

Redness and Painful Ulcerations in the Perineal Area

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A 7-week-old boy with ambiguous genitalia presented for evaluation of what the parents described as progressively worsening diaper rash. The patient was born at full-term after an uncomplicated gestation via normal spontaneous vaginal delivery. Examination of the external genitalia revealed microphallus with phimosis and a bifid scrotum. Two weeks after birth, the patient developed redness and painful ulcerations in the diaper area. At the time of presentation, the patient had bright red

plaques along the suprapubic lines, inguinal creases, and in the perineal region. Physical examination also was notable for tender ulcerations of the inguinal creases and perineum and a perineal skin tag.

What's the diagnosis?

- infantile psoriasis
- kaposiform hemangioendothelioma
- PELVIS syndrome
- PHACE syndrome
- Sturge-Weber syndrome

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The Diagnosis: PELVIS Syndrome

Infantile hemangiomas (IHs) are present in up to 10% of infants by 1 year of age and are most commonly located on the face and upper extremities. Less than 10% of IHs develop in the perineum.¹ Perineal IHs are benign tumors of the vascular endothelium that present as plaques and commonly are accompanied by painful ulcerations. Ulceration is more common in the diaper area secondary to irritation from urine, stool, and friction.² Although most IHs are benign isolated findings, facial IHs have been associated with several syndromes including Sturge-Weber and PHACE (posterior fossa brain malformations, hemangiomas, arterial anomalies, cardiac anomalies and coarctation of the aorta, and eye and endocrine abnormalities) syndromes.³ Researchers also have identified an association between lumbosacral IHs and spinal dysraphism (tethered spinal cord).⁴

A smaller number of studies have investigated congenital anomalies related to perineal IH,^{1,5} specifically PELVIS syndrome. The acronym PELVIS has been used to describe a syndrome of congenital malformations including perineal hemangioma, external genital malformations, lipomyelomeningocele, vesicorenal abnormalities, imperforate anus, and skin tag.¹ An alternative description of similar findings is LUMBAR (lower body hemangioma and other cutaneous defects; urogenital anomalies, ulceration; myelopathy; bony deformities; anorectal malformations, arterial anomalies; and renal anomalies).⁵ Researchers have suggested that both of these acronyms describe the same syndrome, and it is common for the syndrome to be incomplete.⁶ One study (N=11) found that perineal hemangiomas are most commonly associated with anal malformations (8 patients), followed by urinary tract abnormalities (7 patients) and malformation of the external genitalia (7 patients). A skin tag was present in 5 patients.¹ The pathogenesis of PELVIS syndrome is unknown.

When an infant presents with a perineal hemangioma and physical examination suggests PELVIS syndrome, imaging should be performed to evaluate for other anomalies. Before 4 months of age, ultrasound should be utilized to investigate the presence of reno-genitourinary or spinal malformations. Magnetic resonance imaging is the preferred imaging modality in children older than 4 months.⁷ Management of PELVIS syndrome requires a multidisciplinary approach and early recognition of the full extent of congenital malformations. Pediatric dermatologists, urologists, endocrinologists, and neonatologists have a role in its diagnosis and treatment.

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