INFANTILE HEMANGIOMA MASQUERADING AS PERSISTENT DIAPER DERMATITIS IN THE SETTING OF LUMBAR SYNDROME WITH A CONGENITAL CUTANEOUS SKELETAL MUSCLE HAMARTOMA

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Abbreviations **IH** = infantile hemangioma; **IH-MAG** = infantile hemangioma with minimal or arrested growth, **LUMBAR** = *L*ower body segmental IH, *U*rogenital anomalies, spinal cord *M*alformations, *B*ony defects, *A*norectal malformations/Arterial anomalies, *R*enal anomalies, **RMH** = rhabdomyomatous mesenchymal hamartoma.

Case report. A 3-month-old male with history of caudal appendage, tethered spinal cord and spinal lipoma complicated by neurogenic bladder and penoscrotal webbing was referred to dermatology for primary concern of persistent diaper rash. The referral noted discomfort in the diaper region as patient cried with each diaper change and symptoms were non-responsive to barrier protection and recent initiation of antifungals (Nystatin) for consideration of cutaneous candidiasis. Evaluation by pediatric dermatology revealed a large bright red thin plaque of approximately 20 cm in diameter composed of numerous coalescing bright red papules with similar adjacent multifocal papules extending from the lumbosacral region to the gluteal cleft, perineum, scrotum, and penis on a background pink telangiectatic patch showing coarse branching linear vessels on dermoscopy. Within the gluteal cleft, areas of ulceration and skin breakdown were noted within the vascular plaque. The vascular lesion did not extend to the lower extremities. The sacrococcygeal area further revealed the previously noted firm, exophytic appendage near the left superior gluteal cleft (Fig. 1, 2). The skin exam was consistent with a segmental infantile hemangiomas with minimal or arrested growth (IH-MAG) given the concomitant proliferative vascular papule / plaque component overlying a predominant larger area of prominent telangiectases.

Based on the clinical presentation, the patient met diagnostic criteria for LUMBAR syndrome – lower body segmental IH plus one additional LUMBAR feature – (6) with concomitant urogenital anomalies (penoscrotal webbing) and spinal cord malformations (tethered cord and spinal lipoma). At

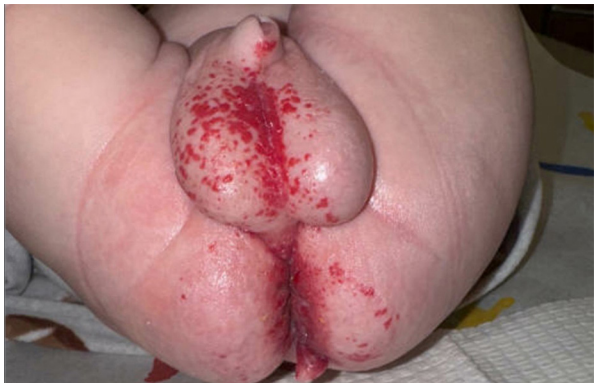

Fig. 1

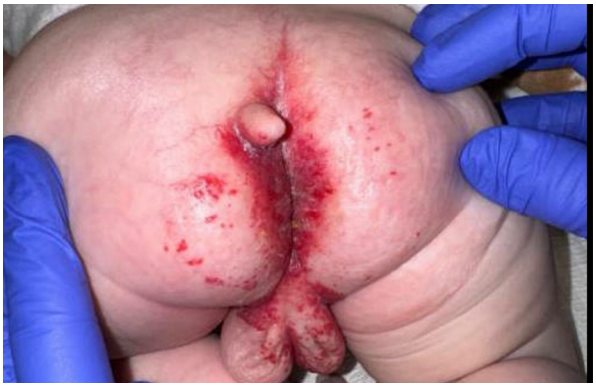

Fig. 2

Figs. 1, 2: Anogenital infantile hemangioma with caudal appendage in the setting of LUMBAR syndrome.

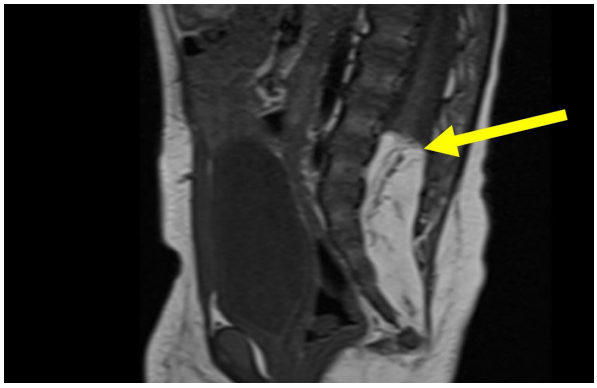

Fig. 3

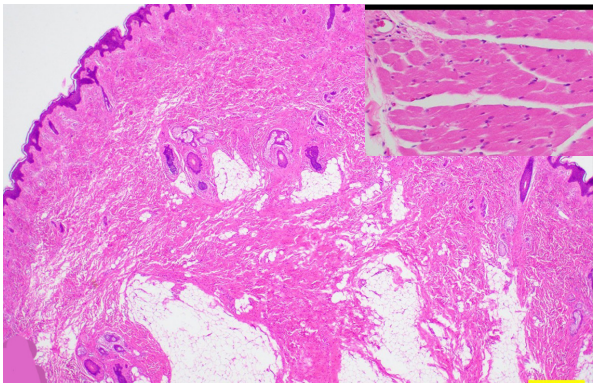

Fig. 4

Figs. 3, 4: MRI (Fig. 3) shows tethered spinal cord with intradural lipoma (yellow arrow). Histological examination of the skin appendage (Fig. 4, H&E, x10 and inset Fig. 4, H&E x40) reveals rhabdomyomatous mesenchymal hamartoma.

