Case Report
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PELVIS Syndrome: A Case Report

Iva Palčić[1], Andrea Cvitković Roić[1,2], Alemka Jaklin Kekez[1], Goran Roić[3]

[1]Helena Clinic for Pediatric Diseases, Zagreb, Croatia, [2]Josip Juraj Strossmayer University of Osijek, Faculty of Medicine, [3]Zagreb Children´s Hospital, Zagreb, Croatia

Correspondence:
iva.palcic@yahoo.com
Tel.: 385 1 231 00 67
Fax.: 385 1 231 019

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Objective – PELVIS syndrome is an association between large perineal hemangiomas and congenital anomalies, including anorectal, urinary tract, spine, and external genitalia malformations. We present a 7-year-old boy with PELVIS syndrome. **Case report –**. The boy had been monitored since early intrauterine age by ultrasound (US) due to left kidney cysts. Clinical examination at birth found large segmental hemangiomas in the perineal, gluteal region, and the left leg. At the age of 4 weeks US showed a multicystic, dysplastic left kidney with compensatory hypertrophy of the right kidney. Voiding cystography (VCUG) did not show vesicoureteral reflux and morphology of the bladder, and the urethra was normal. PELVIS syndrome was suspected and the child underwent further diagnostic tests which showed: undescended left testis with a hypoplastic left scrotum and micropenis, right-sided angulation of the sacrum and closed spinal dysraphism from S1 to S5, with lipoma of the filum terminale, without tethering. The urodynamics investigation was normal. During the seven-year follow-up there was spontaneous regression of the left kidney cysts; the growth and function of the right kidney was normal, as was also sphincter control. There was significant spontaneous regression of the hemangiomas. Follow-up magnetic resonance imaging (MRI) of the lumbosacral spine showed no progression of the lipoma. **Conclusion –** Large perineal hemangiomas should be recognized as indicators of underlying congenital anomalies, including urogenital, anorectal and spinal. Therefore, additional diagnostic evaluation, including kidney ultrasound and spine imaging, are necessary.

Introduction

PELVIS syndrome is an association between segmental infantile hemangiomas of the perineum, and congenital anomalies of the genitalia, urinary tract, spine, anus and rectum. The acronym PELVIS was proposed in 2006 by Celine Girard and colleagues (1). It stands for: perineal hemangioma, external genitalia malformations, lipomyelomeningocele, vesicorenal abnormalities, imperforate anus, and skin tag. Currently, there are no standardized diagnostic criteria for diagnosis of PELVIS syndrome. However, not all the components of the acronym are necessary to fit the malformation sequence (2, 3). Large perineal hemangiomas should be recognized as indicators of underlying pathological conditions, so patients with segmental hemangiomas over the perineum and/or lower back should be carefully screened for other possible congenital anomalies by appropriate imaging techniques (4).

Here we report a 7-year-old boy with PELVIS syndrome.

Case Report

The boy was monitored from early intrauterine age (16 weeks) by ultrasound (US) due to left kidney cysts. The right kidney, the amount of amniotic fluid and fetus development were completely normal. He was born as a full-term neonate by normal spontaneous vaginal delivery. His birth weight was 3130 g and birth length 48 centimeters. He was the third child of nonconsanguineous parents. There was no family history of birth defects. On the first clinical examination large segmental hemangiomas in the perineal, gluteal region and left leg were detected (Fig. 1). Further, we found an undescended left testis with a hypoplastic left scrotum and micropenis (Fig. 2)

Ultrasound showed a multicystic dysplastic left kidney with compensatory hypertrophy of the right kidney. Voiding cystography did not show vesicoureteral reflux, and the morphology of the bladder and urethra was normal. As it is known that large perineal hemangiomas may be associated with congenital anomalies, and the boy had already been diagnosed as having a multicystic dysplastic kidney, PELVIS syndrome was suspected, and the child underwent further diagnostic tests. MRI of the spine revealed right-sided angulation of the sacrum, spina bifida from S1 to S5, an osseous defect covered with fibrous and subcutaneous fat tissue, and confirmed multicystic dysplastic left kidney, with compensatory hypertrophy of the right kidney (Fig. 3). The urodynamics investigation was entirely normal.

