LUMBAR: Association between Cutaneous Infantile Hemangiomas of the Lower Body and Regional Congenital Anomalies

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Objective To define the clinical spectrum of regional congenital anomalies associated with large cutaneous hemangiomas of the lower half of the body, clarify risk for underlying anomalies on the basis of hemangioma location, and provide imaging guidelines for evaluation.

Study design We conducted a multi-institutional, retrospective case analysis of 24 new patients and review of 29 published cases.

Results Hemangiomas in our series tended to be ''segmental'' and often ''minimal growth'' in morphology. Such lesions were often extensive, covering the entire leg. Extensive limb hemangiomas also showed potential for extracutaneous anomalies, including underlying arterial anomalies, limb underdevelopment, and ulceration. The cutaneous hemangioma and underlying anomalies demonstrated regional correlation. Myelopathies were the most common category of associated anomalies.

Conclusions We propose the acronym ''LUMBAR'' to describe the association of Lower body hemangioma and other cutaneous defects, Urogenital anomalies, Ulceration, Myelopathy, Bony deformities, Anorectal malformations, Arterial anomalies, and Renal anomalies. There are many similarities between LUMBAR and PHACE syndrome, which might be considered regional variations of the same. Although guidelines for imaging are suggested, prospective studies will lead to precise imaging recommendations and help determine true incidence, risk and long-term outcomes. *(J Pediatr 2010; ■: ■-■).*

Infantile hemangioma (IH) is the most common childhood tumor, with an estimated incidence of 4% to 5%.[1] The recognition that IH in certain locations on the skin can be associated with unique medical concerns, including the potential presence of underlying congenital anomalies, has been increasingly appreciated.[2] The IH morphologic subtype termed "segmental" (ie, large, plaque-like, involving a specific territory), although less common, is known to be at much higher risk for complications.[3]

The best known example of syndromic IH is PHACE (Online Mendelian Inheritance in Man database [OMIM] #606519), the association between large, characteristically segmental IH on the face and developmental defects of the cerebrovasculature, cardiovasculature, eyes, and chest wall. In comparison, the association between large IH of the lower half of the body and regional congenital anomalies is seemingly rarer and poorly understood.[4] Although 2 recent publications, both composed of small case series, proposed acronyms for this particular association,[5,6] each suffers from some limitations and neither has gained universal acceptance. Furthermore, despite these efforts, this distinct presentation has yet to be fully defined or recognized by OMIM.

The potential severity, complexity, and clinical implications of the anomalies associated with IH in the lumbosacral, perineal, or lower extremity location merits special attention. We thus present the largest series of 24 new cases and review 29 previously published cases to more fully define the clinical spectrum, clarify risk for underlying anomalies on the basis of specific IH location, and provide imaging recommendations. We also propose what we believe is a more inclusive and representative acronym for this syndrome: LUMBAR, for the association between Lower body IH and other skin defects, Urogenital anomalies and Ulceration, Myelopathy, Bony deformities, Anorectal malformations and Arterial anomalies, and Renal anomalies (**Figure 1**; available at www.jpeds.com).

IH	Infantile hemangioma
MG	Minimal growth
MRA	Magnetic resonance angiography
MRI	Magnetic resonance imaging
MRV	Magnetic resonance venography
OMIM	Online Mendelian Inheritance in Man database

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Methods

Institutional review board (IRB) approval was obtained for this study, allowing for the collection of non-identifiable patient information from outside institutions. Because of the known rarity of this association, a call for unpublished cases of lumbosacral or perineal IH associated with at least 1 other regionally present, congenital anomaly was made through the Society for Pediatric Dermatology membership listserve. Specific information requested included patient's sex, a description of the IH morphology and location, photograph(s) of the IH, presence or absence of ulceration, and a description of the associated anomalies identified, including results of evaluation(s) performed. Anomalies were categorized according to the LUMBAR acronym, with the term "myelopathy" used for disease/disorder of the spinal cord. Twenty-four cases were received from 8 institutions (7 in the United States, 1 in Chile). We also performed a PubMed search for publications that reported patient-specific data, which resulted in an additional 29 cases published through 2008.

To assess the potential risk for underlying anomalies on the basis of IH location, we classified the lumbosacral-perineal anatomy in 4 regions: (A) lumbar (T_{12}-L_5 intervertebral spaces); (B) sacral (area between the anterior iliac crests and perineum/genitals); (C) perineum and genitals (perineum: area between anus and urethral meatus); and (D) lower extremity (**Figure 2**; available at www.jpeds.com). The first and senior authors (I.I. and D.M.) independently reviewed all photographs, assigned regional designation(s) for each patient's IH location, and determined IH morphology as localized or segmental, minimal-growth (MG) or normal growth. Segmental was defined as IH showing linear or geographic patterning. MG was defined as segmental IH that maintained a telangiectatic or patchy appearance with minimal to no proliferation. This independent review yielded high inter-observer concordance. The few conflicting designations were discussed among the authorship and resolved.

