal pallor can be a nonspecific histological finding and may present in the setting of various nutritional deficiencies. This diagnosis was considered, but it was thought to be less likely in this patient who was receiving good nutritional support and had normal laboratory values. Laboratory studies were negative for Anti-SSA and Anti-SSB; Serum zinc and albumin were within normal limits as were newborn screening test results. Chromosomal microarray analysis revealed a 8,165 kbp loss of chromosome Xp22.32p22.2, which is consistent with MiDAS syndrome.

DISCUSSION

MiDAS syndrome (microphthalmia, dermal aplasia, and sclerocornea) or MLS (microphthalmia with linear skin defect) is an X-linked dominant condition first described in the early 1990s. MiDAS syndrome displays great phenotypic variability in females while appearing to exhibit lethality in hemizygous males in utero. MiDAS syndrome is most often caused by a deletion or translocation of Xp22.3 encoding the gene HCCS. HCCS encodes the mitochondrial holocytochrome c type synthase that is involved in complex III of the mitochondrial respiratory chain (MRC) that carries out oxidative phosphorylation.[1] A mutation of the COX7B gene at Xq21.1 has been identified which encodes for cytochrome c oxidase or complex IV of oxidative phosphorylation. and is more commonly associated with patients who only exhibit the dermal aplasia of MiDAS syndrome without ocular or other anomalies.[2]

However, there have been reported cases of MiDAS syndrome with no mutations of the HCCS and COX7B genes, but genetic analysis shows a skewed pattern of inactivation of the X chromosome.[3] Mosaicism of X chromosome inactivation is also believed to contribute to the vast array of phenotypic presentations of sporadic as well as heritable cases of MiDAS syndrome.[1,4]

The linear skin lesions follow the Blaschko lines, which form as embryonic cells migrate during fetal development. The skin findings of MiDAS syndrome are typically confined to the face and neck region. Dermatoscopic examination of the lesions reveals erythematous areas without evidence of sebaceous glands or vellus hair, and although the linear skin defects can appear to be erosions they actually represent profound atrophy of the skin and are similar but distinct from the dermal hypoplasia seen in Goltz syndrome.[5,6] While Goltz syndrome presents with linear hyper and hypopigmented patches, skin involvement is not limited to the head and neck, and patients with this condition will also have fat herniation and skeletal abnormalities.[6,7] Aicardi syndrome can present with similar ocular and neurological features as MiDAS syndrome but without the characteristic linear skin findings. All three of these diseases are X-linked with some overlap of clinical features which has led some authors to posit they may involve a contiguous region of the Xp chromosome.[8] With time, the dermal aplasia of MiDAS syndrome heals with hyperpigmentation, and residual scarring can occur.

Microphthalmia is the most common associated ocular abnormality, and it can present unilaterally or bilaterally. Additional documented ocular findings include sclerocornea, congenital glaucoma, retinal abnormalities, cataracts, microcornea, optic nerve hypoplasia, and anterior chamber defects. Many other clinical features have been associated with MiDAS syndrome such as central nervous system abnormalities like agenesis of the corpus callosum and microcephaly, speech and cognitive delay, genitourinary abnormalities, short stature, and cardiac defects with case reports of sudden cardiac death before one year of age.[7,9] Due to the many conditions that can be associated with MiDAS syndrome, patients with this diagnosis should have full cardiac workup, MRI of the brain, audiology testing, and ophthalmologic exam.

When a newborn exhibits dermal aplasia on the face and neck, especially if there are ocular anomalies, further investigation is needed to determine if they have MiDAS or other syndromes with associated skin findings. It is essential to diagnose MiDAS syndrome early in life to allow for a thorough workup to determine if there are any associated abnormalities in the child that require treatment. While the skin lesions of MiDAS syndrome heal spontaneously, other associated abnormalities require early intervention and can be life threatening.[10]

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Corresponding Author:
Matthew LaCour, BS
University of Texas Medical Branch
Galveston, TX
mdlacour@utmb.edu

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