MRI also showed lipoma of the filum terminale, but without signs of a tethered cord (Fig. 4).

During the seven-year follow-up there was spontaneous regression of the left kidney cysts; the growth and function of the right kidney was normal, as well as sphincter control. Orchidopexy was done at the age of 2.

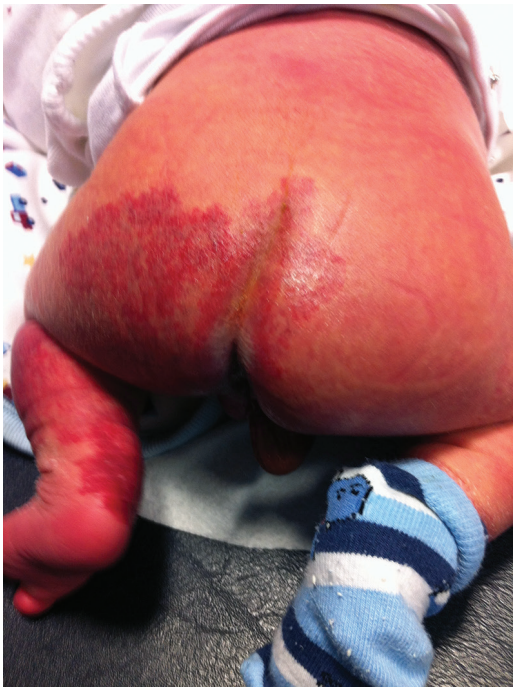

Fig. 1. *Large Segmental Hemangiomas in the Perineal, Gluteal Region and Left Leg.*