Results

Table I (available at www.jpeds.com) and **Figures 3** and **4** describe the clinical features of 24 new, unpublished cases. There were 15 female and 9 male patients, resulting in a sex ratio of 1.7 to 1. The most common IH location was region B (sacral; 20/24; 83.3%), followed by regions A (lumbar; 18/24; 75%) and C (perineum/genitals; 16/24; 66.6%). Region D (lower extremity) was affected in 10 of 24 cases (41.7%). One case (case 1) had an additional retroperitoneal IH. Most patients (66%) had extensive IH with involvement of >1 region, most commonly all (A through D). IHs in all cases were segmental, with the exception of case 4. In 16 of 24 cases (67%), the IH was both segmental and MG, and in 9 of those patients the IH involved the entire lower

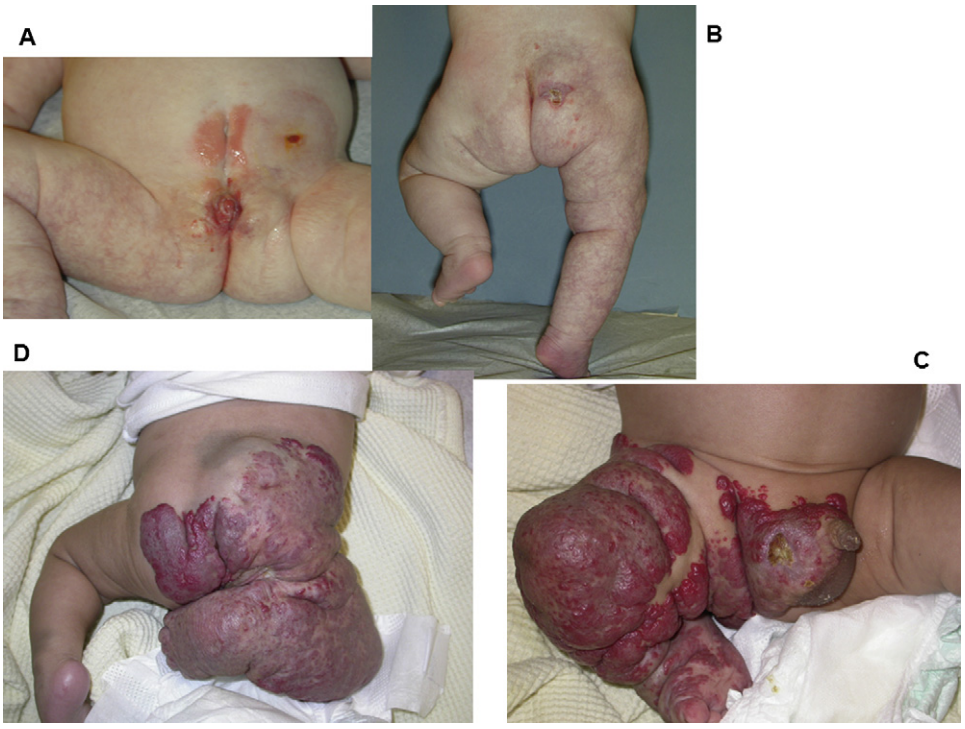

Figure 3. Clinical features of LUMBAR association. **A** and **B,** case 19; **C** and **D,** case 1. Both cases involve regions ABCD, but case 19 is segmental minimal-growth phenotype and case 1 segmental normal growth. Note the ulceration present in both cases, the lumbar lipoma and urogenital anomalies in case 19, and the pterygium in case 1 (**Table I**).

extremity. The IH extended over 3 or more regions in 73% of MG versus 33% of cases demonstrating normal growth. Only one patient (case 1) had an IH of the lower extremity that showed normal growth. Overall, ulceration was reported in 17 of 24 cases (70%). Ulceration was more likely when the IH was extensive (>87% with 3 or 4 regions involved and <50% with 1 or 2 regions involved) or MG (80% MG versus 55% normal growth).

Thirty percent of cases had only one reported extracutaneous anomaly, most commonly tethered cord. Only 25% of cases reported the presence of 3 or more extracutaneous anomalies in addition to the IH; 66% of these were MG. Twenty of 24 patients (83%) overall had magnetic resonance imaging (MRI) evidence of myelopathy, making this the most common category of extracutaneous anomalies. Twenty-nine percent of cases had a lumbosacral lipoma, 100% of which had underlying myelopathy. Four cases of myelopathy had no IH of region A, but 3 of these had a lumbosacral lipoma and the other had a lipomeningocele. All IH of region A associated with myelopathy involved and extended across the midline.

Twenty-nine percent of patients had anorectal malformations, most commonly imperforate anus. All cases of anorectal malformations had IH localized to region B (17.6% specificity). Twenty-five percent of cases had renal anomalies, most commonly single kidney. All cases of renal anomalies had IH localized to region B (28.5% specificity). Nineteen percent of patients had urogenital anomalies, all of whom had IH localized to region B (15% specificity). IH of region C yielded a 25% positive predictive value for urogenital anomalies and an 87.5% negative predictive value. Bony deformities of the lower extremities or pelvis were reported in two cases (8.3%). Two patients, both of whom had IH extending over the entirety of the right lower limb, had significant arterial anomalies of that limb. Less frequently reported anomalies are described in **Table I**. **Table II** (available at

Table III. Comparative incidence of LUMBAR anomalies in new and previously reported cases

