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Case Report Rapport de cas

A perineal cystic hamartoma causing constipation in an intact female Irish water spaniel

Alejandro Alvarez-Sanchez, Lauren Charnock, Paul Hanna, James Dundas

Abstract — A perineal fluid-filled structure was discovered in a 6-year-old intact female Irish water spaniel suffering from intermittent constipation. Diagnostic tests revealed the structure was immediately caudal to the vagina and compatible with a cyst. Surgical excision was required for resolution of clinical signs. Histology confirmed the structure was a cyst. The exact origin is unknown; however, the variety of lining epithelia, including sections with mucin production, and a well-differentiated smooth muscle layer, were most consistent with development from the lower hindgut or urogenital sinus during embryonic growth. The histologic and anatomical similarities with human retrorectal cystic hamartomas were key in establishing the diagnosis of a perineal cystic hamartoma. Following removal, constipation resolved, and the cyst did not recur.

Résumé — Hamartome kystique périnéal causant de la constipation chez une femelle intacte de race épagneul d'eau irlandais. Une structure périnéale remplie de liquide fut découverte chez une chienne intacte de race épagneul d'eau irlandais âgée de 6 ans souffrant de constipation intermittente. Les tests diagnostiques ont révélé que la structure était immédiatement caudale au vagin et était compatible avec un kyste. L'excision chirurgicale était requise pour la résolution des signes cliniques. L'histologie confirma que la structure était un kyste. L'origine exacte est inconnue; toutefois, la diversité de l'épithélium de couverture, incluant des sections avec production de mucine, et une couche bien différenciée de muscle lisse, étaient plus cohérentes avec un développement à partir du tractus digestif postérieur ou du sinus urogénital lors de la croissance embryonnaire. Les similarités histologique et anatomique avec l'hamartome rétro-rectal humain étaient critiques pour établir le diagnostic d'hamartome kystique périnéal. À la suite du retrait, la constipation s'est résolue, et il n'y a pas eu de récurrence du kyste.

(Traduit par Dr Serge Messier)

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During embryonic growth, the gastrointestinal tract divides into the foregut, midgut, and hindgut (1). Abnormal development of the hindgut can give rise to malformations affecting the transverse colon, descending colon, rectum, and the embryonic cloaca. Well-documented malformations that may affect the lower intestinal tract in humans are developmental cysts, such as cystic hamartomas and duplication cysts. In humans, these cysts are frequently located in the retrorectal region (2). Of these cysts, retrorectal cystic hamartomas share similar histologic characteristics to the perineal cystic hamartoma of the present report, also believed to be a type of developmental cyst (3,4). Rare reports of developmental cysts are described in the veterinary literature (5–7). Although most of these cysts are asymptomatic, over time they may cause clinical signs due to their mass effect. The purpose of this study was to document the clinical manifestation, diagnostic steps, histologic characteristics, and treatment of a perineal developmental cyst, thereby expanding the veterinary knowledge base on this atypical malformation, as well as the management and treatment options.

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Case description

A 6-year-old intact female Irish water spaniel dog, weighing 28.6 kg, was presented to the Atlantic Veterinary College (AVC) Small Animal Surgery Service in December 2017 for a mass in the perineum. The condition had been noted on routine physical examination 2 mo earlier by the AVC Small Animal Internal Medicine Service. The dog had not been previously bred and had no significant medical concerns other than difficult defecation associated with what the owner described as "ribbon-like" stools. The perineal mass had been noted by the owner previously; however, its initial time of appearance was undetermined. The owner believed the mass increased in size with the patient's heat cycles. On physical examination, no significant findings

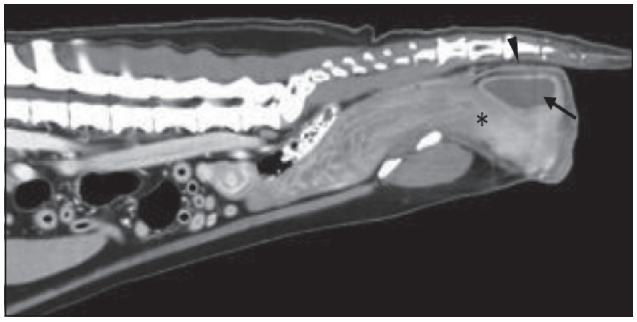

Figure 1. Post-contrast lateral computed tomography (CT) scan. The well-circumscribed cyst (arrow) can be seen immediately dorsal to the vagina (asterisk) and vulva, and ventral to the anus and rectum (arrowhead). Note the dorsal deviation of the ventral wall of the rectum and anus.