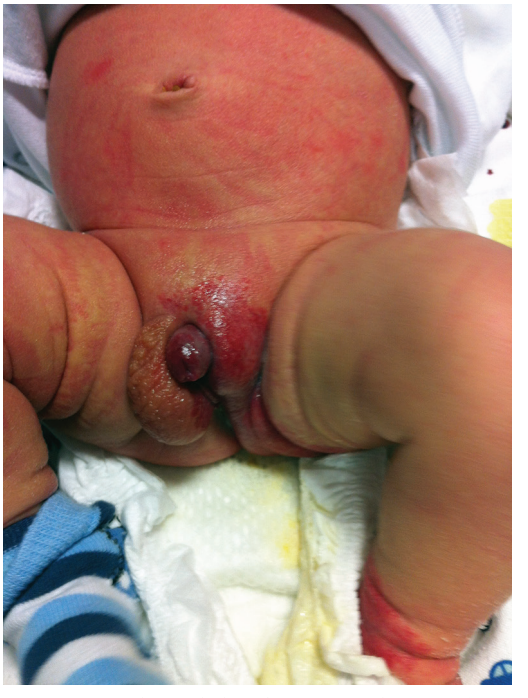

Fig. 2. *Undescended Left Testis with Hypoplastic Left Scrotum and Micropenis.*

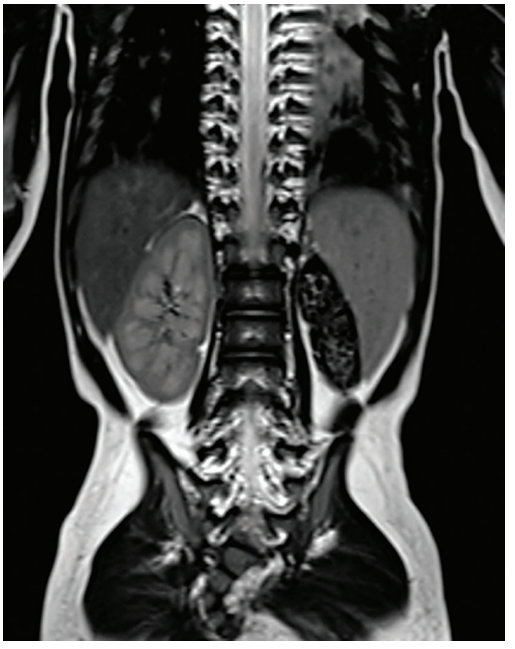

Fig. 3. *MRI of the Spine: Right-Sided Angulation of the Sacrum, Spina Bifida From S1 to S5, Osseous Defect, Covered with Fibrous and Subcutaneous Fat Tissues. We Can Also See the Multicystic Dysplastic Left Kidney, with Compensatory Hypertrophy of the Right Kidney.*

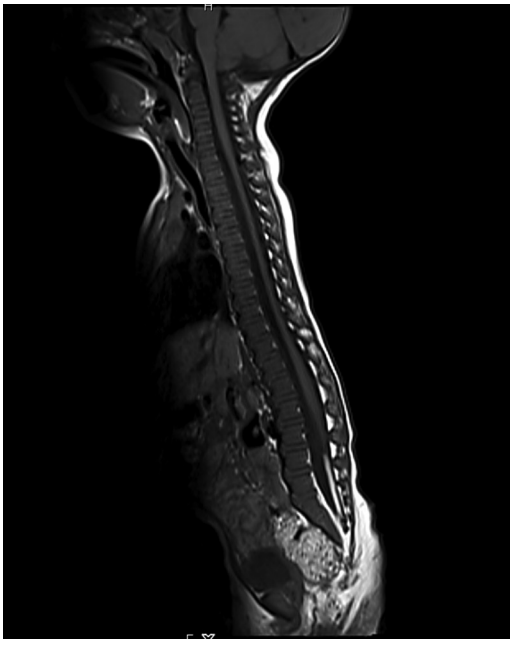

Fig. 4. *MRI of the Spine: Lipoma of the Filum Terminale, no Signs of Tethered Cord.*