Category of anomaly	Incidence (24 new cases), % (n)	Incidence (29 published cases), % (N)	Specific defects noted in new or published cases (% new cases/% published cases for the most frequent)
Lipoma and other cutaneous defects	45.8% (11/24)	51.7% (15/29)	•**Lipoma (29.1/27.6)** •Skin tag, caudal appendix, acrochordon •Tuft of hair, nevus •Sacral dimple
Urogenital	20.8% (5/24)	41.4% (12/29)	•Bladder (extrophy, elongated, problems) •Ureters (reflux, pyelo-ureteral duplication) •Clitoris (clitoromegaly, hemiclitoris) •Labia majora (incomplete, hypertrophied, asymmetric, atrophic) •Labia minora (absence) •Vagina (duplication, atresia) •Uterus (2 uterine cavities) •Testis (undescended, hydrocele, small remnant) •Penis (torqued, hypospadias, micropenis) •Scrotum (bifid, vulviform) •Ambiguous genitalia
Ulceration	70.8% (17/24)	37.9% (11/29)	
Myelopathy	83.3% (20/24)	68.96% (20/29)	•**Tethered cord (75/56.66)** •Lipomyelocele or Lipomyelomeningocele (20.8/23.33) •Syrinx •Spinal dysraphism •Abnormal termination of conus medullaris (high or low) •Abnormally thickened/fatty filum terminale
Bony	8.3% (2/24)	17.24% (5/29)	•**Foot deformity** •Leg discrepancy in length or diameter •Hip dysplasia •Sacrum abnormality •Scoliosis
Anorectal	29.1% (7/24)	37.93% (11/29)	•**Imperforate anus (20.8/13.33)** •Fistulas (recto-vestibular, recto-vaginal, recto-scrotal, rectal-fourchette, recto-perineal) •Anus (anterior displacement, stenosis, vestibular) •Complex cloacal anomaly •Deviated gluteal cleft
Arterial	8.3% (2/24)	13.79% (4/29)	•**Dysplasia, narrowing** •Aberrant course or origin •Persistence of embryonic anastomoses
Renal	25% (6/24)	31% (9/29)	•**Single kidney (8.3/16.66)** •Pelvic kidney •Pelviectasia, Pelvic diastasis •Nephromegaly or hydronephrosis •Hypoplastic kidney, Duplex left kidney
Other	33.3% (8/24)	20.70% (6/29)	•**Digestive (constipation, colostomy, megacolon)** •Omphalocele, Patent urachus •Atrophy of affected extremity

www.jpeds.com) summarizes the published cases that fit the inclusion criteria in LUMBAR, and Table III compares the incidence of LUMBAR anomalies in new and previously reported cases.

"Atypical" cases

There were two additional patients who, because of unusual clinical findings, were not included in our 24 patients, but we feel deserved mention. The first is a female infant with an ulcerated segmental IH involving regions B and C. She had no regional anomalies, but a bifid nose. The second is a female infant with a large segmental IH of the right supraorbit, forehead, and regions A, B, and C. Ulceration occurred only over her facial IH. An additional subglottic IH necessitated tracheostomy. MRI results of her lumbar spine were normal, but MRI of her thorax showed a 5- by 10-mm in diameter, 3- to 4-cm long, oval-shaped right paravertebral enhancing mass extending from the mid to lower thoracic spine, possibly representing IH. Also noted was a 15-mm triangular-shaped enhancing mass in the left adrenal region extending inferiorly to surround the abdominal aorta that was also suspected to be IH. With brain MRI, a supravermian lipoma with associated dysgenesis of the superior vermis was identified.

Discussion

The term "segmental" or what some authors refer to as "regional" describes IHs that show linear patterning, geographic patterning, or both over a specific cutaneous territory, respecting embryologic anatomic boundaries, rather than appearing to arise from a single focal point. Segmental IHs show reproducible patterns associated with developmental segments, which arise from the neuroectoderm and later correspond to a specific region of skin and soft tissues.[7] Clinically, segmental IH can manifest as a solitary, confluent plaque or small individual papules clustered in a patterned distribution. In the newborn period, segmental IHs may have a unique, "telangiectatic" appearance or present as a faintly erythematous patch, and thus they are often initially mistaken for capillary malformations,[8] also known as "port-wine stains." The presence of coarse telangiectasias is a feature more indicative of IH. In some instances, segmental IHs fail to proliferate and maintain this telangiectatic appearance, which has been described with various terms including "minimal growth (MG),"[9] "plaque-telangiectatic,"[3] and "reticular."[10]

When located on the face, a segmental IH is most commonly recognized as a potential indicator of PHACE syndrome. In particular, it has been suggested that segmental IH with MG morphology, when associated with PHACE, may possibly indicate more severe neurologic disease and portend a higher risk for ulceration.[11] Our data show that IHs associated with LUMBAR association are also most commonly, although not exclusively, segmental and in addition are often MG in morphology (Figure 3, A and B). A recent study of MG IH showed a strong tendency for such lesions to localize to the lower half of the body compared with the upper half.[9] MG IHs in our study were always segmental, showed a higher incidence of ulceration, were generally extensive, often involving the entirety of the lower limb, and had a higher incidence of extracutaneous anomalies. These extensive cases were more common among girls (3:1 ratio), in concordance with the findings observed by Mulliken.[10] However, the overall female predominance in LUMBAR (62.5%) appears to be lower than that observed in PHACE, which affects female patients in approximately 80% to 90% of cases.

Studies of PHACE syndrome have demonstrated a general regional correlation between the cutaneous IH and underlying anomalies present,[12] and our findings support a similar observation in LUMBAR. Myelopathies, notably tethered cord or lipomyelocele/lipo-myelomeningocele, were the most common category of anomalies seen in our patients and were strongly, although not exclusively, correlated with IH of region A. All region A IHs associated with myelopathy were segmental, involving and extending across the midline. Region B involvement with IH was highly sensitive (100%) for urogenital anomalies, anorectal malformations, and renal anomalies, but not specific. The reported incidence of urogenital anomalies may represent an underestimation, because anomalies of the internal genitalia (ie, ovaries, fallopian tubes and, prostate) may not have been specifically evaluated. Although we reviewed published cases for evidence of regional correlation, those assessments were often inconclusive because of either a lack of IH photographs or non-specific descriptions of IH location (eg, "pelvic localization"). Also, there exists a known regional correlation between cutaneous, generally segmental IHs and IHs affecting internal structures, either by direct extension or as a separate, underlying IH, as discovered in case 1 (Figure 4). Such findings parallel earlier observations in which IHs may be present either in direct contiguity or anatomic distinct, such as the association of "beard area" IH and airway IH,[13] IH of the ear and the scalp with IH affecting auditory structures,[14] the presence of cutaneous and underlying intraspinal IH,[15] and the presence of intracranial IH in patients with PHACE syndrome.[16]

