01. **Title: Diagnostic Evaluation of Cutaneous Mastocytosis with Atypical Urinary Manifestations: A Case Report**

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Background: This case study involves the progression of a diagnosed incidence of cutaneous mastocytosis from birth to 9 months. Cutaneous mastocytosis is a rare condition, seen in all ages, presenting with progressive inflammatory dermatologic symptoms and rare systemic organ damage. Mastocytosis is classified into cutaneous and systemic categories, with a predilection for children and adults, respectively. The pathogenesis of cutaneous mastocytosis involves increased mast cell proliferation and hyperactive inflammatory responses, progressively worse with age. Diagnosis is confirmed with a skin biopsy and genetic testing. Treatment primarily involves symptomatic control, utilizing anti-inflammatories, steroids, light therapy, immunologic mediators, and KIT gene-targeted therapy. This study is especially unique in that systemic symptoms, such as hydronephrosis and vesicourethral reflux, are rare, however plausible, as supported by pathogenesis. Due to the difficulties of proper dermatologic testing and availability of genetic testing, different diagnostic and treatment modalities were experimented.

The Case: A 7-month year old female presents to the pediatrician clinic after a diagnosis of cutaneous mastocytosis. While patient is stable and in no signs of distress, signs of the condition are still evident with remitting and relapsing maculopapular lesions throughout the central trunk. Additionally, a primary healing scar is present on the left wrist, persisting as an open wound with a central pink clearing. Recurrent episodes of systemic flushing and polymorphic maculopapular rashes have occurred every month since birth, however, these symptoms have been treated supportively with Benadryl as needed. Concurrently, a comorbid nodule on the left ear, a left nasolacrimal duct stenosis, signs of urinary tract infection with incidental hydronephrosis, and vesicourethral reflux are also being monitored as either incidental or provoked by the mastocytosis.

Conclusion: While this patient's renal and urethral anomalies are benign, they will be monitored with serial imaging. After being treated symptomatically for cutaneous and urinary tract symptoms, the patient is advised to undergo genetic testing for both cutaneous mastocytosis and urinary tract anomalies for more individualized treatments.

Key Words: Cutaneous Mastocytosis, Systemic Mastocytosis, Hydronephrosis, Vesicourethral reflex, Maculopapular rash

Figure or Table:

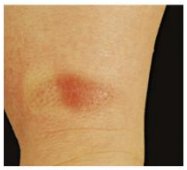


Figure 1A

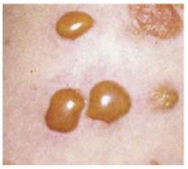
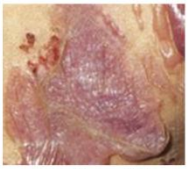


Figure 1B



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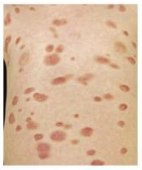

Figure 2A

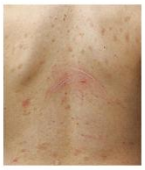

Figure 2B

Figure 1A: Darier's Sign. A pathognomonic indication of Cutaneous Mastocytosis is a Darier's Sign, defined as a wheel-and-flare reaction initiated by rubbing lesional skin with a tongue spatula. While not confirmed, it is hypothesized to have been potentially been present in this case at the left wrist of this neonate, potentially triggered by some mechanical irritation, postpartum.

Figure 1B: Bullous Cutaneous Mastocytosis. As classically named xanthelasmoid cutaneous mastocytosis, the primary lesion on the left wrist progressed into a single bullous serous lesion, predicted to be filled with yellow pustular fluid, histamines, other inflammatory biomarkers.

Figure 1C: Scar Remanent. The primary bullous lesion eventually burst and healed into a brown plaque with a central pink erythematous patch.

Figure 2A: Polymorphic: Urticaria Pigmentosa. During monthly flares, polymorphic maculopapular blisters filled the patient's trunk, face, and proximal extremities.

Figure 2B: Subsided Urticaria Pigmentosa. After persisting for less than 1 hour and alleviating symptoms with Benadryl cream, patient would be left with minimal, indistinct scars throughout the body. As noted in previous studies, polymorphic lesions are more common in children and adolescence, while monomorphic, uniform lesions are more prevalent in adults. Both are classified as urticaria pigmentosa.

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