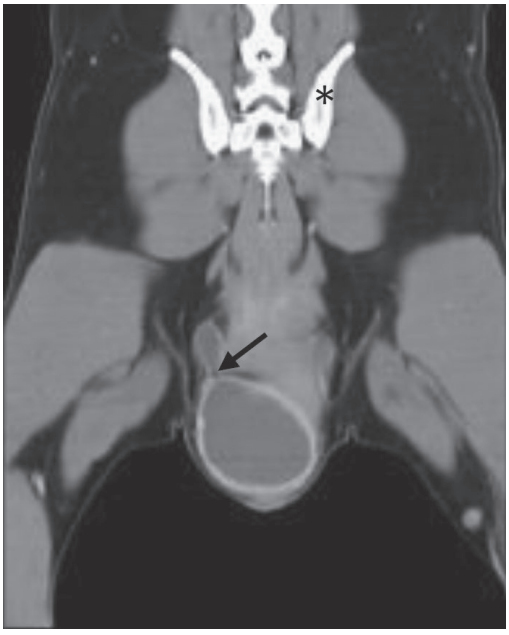

Figure 2. Post-contrast coronal computed tomography (CT) scan. The ilial wings have been left as references (asterisk). Note the proximity between the cyst and the right anal gland (arrow).

were identified aside from the mass. Palpation of the mass revealed that it was non-painful, fluctuant, measuring approximately 5 cm × 6 cm, and the overlying perineal skin was haired. The well-circumscribed, but deeply fixed structure could be felt within the subcutaneous tissues, immediately superficial to the perineal body. On rectal examination, dorsal deviation of the ventral rectal wall could be detected immediately above the location of the structure. Vaginal examination was unremarkable.

A Board-certified radiologist performed an ultrasonographic assessment of the perineal structure and abdomen when the patient was seen by the Internal Medicine Service, revealing that the mass was well-circumscribed, contained echogenic fluid, and was surrounded by a thick hyperechoic rim. The mass did not appear to be associated with any other anatomic structures despite being situated immediately caudal to the caudal vaginal wall. Ultrasound-guided aspiration of the structure resulted in drainage of 95 mL of light brown fluid that was submitted for cytologic examination. Cytology revealed proteinaceous fluid with mild neutrophilic inflammation. Based on cytological and imaging findings, a cyst was suspected.

The patient returned to the Small Animal Surgery Service in January 2018 for additional tests in preparation for surgical removal of the presumed cyst and ovariohysterectomy. At this visit, the patient was in estrus. Aside from the perineal structure and concurrent estrus, physical examination was normal; rectal and vaginal examination remained unchanged. Complete blood (cell) count (CBC) was unremarkable, and a biochemical profile showed a mild hyperbilirubinemia [5 μmol/L, reference range (RR): 0 to 4 μmol/L] and a mild hypophosphatemia (0.68 mmol/L, RR: 0.84 to 1.83 mmol/L). Contrast-enhanced computed tomography (CT) scan (Aquilion TSX-101A; Toshiba Medical Systems, Shimoishigami, Japan) of the abdomen and pelvis was performed under general anesthesia. Noted abnormalities included a right ovarian cyst, vaginal, vulvar, and uterine hypertrophy, and mild fluid accumulation in the uterus — these findings were attributed to the concurrent estrus. The perineal structure was cavitated, well-encapsulated, and appeared to be within 5 mm of the right anal gland and vaginal wall (Figures 1 and 2).

Fine-needle aspirates of the perineal structure were repeated due to its suspected association with the right anal gland as seen on CT. After the aspirates were taken, the structure was fully drained, emptying 45 mL of light brown fluid. Based on the new cytology results, a cyst remained our primary differential diagnosis. Aspirates of the right anal gland were obtained and were consistent with anal sac secretory material, confirming communication between both structures was unlikely. Surgical removal of the presumed cyst and ovariohysterectomy were delayed at this point as the patient was in estrus.

Routine ovariohysterectomy was performed 1 mo later. At this time, the presumed cyst had subjectively decreased in size and could not be easily palpated; therefore, removal was not attempted. The ovaries, cervix, and uterus were submitted for histopathology — no gross or microscopic abnormalities were detected. Despite the ovariohysterectomy, the structure did not regress, and the patient was presented to the surgery service in April 2018 for recurrence of the presumed cyst, as well as difficult defecation. The structure was palpable (~4 cm diameter) externally through the perineal skin, as well as on rectal palpation. Surgical excision was recommended at this time.