An important association in LUMBAR that previously has not been emphasized is the potential for underlying arterial anomalies particularly when the IH extends over the entirety of a lower limb, as noted in 2 of our cases. Only a few cases of arterial anomalies have been described in this setting, some of which were found incidentally. Mulliken[10] reported 6 patients with reticular, segmental IH involving the lumbosacral area and lower extremity. Two had arterial anomalies: 1 with a hypoplastic iliofemoral artery and the other revealed dilatation of the right common iliac artery with extensive arteriovenous shunting in the pelvis and lower extremity, particularly the foot. Another case of an extensive, lumbosacral and limb IH reported abrupt tapering of the ipsilateral superficial femoral artery, with reconstitution from an

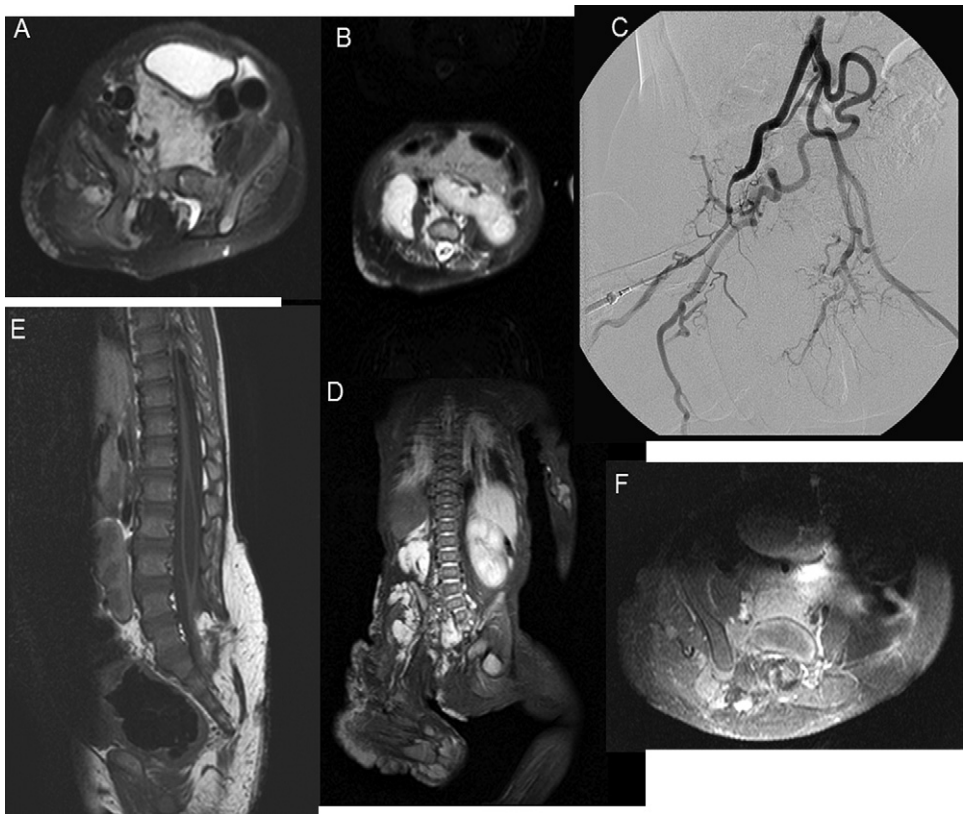

Figure 4. Representative images for case 1. **A, B,** and **D,** 4 months of age. **C, E,** and **F,** 10 months old, after treatment with corticosteroid and vincristine. **A,** Note the retroperitoneal hemangioma, displacing the urinary bladder, dysraphic sacrum, cutaneous and intramuscular hemangioma, neural tube defect with extension of fat and hemangioma into the sacral spinal canal. **B,** Left abdominal crossed fused renal ectopia and retroperitoneal hemangioma. **C,** Angiogram demonstrating the right primitive sciatic artery (aberrant right internal iliac and profunda femoral arteries) arising from the left common iliac artery (courtesy of Glen Siedel). **D,** Coronal view demonstrating the pterygium and hemangiomas in the retroperitoneum, gluteal muscles, thigh, and skin of the right lower extremity. **E** and **F,** Syrinx, tethered cord, cutaneous hemangioma, abnormal rectum, intra-pelvic hemangioma replaced by adipose tissue.

anomalous dominant artery. The affected limb in this case was also described as shorter and atrophic in comparison with the unaffected (Amy Nopper, personal communication). Atrophy was also noted in 1 of our cases, and the other case demonstrated grossly abnormal limb development, with an unusual "pterygium-like" structure that fused the right foot to buttock (Figure 3, D). Although previously it has been suggested that extremity IHs do not affect limb development, these findings suggest that atrophy can occur in this setting and may be related to underlying arterial anomalies of the affected limb. This feature may also help distinguish LUMBAR from other vascular syndromes that affect the limb, particularly vascular malformations that more often result in hypertrophy.[17]

