Pediatric pedunculated perianal problems

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Abstract

Congenital perianal masses, distinct from sacrococcygeal teratomas, are a rare entity and are minimally reported in literature. A retrospective chart review was conducted of all patients who presented to the Children’s Hospital of Wisconsin (CHW) with a benign pedunculated perianal mass between April 2014 and June 2015. Three patients fulfilling these criteria underwent elective resection either under general or local anesthesia. The first patient’s mass was diagnosed as a benign acrochordon, or skin tag. The midline location of this mass prompted a spinal MRI revealing a low-lying conus and fibrolipoma of the filum terminale. The patient underwent prophylactic laminectomy and transection of the filum terminale and he currently remains asymptomatic. The second patient was found to have a hamartomatous polyp and later developed scrotal and perirectal vascular malformations and was treated successfully with topical timolol. Our third patient presented with a hamartomatous lesion and underwent a colonoscopy and stool studies, both of which were negative. We present these cases to bring awareness to this clinically important entity and to the associated anomalies and work-up that should be considered.

While the exact incidence is currently unreported, perianal masses typically occur in older patients and are quite rare in the neonatal population. When congenital, they are often associated with other medical disorders. For example, more common congenital mass lesions, such as sacrococcygeal teratomas, giant congenital melanocytic nevi, Langerhans cell histiocytosis, rectal duplication cysts, and rhabdomyosarcomas, rarely present as a clinically isolated mass. These lesions have been well documented [1–11]; however, benign congenital perianal masses and their important clinical associations are underrepresented in the literature. The aim of this series, therefore, is to bring this exceedingly rare diagnosis into awareness, to alert clinicians of associated conditions, and to present an effective diagnostic and treatment plan.

1. Methods

Institutional review board (IRB) approval was received to perform a retrospective chart review of all patients born with a perianal mass who were treated at the Children’s Hospital of Wisconsin (CHW) from 4/1/2014 to 6/30/2015. Patients with sacrococcygeal teratomas were excluded, as those masses are well described in the literature and distinct from the entity we sought to examine. For each patient, the following information was collected from the electronic medical record: age, gender, weight, drug history, findings on physical examination, operative and pathology reports, radiological and laboratory investigations and complications.

2. Results

Three patients met the above mentioned criteria and their charts were reviewed in detail.

2.1. Case 1

This patient was diagnosed with a perianal mass by prenatal ultrasound at 24 weeks gestational age. The mass remained stable throughout the duration of the pregnancy. On initial physical examination, the mass was found to be a 1.5 x 1.0 x 1.0 cm skin tag with a broad base protruding from the anal canal at the 5:00 position. There were no other abnormalities noted on visual inspection and a 10 hegar was passed through the anal canal with ease (Fig. 1a). The patient was feeding and passing stool without difficulty, and thus operative resection was postponed until
50 weeks post-conceptual age to minimize anesthesia risk and the need for inpatient monitoring. Histologic examination of the resected specimen revealed a polypoid lesion with an orthokeratotic epidermis and underlying dermis composed of dense, bland connective tissue, multiple small blood vessels, and scattered nerve bundles. The lesion was diagnosed as a benign acrochordon, or skin tag (Fig. 1b).

Because of the midline location of the mass, there was concern for anatomic abnormalities of the lower spine, and this patient thus underwent further work-up with a lumbar spine MRI. The MRI documented a low-lying conus and linear fibrolipoma of the filum terminale. The patient continued to be asymptomatic without any extremity weakness or bowel or bladder dysfunction. Patient underwent a laminectomy with transection of the filum terminale as a

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**Fig. 1.** a: The patient from case one underwent excision at 50 weeks of age. b: Representative photomicrograph of excised specimen from Case 1, with histologic features of benign acrochordon (H&E, 2×).

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**Fig. 2.** a: The patient from case two underwent surgical excision on day two of life. b: Focus of lesion from Case 2 with separation of epidermis from underlying dermis (H&E, 2×). c: Lobules of mature adipocytes from Case 2 (H&E, 4×). d: Intermediate magnification photomicrograph from Case 2 displaying extensive hemorrhage between and within lobules of adipocytes (H&E, 10×). e: Patient from case two with a medial right scrotal vascular malformation that was diagnosed at one and a half months of age. f: Patient from case two with a left buttocks vascular malformation that was diagnosed at one and a half months of age.
prophylactic measure to avoid tethering of the spinal cord. The patient tolerated both operations well without post-operative complications, and remains asymptomatic.

2.2. Case 2

The patient's perianal mass was discovered by visual inspection of the anal area on day one of life. Examination revealed a pedunculated and thrombosed appearing mass on a narrow stalk emanating from the anus at the 7:00 position (Fig. 2a). The patient was feeding well and passing adequate meconium. He underwent surgical resection in the operating room on day two of life. Microscopic examination revealed a polypoid tissue fragment focally covered by histologically unremarkable epidermis, while other areas displayed attenuation and separation of the epidermis from the remainder of the tissue fragment. Centrally, the specimen consisted of lobules of mature adipose tissue separated by extensive hemorrhagic fibrovascular tissue, and a diagnosis of hamartomatous polyp with ischemic/hemorrhagic changes was rendered (Fig. 2b–d).