For surgical removal, an incision was created immediately below the anus, extending ventrally to the vulva. The structure was bluntly dissected from the surrounding tissues — fibrous and vascular attachments were noted where the structure abutted the caudal vaginal vault. The proliferation was spherical, ~5 cm in diameter, encapsulated, and with a relatively white and smooth exterior surface. The perineal structure, which was intact upon removal, was placed in 10% formalin for histopathological analysis. No communications were noted between the presumed cyst and other nearby anatomic structures. Routine surgical closure of the site was performed, and the patient recovered from surgery and anesthesia uneventfully.

At trimming, the cyst-like structure was unilocular, roughly 4 cm diameter, filled with brown watery fluid and had a

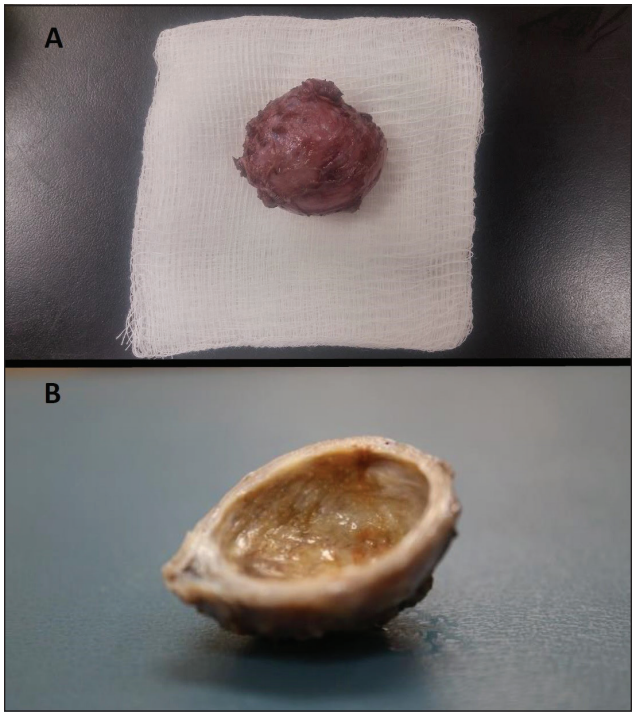

Figure 3. Perineal cystic hamartoma. On removal, the cyst was well-circumscribed, firm, and roughly 4 cm in diameter (A). The hemisectioned cyst had a large lumen with a relatively smooth inner surface (B).

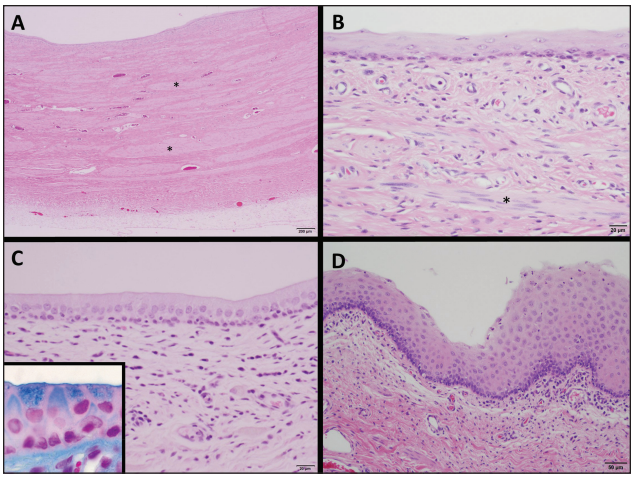

Figure 4. Perineal cystic hamartoma. The relatively thick cyst wall has numerous circumferential bundles of smooth muscle (asterisks) embedded in a dense fibrous stroma (A, B). The cyst is lined by a variety of epithelia, including various forms of stratified squamous (B, D), and stratified (bilayered) columnar epithelia (C). Hematoxylin and eosin (H&E) stain. Inset (C): High-magnification with Alcian blue (pH 2.5) staining showing mucin production in the apical cytoplasm of some epithelial cells.

relatively smooth inner lining (Figure 3). On histopathological examination, the inner aspect of the wall was lined by various types of epithelium including stratified squamous and stratified columnar to pseudostratified columnar with some of the columnar cells showing apical mucin production (Figure 4). The presence of an inner epithelial lining confirmed the structure was a cyst. Subjacent to the epithelium was a variably thick layer of propria-like connective tissue which showed a patchy interstitial infiltrate of generally small numbers of macrophages (many containing yellow-brown cytoplasmic pigment), lymphocytes, and plasma cells. Beneath this, the thickest layer of the wall was composed of multiple circumferential thin bands of well-differentiated smooth muscle within a relatively dense fibrovascular stroma. Loose connective tissue was present along the outer margins of the wall. These findings were consistent with a developmental cyst, with histologic features similar to those described for human retrorectal cystic hamartomas ("tailgut cysts"), but in a perineal location (2,4,8).