These reports of arterial anomalies are not surprising because of their known frequency in PHACE syndrome, and they are probably more common than recognized because they have not been routinely and specifically evaluated. The few reports of vessel anomalies described in LUMBAR bear striking resemblance to those observed in PHACE: affecting large arteries, occurring ipsilateral and underlying the cutaneous IH, and describing various degrees of dysplasia, narrowing, aberrant course or origin, and persistence of embryonic anastomoses (Figure 4, C). Renal artery stenosis has been reported in one patient with PHACE syndrome, which was thought to be consistent with his other abnormal vasculature.[18] In PHACE patients with cerebrovascular anomalies, there is a known risk of progressive vasculopathy leading to stenotic and occlusive changes and a rare risk of ischemic stroke.[12] The mechanism(s) for stroke in PHACE are unknown, but the greatest risk is seemingly during infancy, with most occurrences reported between birth and 18 months of age. Although to our knowledge no clinical consequences from LUMBAR-associated vasculopathy have been reported, in addition to the aforementioned possible effects on limb development, one of our cases had significantly abnormal vasculature that coursed through an unusual pterygium-like structure. Despite maximal medical therapy and surgical attempts at revision, this child eventually required a below-the-knee amputation with partial release of his popliteal contracture to preserve some function. Finally, we

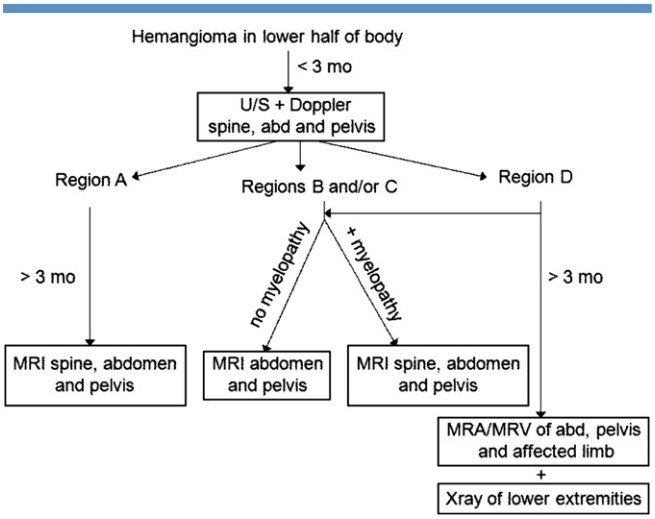

Figure 5. Algorithm for comprehensive diagnostic imaging guidelines of LUMBAR association.

are aware of two reports of complications caused by regional vasculopathies in association with extensive segmental IH of the upper limb. Heyer et al described a PHACE infant with a MG IH of the shoulder and arm with related occlusion of the small arteries of the hand and fingertip necrosis,[19] and Metry et al reported a similar patient with a shoulder IH and diminished ipsilateral radial and ulnar pulses.[20]

Like PHACE, the pathogenesis of LUMBAR is unknown, and we hope a comprehensive description of the clinical picture will provide insight into the possible etiologies. One of the main purposes of further defining an association is to provide guidelines for diagnosis and management. In children at risk for LUMBAR, the first and most important element in diagnosis is a thorough physical examination of the abdomen, pelvis, and lower extremities, with emphasis on the genital and paraspinal regions. Although this study supports a general regional association between the cutaneous IH and underlying anomalies, a limitation of its retrospective nature is the lack of consistent and thorough imaging for all potentially associated anomalies in all patients. It is therefore possible, for example, that a patient without any detectable cutaneous findings in region A, but a segmental IH in a different region, could still have spinal dysraphism. Our "atypical" cases also speak to the possibility of a more global than regional event in some patients. Until more is known, we suggest that all patients with segmental IH of any lumbosacral or perineal region <3 months of age undergo ultrasound scanning of the spine, abdomen, and pelvis with color Doppler. This should provide adequate screening for the presence of larger anomalies, internal IH, and spinal dysraphism without the risks of sedation. For infants >3 months old with segmental IH or lipoma involving and extending across the midline of region A, even when the results of neonatal imaging studies such as ultrasound scanning are normal, MRI of the spine at 3 to 6 months of age is recommended, because of the extremely high risk of myelopathy. When region A is not affected, MRI of the spine remains at the discretion of the provider. MRI of the abdomen and pelvis with intravenous contrast, plus magnetic resonance angiography (MRA)/magnetic resonance venography (MRV), will show the extent of internal IH, vascular anomalies, most malformations, and spinal dysraphism. When a time-resolved MRA technique is used, the MRV is obtained as part of the MRA and is recommended because although the anomalies tend to be arterial, a large IH can obstruct the ileofemoral veins. Computed tomography angiogram is also acceptable, but delivers a high radiation dose to the infant. When the lower limb is extensively involved with IH, patients should have MRI/MRA/MRV (preferably time resolved MRA) of the affected limb. Optimal imaging should include the abdominal aorta, iliacs, and femorals down to the pedal vessels. Bony abnormalities are usually detectable on physical examination, but we recommend a radiographic scanogram of the lower limbs in the early pre-school years to detect subtle leg discrepancy and that repeat imaging be considered at 6 years of age. An algorithm for these diagnostic imaging guidelines is depicted in Figure 5.

The diagnosis of LUMBAR, particularly when significant arterial anomalies are detected, may influence therapeutic decisions about treatment of the IH, when indicated. Systemic corticosteroids, beta-blockers, interferon, and vincristine are all therapeutic options for potentially life-threatening or life-altering IHs. Although the real risk is unknown, the presence of severe arterial anomalies of the limb or elsewhere may preclude or restrict the use of beta-blockers in this population, because potential hypotension and reduced perfusion through already stenosed or tortuous vessels could have potentially serious consequences in this important subset of patients. Although the diagnosis of LUMBAR association is not an absolute contraindication to the use of beta-blockers, we advocate caution and initiation of this therapy only after a complete and thorough investigation for significant internal vascular anomalies and appropriate subspecialty consultation.