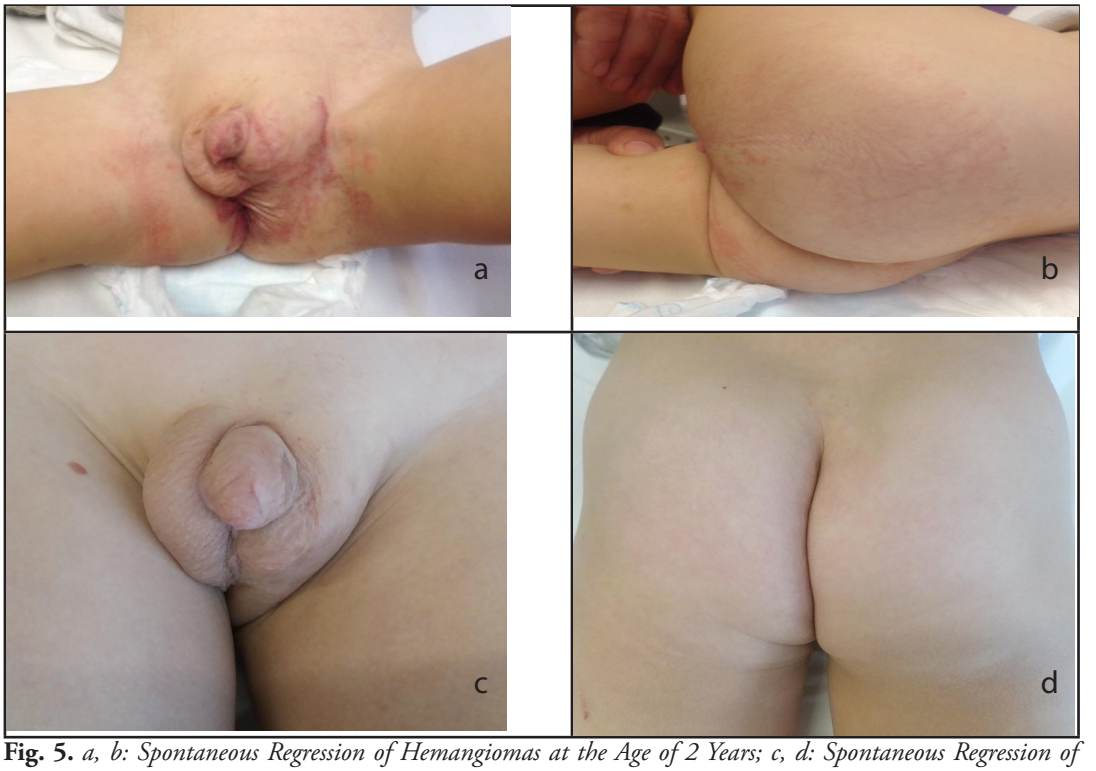

Fig. 5. *a, b: Spontaneous Regression of Hemangiomas at the Age of 2 Years; c, d: Spontaneous Regression of Hemangiomas at the Age of 7 Years.*

The parents refused treatment of the hemangiomas with corticosteroids or propranolol, but significant spontaneous regression occurred over time (Fig. 5, a, b, c, d). Follow up MRI of the lumbosacral spine every 2 years showed no progression of the lipoma.

Discussion

Hemangiomas are the most common benign tumors of infancy, occurring in up to 10% of children by 1 year of age, with a striking predilection for the head and neck region. Approximately 10% of hemangiomas are located in the perineal area. Perineal hemangiomas are more prone to ulceration because of irritation from stools, urine, friction, and maceration (5). In rare instances, perineal hemangiomas may be associated with congenital anomalies, including anorectal, urinary tract, spine, and external genitalia malformations. Gonzalez Martin et al. initially described this type of association in an 8-month-old girl in 1982 (6). In 1986, Goldberg et al. published the cases of five infants with sacral hemangiomas associated with a variety of congenital malformations involving the anorectal area, urinary tract, spine, and external genitalia (7).

Two further cases of infants with large sacral and perineal hemangiomas and visceral malformations were published by Girad and al. who proposed the acronym PELVIS syndrome (perineal hemangioma, external genitalia malformations, lipomyelomeningocele, vesicorenal abnormalities, imperforate anus, and skin tag) to denote the characteristic findings of this syndrome (1). The initial clinical presentation of infants with PELVIS syndrome is usually the finding of a large sacral or/and perineal hemangiomas after birth, as it was in our patient. This localization of hemangiomas should alert the physician to possible underlying abnormalities, so appropriate imaging techniques need to be used (1, 2, 4).

Our boy had a multicystic dysplastic kidney, undescended left testis with hypoplastic left scrotum and micropenis, right-sided angulation of the sacrum, spina bifida occulta from S1 to S5, and lipoma of the filum terminale without tethering. The association of anorectal, urinary tract, spinal and genital defects were grouped under the term "urorectal septum malformation sequence, partial or complete" (8, 9). These abnormalities are thought to arise early, during the fourth to sixth weeks of development from the insufficient breakdown of the cloacal membrane and a lack of mesodermal cells in the caudal region of the embryo.

Conclusion

We report a case of a boy with PELVIS syndrome. Through this case we wish to point out that large perineal hemangiomas should be recognized as indicators of possible underlying congenital anomalies, including urogenital, anorectal and spinal. Therefore, additional diagnostic evaluation is essential, especially ultrasound of the urinary tract and spinal imaging.

Authors' Contributions: Conception and design: IP and AJK; Acquisition, analysis and interpretation of data: IP, ACR, and AJK; Drafting the article: IP; Revising the article critically for intellectual content: ACR, GR, and AJK; Approved final version of the manuscript: IP, and GR.

Conflict of Interest: The authors declare that they have no conflict of interest.

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