Two weeks after surgery, the surgical site had healed without any complications. On follow-up 4 mo after removal of the cyst, there had been no recurrence. Stools had also returned to normal consistency, and constipation had not been observed since surgery.

Discussion

The case reported herein describes a rare type of perineal developmental cyst, and the associated clinical signs, tests, and histologic findings. In humans, developmental cysts in the retrorectal region are believed to arise from incomplete tailgut regression during embryonic growth, and histologically present an epithelial layer which frequently contains smooth muscle beneath it (3). The presence of epithelium, with or without subjacent smooth muscle, indicates that these cysts are developmental. The perineal cyst was also classified as developmental given the similar wall layering. However, incomplete tailgut regression alone does not explain the anatomic location of this cyst. During development, the epithelium of the lower intestinal tract and part of the urinary tract (the urethra and most of the urinary bladder lining) derive from the hindgut and the urogenital sinus. The hindgut and urogenital sinus develop from the endoderm, whereas the smooth muscle subjacent to the epithelia and non-epithelial portion of the urinary bladder develop from the mesoderm (1). Considering the mixed endodermal-mesodermal origin of portions of the lower intestinal and urinary tracts, the origin of this perineal cyst is compatible with either urogenital sinus or hindgut.

The diagnosis of a cyst was established on histologic analysis upon identification of the epithelial lining within the fluid-filled structure (9). The epithelium had some areas of mucin production. Mucinous vacuolation occurs within goblet cells, which are commonly located throughout the intestines up to the colorectal mucosa (10). Goblet cells are absent caudal to the colorectal mucosa, suggesting that the origin of the present cyst is unlikely related to the anal canal, an ectodermal derivative.

Tailgut cysts (retrorectal cystic hamartomas) are an example of a hindgut developmental cyst well-documented in the human literature, and they are histologically very similar to the cyst reported herein. Tailgut cysts can be multilocular or unilocular and are lined by various epithelial types including squamous, transitional, columnar, ciliated columnar, or mucinous epithelium. These structures are classified as hamartomas because they present a well-differentiated cell population, the epithelial lining, and proliferate from a normal organ in development, the

hindgut (9,11). Tailgut cysts can also contain various amounts of organized to disorganized bands of concentrically arranged smooth muscle beneath the epithelium and they always lack neural plexuses (2,3,8). The histologic similarities this cyst shares with human retrorectal cystic hamartomas aided in establishing the diagnosis of a perineal cystic hamartoma.

Other types of developmental cysts deriving from the hindgut reported in the human literature include epidermoid cysts, dermoid cyst, teratomas, and duplication cysts. Epidermoid cysts are unilocular cysts, lined by squamous epithelium without any skin adnexa (9,12). Smooth muscle fibers are absent within the cystic wall. Dermoid cysts present skin adnexa, such as hair follicles and sweat glands, underneath the squamous epithelium (2,9,12). Teratomas possess all 3 germ layers, none of which are native to the area in which they proliferate, and generally refer to cases with dermal appendages, neural elements, cartilage, or bone (9,12). Duplication cysts are unilocular and can be cystic or tubular in conformation. They are lined by gastrointestinal and respiratory epithelium commonly infiltrated with villi or crypts, and have a muscle layer divided into muscularis propria and muscularis mucosa and connected by a neural plexus (12).

Due to the initial suspicion that the cyst was linked to the reproductive tract, aberrant cystic proliferations of the vagina that could be responsible for dyschezia or tenesmus had also been considered (13,14). In human medicine, the most common vaginal cysts are Müllerian cysts and Gartner duct cysts, both typically located along the vaginal wall (13,15,16), and Bartholin's duct cysts, which arise from the Bartholin glands within the vulva (16). Müllerian cysts are characterized by tall columnar mucinous epithelium (16), contrasting with Gartner duct cysts which are lined by a single layer of cuboidal to low columnar epithelium (15,17). Bartholin's duct cysts can be lined by 3 types of epithelium: mucus secreting, transitional, and squamous. The extra-vaginal location of the perineal cyst in the present case, combined with a different type of epithelium (varying from pseudostratified columnar to squamous), suggests that the cyst had not derived from the reproductive tract.

Some developmental cysts, such as dermoid cysts, are common findings in veterinary patients (9,11,18). However, developmental cysts of similar histologic characteristics to the cyst of the present report have been less frequently documented in the veterinary literature (5,7). Dunkel et al (5) documented a foal with a rectal cyst with comparable histologic features to human retrorectal cystic hamartomas and juvenile polyps. Another author reported a dog diagnosed with an endodermal cyst in the cranial abdomen. The diagnosis was based on the histological resemblance between the cyst's epithelial lining and the patient's small intestine (7). The histologic features and anatomic location were fundamental in establishing a diagnosis in both studies.