There are many aforementioned similarities between LUMBAR and PHACE: association with segmental IH (often of minimal-growth morphology), female predominance, general regional occurrence of the cutaneous IH and underlying anomalies, and potential for underlying vascular (generally arterial) anomalies. LUMBAR might thus be considered analogous to PHACE, although affecting the lower half of the body. Like PHACE, it appears rare for patients with LUMBAR to manifest the complete spectrum of anomalies, although the incidence of anomalies reported in both new and previously described cases of LUMBAR likely represent minimum indices because most patients have not undergone complete evaluations for every potential category of anomalies. We suspect that LUMBAR is likely underrecognized because of the often subtle morphology of the MG IH, resulting in misdiagnosis as "reticulate capillary malformation" or, particularly when limb underdevelopment is

associated, cutis marmorata telangiectatica congenita. This study was intended to more fully define the spectrum of anomalies observed in LUMBAR, increase awareness, and provide imaging guidelines for diagnosis. Prospective studies are needed to determine true incidence, risk and long-term outcomes. ■

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Table I. Twenty-four new cases of patients who fit the inclusion criteria of LUMBAR association

Case, sex	Pattern/ region	Lower body hemangioma localization	Lipoma or other skin defect	Ulceration	Urogenital anomalies	Myelopathy	Bony anomalies	Anorectal anomalies	Arterial anomalies	Renal anomalies	Other anomalies
*1, M	NP/ ABCD	Buttocks, pelvis, right retroperitoneum to the level of the subhepatic space, entire right lower extremity	"Web" fusing right posterior heel and calf to right posterior thigh and buttock	Yes		Tethered cord associated with L5 and sacral dysraphism; - 2 mm syrinx at L2-L4 - lipomyelo-meningocele			abnormal and tortuous single tibial artery extending to the ankle, dominant arterial flow to the foot comes from an aberrant/ embryonic branch off the left iliac artery; aortic bifurcation extremely high at the level of L2-L3; renal arteries not demonstrated	Pelvic kidney with crossed fused renal ectopia from right to left	Venous drainage from the left leg is into the hemiazygos system on the left
2, F	NP/BC	Posterior introitus/vulva to gluteal cleft		No			Bilateral hip dysplasia	Recto-vestibular fistula. Imperforate anus			
3, M	MP/ABCD	Buttocks, left hemiscrotum, entire left lower extremity	Caudal appendage (finger-like), lipoma	Yes		Sacral dysraphism, asymmetric and deficient on the left, tethered cord, lower extremity paresis, lipomyelocele					
4, F	NP/A	Midline lumbosacral (5 cm, superficial)		Yes		Soft tissue band connecting hemanigoma to posterior elements of spine at S1 level and low-lying conus medullaris, possible tethered cord; mild delayed motor development and mild gait disturbance/ intoeing					
5, M	MP/BC	Lumbosacral and perineal	Lipoma extending from L4 to end of sacrum, small keratotic area resembling an epidermal nevus in the gluteal crease	Yes		Tethered cord; small spina bifida occulta defect at the S1 level; conus medullaris terminates at the L2-L3 level; abnormally thickened and fatty filum terminale					

(*continued*)

Table I. Continued

Case, sex	Pattern/ region	Lower body hemangioma localization	Lipoma or other skin defect	Ulceration	Urogenital anomalies	Myelopathy	Bony anomalies	Anorectal anomalies	Arterial anomalies	Renal anomalies	Other anomalies
6, F	MP/ABCD	Buttocks, perineum, entire left lower extremity			Clitoro-megaly made more apparent by diminutive surrounding labial tissue; small patent vaginal orifice with a relatively small perineal body			Complex cloacal anomaly with imperforate anus		Mild pelviectasias of left kidney	
7, F	MP/ABCD	Lumbosacral, entire left lower extremity		Yes		Tethered cord					
8, F	MP/ABC	Lumbosacral, gluteal cleft		Yes		Tethered cord: low-lying conus and thickened filum					
9, M	NP/A	Lumbosacral		No	Undescended right testicle and large left hydrocele; testicular remnant right scrotum; small left testes	Tethered cord				Mild bilateral nephromegaly	
10, M	MP/A	Lumbosacral	Lipoma, sacral dimple	No		Tethered cord L1-L3					
11, F	NP/ABC	Entire groin with extension to right inner buttock, circumferential around anus	"Fawn tail"	Yes	Reflux	Spinal dysraphism with tethered cord					Congeni-tal pulmonary HTN (term infant); normal ECHO
12, F	MP/B	Sacral, perianal	Lipoma	Yes		Tethered cord Syrinx L1 Conus		Deviated gluteal cleft			
13, F	MP/ABC	Lumbosacral	Lipoma, sacral dimple	Yes		Tethered cord Small syrinx T5 conus					
14, M	MP/A	Lumbosacral	Dermal sinus			Tethered cord Spina bifida occulta Syrinx					Motor delay

(*continued*)

