ARE THERE OTHERS

Yes! As rare of a syndrome as this is, there is a group of parents who are connecting with each other and raising awareness of LUMBAR syndrome for our children. We want every patient with LUMBAR to get the necessary care he or she needs

We are pioneering this syndrome! The children, families and parents of children with this syndrome are coming together

LUMBAR/PELVIS SYNDROME COMMUNITY

As more information about LUMBAR syndrome becomes available, please visit our page on the PHACE Syndrome Community website at :

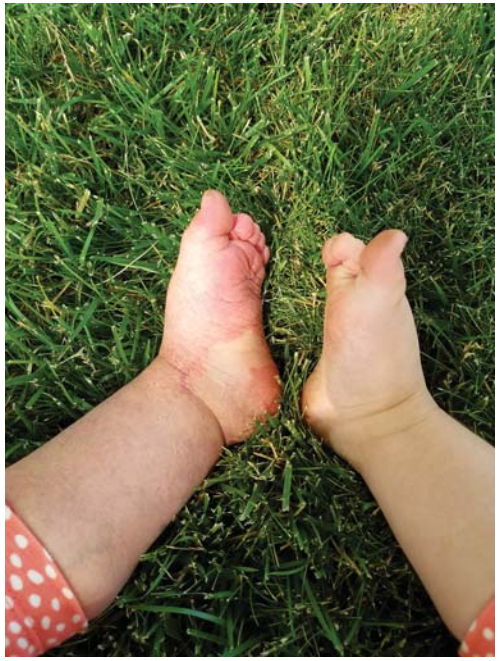
www.phacesyndromecommunity.org

References

1 https://www.dermnetnz.org/topics/pelvis-and-lumbar-syndromes

HELP OUR RESEARCH EFFORTS

If you or your child has been diagnosed with LUMBAR, one of the most important things you can do is join the LUMBAR and PHACE syndrome registry: http:// www.phaceregistry.com



We know this is difficult and yes we are pioneering this syndrome. Just know you are not alone.

There is great strength in numbers!

LUMBAR SYNDROME

Some Facts and Support

LUMBAR or PELVIS

PELVIS and SACRAL are both acronyms that have been used to describe a collection of characteristics which formed similar syndromes, with PELVIS being the more recently acknowledged acronym.

P: Perineal hemangioma[1]
E: External genital malformations
L: Lipomyelomeningocele
V: Vesicorenal anomalies
I : Imperforate anus
S: Skin Tag

Since the first use of the acronym PELVIS in 2006 by Dr. Celine Girard and colleagues[1] to correlate these conditions, there have been new discoveries that have doctors using a different acronym to incorporate all these possibilities; LUMBAR. This new name accurately describes the current understanding of all the complexities this single syndrome can hold. This is important because it shows that we now believe these are all part of one syndrome, where each person has a unique combination.

L: Lower Segmented Hemangioma[1]
U: Urogenital Defects
M: Myelopathy of the spinal cord
B: Boney Deformities
A: Arterial and Anorectal defects
R: Renal anomalies

DIAGNOSIS

The most prominent feature of LUMBAR is a large infantile hemangioma on the lower back often extending down to the leg. An infantile hemangioma is a special type of vascular birthmark that can grow rapidly. Infantile hemangiomas in children with LUMBAR may ulcerate and can be very painful. Early evaluation and treatment will help to stop the growth of the infantile hemangioma, heal ulcerations, and prevent complications. Infantile hemangiomas can be difficult to diagnose, since they appear more bruise-like and subtle at birth. Within a few weeks they can grow and cause ulcers that are difficult to care for. One of the most important care teams for someone diagnosed with LUMBAR is a dermatologist. With their guidance towards the correct medicine regimen, the infantile hemangioma tissue can be managed during its aggressive growth period.

Sometimes this birthmark is the first part of the puzzle to be recognized, but in other babies, it may be another characteristic such as a displaced anus, single kidney or bony defect, such as a shorter limb. Radiology tests and imaging, such as ultrasounds or MRIs might be needed to help make the diagnosis.

Once the hemangioma and one or more of the additional complications are identified, the diagnosis of LUMBAR may be made.

CHARACTERISTICS

Additional characteristics that can be associated with LUMBAR are:

-Bladder extrophy
-Spina Bifida
-Shorter limbs
-Displaced anus
-Single Kidney

Not every LUMBAR patient will have all of the characteristics listed. The severity of each characteristic associated with LUMBAR can vary from mild to severe.

Every LUMBAR case is unique, getting the appropriate care teams in place as soon as possible is key.

Other care teams, in addition to Dermatology, that could be involved in the care of someone with LUMBAR would be:

-Urology
-Nephrology
-Neurology
-Neurosurgery
-Orthopedics