diagnosis, a renal ultrasound was performed that showed no evidence of associated renal anomalies. Pelvic and scrotal ultrasounds confirmed superficial vascularity at the site of the IH without evidence of a deep component. Following pediatric cardiology clearance, the patient was started on propranolol at 0.5 mg/kg/day with up-titration to 2 mg/kg/day for the treatment of the ulcerated IH. The patient exhibited progressive improvements in ulceration and eventually steady involution of the IH over time with propranolol tapered off thereafter.

Magnetic resonance imaging (MRI) of the lumbar spine showed a hypoplastic distal sacrum and coccygeal segments along with open sacral spinal canal and terminal lipoma measuring 5.9 x 1.3 cm at maximum axial diameter (Fig. 3). MR angiography of the lumbar spine and pelvic area did not reveal any arterial abnormalities. Further surgical intervention of the tethered cord would require concurrent subtotal lipoma resection by neurosurgery, thus was deferred for clinical monitoring with consideration if development of further neurologic deficit given the significant risks associated with the procedure.

The congenital caudal appendage was further considered a common cutaneous manifestation of LUMBAR syndrome (12), and was evaluated by plastic surgery for superficial excisional removal. Histopathology of the caudal appendage revealed a benign polyp with increased adipose tissue and several intermixed regions of mature skeletal muscle differentiation (Fig. 4) consistent with a congenital rhabdomyomatous mesenchymal hamartoma (RMH).

Discussion. The case presented further highlights that IH (often IH-MAG) is a crucial consideration in the differential of recalcitrant diaper dermatitis. This requires careful visual inspection and early referral to pediatric dermatology given the potential implications of multisystem involvement in the case of LUMBAR syndrome. IH-MAG is particularly more inconspicuous compared to other typical IH morphologies; IH-MAG has a predominant telangiectatic patch morphology and minimal proliferation which may benefit from close evaluation, including dermoscopy (13). Although the spinal malformations and urogenital complications were already identified prior to the diagnosis of LUMBAR syndrome in this case, the formal classification allowed for a unifying diagnosis and a more comprehensive investigation into features that may otherwise remain clinically unapparent, such as arterial or renal anomalies. Initiation of propranolol was also essential to manage the ulcerating complications of the IH-MAG and support involution to altogether reduce the risk of additional coinciding sequelae,

including superimposed infection, pain, and scarring (14). Thus, early identification of IHs is critical to guide monitoring, management, and further workup for associated clinical features as warranted.

In fact, recent literature has emphasized the association between the presence of complex cutaneous hamartomas, particularly those with skeletal muscle differentiation (i.e., RMH), and both PHACES (15) and LUMBAR (12) syndromes. Along with cutaneous RMH, cases of PHACES syndrome have further demonstrated the possibility of developing intracranial RMH (16). Other hamartomas have also been reported in PHACES syndrome including folliculosebaceous cystic subtypes (15), in comparison to LUMBAR syndrome where most have been identified as hamartomas with skeletal muscle components – predominately RMH – (6, 12, 17). Similarly to previous case reports, the histopathologically confirmed congenital RMH in our case of LUMBAR syndrome was located in the paramedian area rather than in the classic midline location (12). Additional investigation into the histopathologic features of cutaneous appendages in both PHACES and LUMBAR syndromes is warranted to determine the incidence of the specific hamartoma subtypes and their relationship with the additional comorbidities of these conditions. This may further broaden our understanding of the embryological origin and pathophysiologic mechanisms behind the various components of LUMBAR syndrome including segmental IH.

The corresponding clinical features of PHACES and LUMBAR syndromes raise the question of common pathologic mechanisms at opposite cranial and caudal embryologic poles, respectively. In addition to the shared findings of extracutaneous congenital anomalies, IHs, and hamartomas such as RMH, LUMBAR syndrome was also recently recognized to intermittently exhibit a ventral midline defect known as umbilical raphe. Interestingly, the umbilical raphe in LUMBAR syndrome is infraumbilical (18) and directly mirrors the supraumbilical raphe noted in PHACES syndrome (19). Such midline and paramedian defects have been attributed to aberrant mesenchymal cell migration (18), while the prevailing theory of IH development is via placental embolic origin based on comparable transcriptomic signatures (20) and placental antigen expression (21) – i.e., GLUT-1 (11) –. The mechanistic relationships between placental complications, the localization and extent of the associated IH, and the development of multisystem congenital anomalies in IH-related syndromes remain poorly understood. Further investigation may reveal additional in utero risk factors and screening tools for pre-clinical identification of complex IHs towards developing prevention and early intervention strategies.

Conclusion. The current work was presented to alert neonatologists and pediatricians to the need to consider a possible infantile hemangioma in cases of diaper rash resistant to therapy.

Conflicts of interest

The authors declare that they have no conflicts of interest.

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