Table I. Continued

Case, sex	Pattern/ region	Lower body hemangioma localization	Lipoma or other skin defect	Ulceration	Urogenital anomalies	Myelopathy	Bony anomalies	Anorectal anomalies	Arterial anomalies	Renal anomalies	Other anomalies
15, F	MP/ABCD	Perianal, perineal, Left leg		Yes				Imperforate anus, recto-vaginal fistula		Left bifid ureter with hydronephrosis	
16, M	NP/B	Lumbosacral, perianal		No		Fatty filum, tethered cord, low-lying L3 conus		Anterior displaced anus			Constipation
17, F	MP/ABCD	Perianal, lumbosacral, labia		Yes		Tethered cord					
18, F	MP/ABC	Lumbosacral		Yes		Tethered cord					Colostomy
19, F*	MP/ABCD	Pelvic, right lower extremity	Lipoma	Yes	Vaginal atresia	Tethered spinal cord, low lying conus with juxtamedullary lipoma at the L2-3 level		Imperforate anus, rectovaginal fistula	i. anomalies of the right iliac artery; ii. superficial femoral artery is not continuous. Persistent right sciatic artery which probably comes off the hypogastric artery.	Single pelvic kidney, two ureters, a small atrophic ureter draining the right side and ending in a right-sided ureterocele, pelvic diastasis	i. omphalocele ii. end sigmoid colostomy iii. right leg more atrophic than left
20, F	MP/BCD	Left leg, vulva, buttock		Yes						Solitary left kidney	Venous insufficiency
21, F	MP/ABCD	Right buttock, leg, lumbosacral		Yes	Right ureteral reflux, recurrent UTI	Tethered cord, lipomyelo-meningocele	Leg length discrepancy right longer than left, scoliosis	Imperforate anus, cloacal anomaly			Colostomy, pull-through, constipation
22, M	NP/B	Lumbosacral	Lipoma	Yes		Tethered cord					
23, M	NP/B	Buttocks, sacral	Acrochordon right buttock	No		Tethered cord, Lipomeningocele					Constipation, enco-presis
24, F	NP/ABC	Buttocks, perianal, lumbosacral		Yes		Closed complex terminal myelocele with terminal lipoma					

Regions involved: A, B, C, D, or any combination.
*Additional information is available in Figures 3 and/or 4.
MP, Minimal proliferative; *NP,* normal proliferative; *M,* male; *F,* female; *HTN,* hypertension; *UTI,* urinary tract infection.

Table II. Published cases of patients who fit the inclusion criteria in the LUMBAR association

Case, Sex	Lower Body Hemangioma Location	Lipoma or other cutaneous defects	Ulceration	Urogenital anomalies	Myelopathy	Bony deformities	Anorectal anomalies	Arterial anomalies	Renal anomalies	Other anomalies	Source
1, F	External genitalia, perineum, lower extremity			Bladder extrophy, undifferentiated and asymmetric external genitalia			Anterior anus		Right pyelo-ureteral duplication, Left renal agenesis	megacolon	Gonzalez Martin 1982[21]
2, M	Right buttock, macular, bluish areas	Skin tag		Scrotum bifid, torqued penis	Tethered cord Lipomyelmeningocele	Foot deformity (calcaneo-valgus deformity of right leg and equinovarus deformity of the left leg)	Imperforate anus, rectoscrotal fistula		Right renal agenesis		Goldberg, 1986[4]
3, F	Vulva, left buttock, mass, necrotic	Skin tag	Yes	Incomplete labia majora	Tethered cord lipomyelomeningocele	Foot deformity (cavus deformity of left foot and discrepancy in size)			Pelvic left kidney with reflux		Goldberg, 1986[4]
4, F	Sacrum, left leg and foot, macular, teleangiectatic	Skin tag					Imperforate anus, rectal-fourchette fistula				Goldberg, 1986[4]
5, F	Lower extremity, sacrum, telangiectatic	Skin tag				Bony abnormalities (sacrum)	Imperforate anus, rectovaginal fistula		Hypoplastic left kidney		Goldberg, 1986[4]
6, M	Sacrum, gluteal cleft, buttock, mass, telangiectatic				Lipomyelomeningocele	Foot deformity					Goldberg, 1986[4]
7, F	Buttocks, lower back, bladder, left vaginal wall, external genitalia	Lipoma L4	Yes	Hypertrophied left labium major, sexual ambiguity	Tethered cord		Vestibular anus			Stomal prolapse	Bouchard, 1999[22]
8, F	Perineum, pelvic, buttocks, left thigh, vulva, telangiectatic		Yes	Elongated bladder, ureteral reflux, left hemiclitoris, absence of labia minora, labia majora atrophic	L3-L5 spina bifida		Anterior anus	Abnormal vessels: retroperitoneal, presacral, pararectal			Bouchard, 1999[22]
9, F	Perivulvar, telangiectatic	Skin tag		Hypertrophied left labium major			Anal atresia, rectovestibular fistula		Left renal agenesis		Pelaez Mata, 2001[23]
10, M	Scrotum, perineum, buttocks, sacrum, posterior thighs, bladder, rectum, macular, telangiectatic		Yes	Torqued micropenis, vulviform scrotum, incomplete migration of left testis, hypospadias						Constipation	Girard, 2006[5]

(*continued*)

Table II. Continued

Case, Sex	Lower Body Hemangioma Location	Lipoma or other cutaneous defects	Ulceration	Urogenital anomalies	Myelopathy	Bony deformities	Anorectal anomalies	Arterial anomalies	Renal anomalies	Other anomalies	Source
11, F	Sacrum, left buttock, vulva, left thigh, macular, telangiectatic			Bilateral ureteral reflux, 2 uterine cavities, solitary perineal orifice			Anal atresia		Bilateral kidney dilatation with left ureterohydro-nephrosis		Girard, 2006[5]
12, F	Extensive midline superficial, vulva, gluteal and perineal		Yes				Anal atresia, rectoperineal fistula				Stockman, 2007[6]
13, M	Lumbosacral, midline, superficial	Caudal appendix			Tethered cord, lipomyelomeningocele						Stockman, 2007[6]
14, M	Lumbosacral, midline, superficial	Caudal appendix			Tethered cord, lipomyelomeningocele						Stockman, 2007[6]
15, M	Lumbosacral, gluteal, perineal, extensive, midline, superficial	Subcutaneous lipoma		Hypospadias	Tethered cord, lipomyelomeningocele						Stockman, 2007[6]
16, M	Left buttock, nodular	Subcutaneous lipoma			Lipomyelomeningocele						Stockman, 2007[6]
17, F	Left leg, buttock, genitalia, pre and post sacral, adjacent to the uterus on the left, suprapubic, adjacent to the right iliacus muscle, left ankle, telangiectatic plaque	Lipoma L4-L5	Yes	Vesicouretheral reflux gr II-III	Tethered cord, thick filum	Atrophy of left leg, left leg shorter than right		Left superficial femoral artery abruptly tapered, with reconstitution from an anomalous dominant artery	Absent left kidney, right kidney has dilatation of renal pelvis	Patent urachus	Amy Nopper, personal communication
18-24	Lumbar hemangiomas	4/7 intraspinal lipomas			7/7 Tethered cords, 2/7 tight filum terminale						Albright, 1989[24]
25, F	Left lower extremity, buttock and perineum		Yes		Tethered cord		Recto-vaginal fistula, anal atresia	Hypoplastic iliofemoral artery	Solitary kidney	Omphalocele	Mulliken, 2007[10]
26, F	Left lower extremity, buttock and perineum		Yes	Vaginal and uterine duplication			Recto-vaginal fistula, imperforate anus		Duplex left kidney		Mulliken, 2007[10]