In human medicine, ultrasonography, CT, and magnetic resonance imaging (MRI), are used to aid in identification of similar cystic lesions (15,17). However, definitive diagnosis requires histopathology. Computed tomography scan proved instrumental for surgical planning in the dog herein, by ruling out involvement of the vaginal or rectal vault, or the presence of other lesions.

Surgical removal of cysts associated with the gastrointestinal and urogenital tract have been reported in the veterinary literature (5,7,13) and the prognosis after removal is considered good, as *en bloc* resection is generally curative (4,7,8,19). However, it is challenging to standardize when surgical intervention is necessary as the cyst may go undetected until it is large enough to compress surrounding structures, as was the case of the patient in this report and in the patient reported by Cooper (7). In human medicine, symptoms associated with developmental cysts include chronic lower back or perineal pain, urinary retention, dysuria, rectal bleeding, rectal prolapse, or change in stool calibre (2,4,8). Four months following surgical removal of the perineal cyst, our patient's constipation had ceased, and stool consistency normalized. Similar symptoms, such as the change in stool calibre of our patient, may help in the identification of developmental cysts in proximity to the colon, rectum, or anus in animals.

If left untreated, developmental cysts can become a nidus of infection, fistulize, or undergo malignant transformation to adenocarcinomas, carcinoids, or sarcomas (2,3,12,19). In humans, there is an estimated 2% to 13% incidence of malignant transformation of tailgut cysts (3,19) — no such malignant transformation was found in the few cases of developmental cysts reported in the veterinary literature.

The case reported in this study describes a perineal cystic hamartoma that was responsible for signs of constipation, and the steps taken for its diagnosis and treatment. While developmental cysts are a rarely reported finding in domestic animals, they should be included in the differential diagnosis list of cysts appearing in the perineal region. To establish an accurate diagnosis, histologic examination is required, and surgical excision is necessary for complete resolution of clinical signs.

Acknowledgment

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Book Review
Compte rendu de livre

A Color Handbook: Skin Diseases of the Dog and Cat, 3rd edition

Heinrich N, Eisenschenk M, Harvey R, Nuttall T. CRC Press, Boca Raton, Florida, USA. 2019. 296 pp. ISBN: 9781-1383-0870-1.

The approach to dermatology cases can be daunting. Skin can only react in a limited number of ways, and with these limited manifestations, a dazzling array of skin diseases are represented. Good dermatological books, like this color handbook, provide a cartographer's guide directing the general practitioner through the landscape of lesions that represent dermatological disease.

This handbook is arranged into 13 chapters, organized by major symptoms (alopecia or pruritus), by lesion site (nasal dermatoses or nail diseases), or by age of onset (juvenile dermatoses).

The 350 color photographs are a useful accompaniment to the text, though, there are a few diseases without photographs. For example, it would have been beneficial to have a photograph of cutaneous inverted papillomas.

Another strength of this book are the many tables, such as the table on the differential diagnoses of alopecia, organized by whether the hair loss is focal to multifocal or symmetrical to diffuse. This is a helpful starting point when examining a patient with hair loss to determine what testing should be prioritized.

Since skin conditions involve reaction patterns that can be similar across diseases, there are lists of differential diagnoses for each disease. For instance, pemphigus folliaceous, exfoliative staphylococcal pyoderma, and *Trichophyton mentagrophytes* are the 3 conditions in which acanthocytes can be seen on cytology.

Advice is provided on ideal biopsy technique. For example, for bullous pemphigoid the epidermis should be included in the biopsy versus ulcers or erosions.

Lesion terminology is precise. The terms folliculitis or furunculosis are used to describe photographs of lesions, which makes it easy to apply to the practitioner's own cases.

My overall impression is that this book is filled with practical tips that could be applied the next day at your practice. An example is to consider hair plucks for mite checks on the periocular and interdigital skin; sites in which skin scrapings would be painful.

In my opinion, the core books in veterinary dermatology are Muller and Kirk's *"Small Animal Dermatology"* and Hnilica and Patterson's *"Small Animal Dermatology, A Color Atlas and Therapeutic Guide."* This book, however, provides a helpful adjunct to the conscientious general practitioner.

Reviewed by ***Christie-Leigh Capper, BA, BSc, DVM,*** *Mcleod Veterinary Hospital Winnipeg, Manitoba.*