Several weeks later, the patient was found to have visible scrotal and perirectal vascular malformations on physical examination. The patient underwent a flexible sigmoidoscopy that was negative. These vascular malformations are currently being treated with topical timolol ointment and are improving (Fig. 2e–f).

2.3. Case 3

The patient's perianal mass was discovered by initial physical examination of the anal area on day one of life. The patient had spent the first seven days of life at an outside hospital neonatal intensive care unit (NICU) as she was born with meconium aspiration, requiring a week of supplemental oxygen. The mass remained stable in size throughout the NICU stay. On physical examination, the mass was 2.3 x 1.5 x 1.0 cm, was emanating from the rectal canal, and had torsed and become necrotic (Fig. 3a). The patient was feeding and passing stool without difficulty. The mass was excised as an outpatient using local anesthetic. Similar to the previous case, this lesion displayed fibrovascular septa and lobules of mature adipocytes with overlying histologically unremarkable skin; this lesion, however, lacked hemorrhagic changes (Fig. 3b). The final diagnosis was that of a hamartomatous polyp.

3. Discussion

Each of our three patients was born with a benign pedunculated perianal mass adjacent to their anal canal. The first was consistent with an acrochordon, or skin tag, which because of its location in the posterior midline prompted evaluation for a tethered cord. The second patient was diagnosed with an ischemic hemorrhagic hamartomatous polyp, and went on to develop perineal and perianal vascular malformations. In light of the discovery of these vascular malformations, we reviewed the perianal polyp pathology and confirmed the diagnosis of a hamartomatous polyp, rather than a vascular tumor or malformation. The third patient was diagnosed with a soft tissue hamartomatous lesion.

The masses in our reported patients did not fit the pathologic description of any other congenital perianal masses described in the literature. The most common cause of a perianal mass described in a newborn is a sacrococcygeal teratoma [1–4]. However, the differential diagnosis is wide for perianal masses in the neonatal population and the presentations are quite variable; additional and less common diagnostic considerations include masses associated with giant congenital melanocytic nevi, Langerhans cell histiocytosis, rectal duplication cysts, and rhabdomyosarcomas, among others.

Sacrococcygeal teratomas are often diagnosed on prenatal ultrasound and visible as a perianal mass on physical examination at birth. They differ from our population of patients in that sacrococcygeal teratomas develop at the base of the coccyx but do not emanate from the anal canal [1–4]. Congenital melanocytic nevi can be located perianally similar to our patients; however, they also commonly occur in other areas of the body, such as the back, abdomen, or chest, and are clearly visible on external inspection. The masses we report were polypoid and covered with non-pigmented epidermis, while congenital melanocytic nevi are typically flat and pigmented [5]. Langerhans cell histiocytoses are a proliferation of dendritic cells that often show cutaneous involvement, but unlike the patients presented often have simultaneous manifestations of disease in various other organ systems [6,7,10]. Rectal duplication cysts typically adhere to some part of the gastrointestinal tract and present in childhood as an infection, a fistula, or with mass effect on the bowel [8,11]. In contrast, the lesions we describe did not communicate with the gastrointestinal tract and were asymptomatic. Rhabdomyosarcomas are the most common childhood soft tissue sarcoma and usually occur in the head and neck region, but can occur perianally [9]; however, the masses we describe in our series were clearly histologically benign.

To our knowledge, neonatal perianal masses, exclusive of sacrococcygeal teratomas, have rarely been reported. Hamartomatous polyps and skin tags should be included in the differential diagnosis for patients presenting with these lesions. As long as the patient is not exhibiting obstructive symptoms, excision can be safely completed electively. Further work-up may be required depending on the histologic diagnosis, location of the lesion, and the presence of any clinical symptoms. For example, lesions located midline should prompt a neurologic work-up to rule out an underlying spinal pathology. In the presence of a hamartomatous polyp, a

Fig. 3. a: The patient from case three underwent surgical excision on day eight of life. b: Photomicrograph of Case 3 displaying features diagnostic of a hamartomatous polyp (H&E, 2×).
sigmoidoscopy, colonoscopy, or stool study is encouraged to rule out associated bowel pathology. Perineal vascular malformations have been previously associated with perianal masses as part of the PELVIS syndrome (perineal hemangioma, external genitalia malformations, lipomyelomeningocele, vesicorenal abnormalities, imperforate anus, and skin tag), a very rare diagnosis with only a handful of reported cases [12–14]. Thus, in the presence of both perineal vascular malformations and perianal polyps, further neurologic, gastrointestinal and urologic work-up is warranted. By reporting our experience with three cases of perianal masses, we hope to bring this uncommon diagnosis into awareness as well as highlight their association with other conditions.

References