(*continued*)

Table II. Continued

Case, Sex	Lower Body Hemangioma Location	Lipoma or other cutaneous defects	Ulceration	Urogenital anomalies	Myelopathy	Bony deformities	Anorectal anomalies	Arterial anomalies	Renal anomalies	Other anomalies	Source
27	Left lower extremity, buttock and perineum, hepatic hemangiomas		Yes		Tethered cord						Mulliken, 2007[10]
28	Right lower extremity, buttock and perineum, hepatic hemangiomas		Yes		Tethered cord						Mulliken, 2007[10]
29, M	Right lower extremity, hepatic, face, left lower extremity		Yes	Ambiguous genitalia				Dramatic dilatation of the right common iliac artery with extensive arteriovenous shunting in the pelvis and the lower extremity, particularly in the foot		Cardiac failure Pathologic femoral fractures Colostomy due to deep anal and sigmoidal ulcerations Hepatic hemangiomas High -thigh amputation	Mulliken, 2007[10]

One of the patients in cases 27 and 28 is male, the other is female. Authors did not specify.

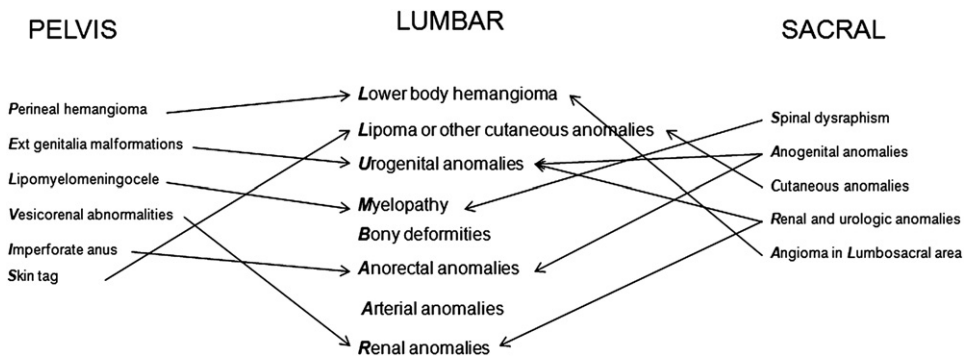

Figure 1. The LUMBAR acronym includes findings described with both the PELVIS and SACRAL acronym.

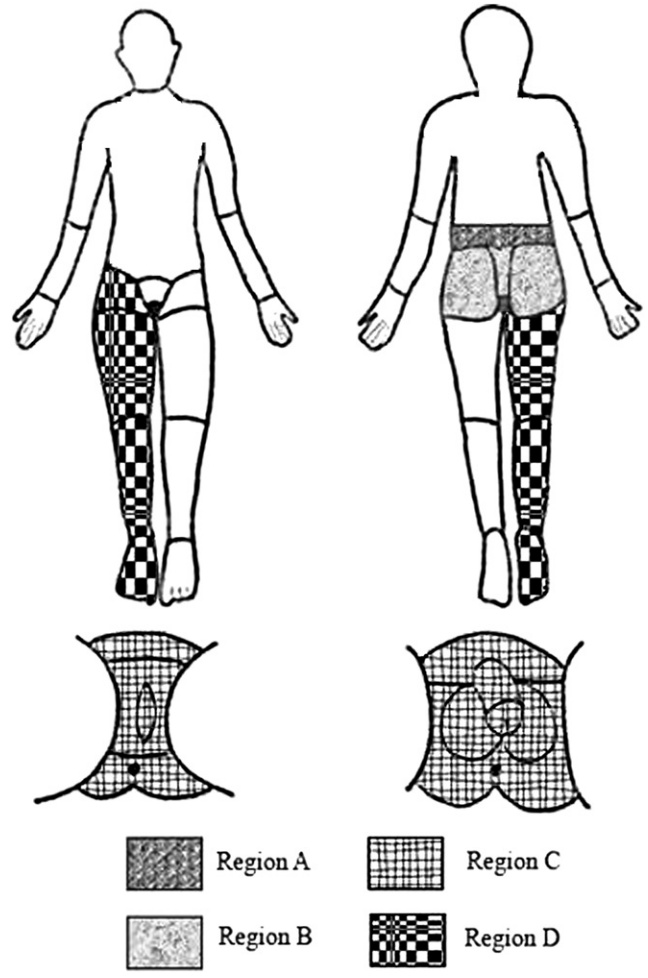

Figure 2. LUMBAR regions.