Umbilical and periumbilical dermatoses

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The umbilicus may be the site of congenital and acquired malformations and may harbor clinical clues to the diagnosis of potentially fatal inherited disorders, primary skin conditions, and a variety of infectious diseases. Both benign and malignant tumors may involve the umbilicus, and some are unique to this site. Finally, cutaneous signs localized to this anatomic location may be found in diverse systemic diseases. (J Am Acad Dermatol 2015;72:1066-73.)

In utero, the umbilical cord lies adjacent to critical embryologic fusion planes of the abdominal wall and is responsible for vascular flow between the growing fetus and the placenta. The umbilicus is formed by the separation of the umbilical stump, which generally occurs 1 week after birth.[1] This article will review congenital and acquired anatomic abnormalities, cutaneous diseases, neoplasms, infectious diseases, and foreign material involving the umbilicus and periumbilical skin.

Abbreviations used:

ACD:	allergic contact dermatitis
AGA:	alfa-galactosidase A
C3:	complement C3
CMPI:	cow's milk protein intolerance
DIF:	direct immunofluorescence
IgA:	immunoglobulin A
IgG:	immunoglobulin G
LAD:	leukocyte adhesion deficiency
SMJN:	Sister Mary Joseph nodule

CONGENITAL MALFORMATIONS

Omphalomesenteric duct malformations

During embryologic development, the omphalomesenteric duct connects the yolk sac to the midgut and typically involutes by 6 weeks' gestation.[2] Failure to involute allows a direct connection between the umbilicus and the ileum. Malformations of the omphalomesenteric duct typically present at birth with umbilical drainage of intestinal contents, but can rarely become symptomatic in adolescence. Other signs and symptoms include a pink to red glistening umbilical nodule, described as a "raspberry tumor,"[3] melena, anemia, abdominal pain, intussusception, or intestinal obstruction.[2] Surgical excision is recommended.[4,5] Persistence of the most external portion of the omphalomesenteric duct is responsible for umbilical polyps, cysts, or sinuses.

Urachal malformations

The urachus connects the fetal bladder to the umbilicus and involutes with normal development. A patent urachus and an umbilical–urachal sinus are malformations that typically present in infancy. Clinical findings include drainage of urine from the umbilicus, an umbilical sinus with recurring periumbilical dermatitis,[6] a painful mass inferior to umbilicus, and recurrent urinary tract infections. Partial involution may present with umbilical drainage and recurrent periumbilical dermatitis or cellulitis, or as a painful mass just inferior to the umbilicus. Surgery is required to prevent secondary infection and potential development of adenocarcinoma.[7]

Abdominal wall defects

Exstrophy of the bladder, more common in males, is a rare, complex abnormality consisting of an

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absent lower abdominal wall inferior to the umbilicus, separation of the abdominal musculature and pubic rami, and externalization of the bladder epithelium (Fig 1, *A*).[3,5] If left untreated, patients are at increased risk of bladder cancer.

Gastroschisis is an abdominal wall defect that is characterized by extrusion of abdominal contents lacking a membranous covering (Fig 1, *C*). The defect arises to the right of the umbilicus and is likely caused by vascular compromise.[8,9] Surgery is necessary. An omphalocele is characterized by partial absence of the abdominal wall with externalization of abdominal contents covered by peritoneal membrane. It can be associated with anomalies of multiple organs, chromosomal abnormalities, and Beckwith–Wiedemann syndrome.[5,10]

CAPSULE SUMMARY

- A wide variety of disorders present with umbilical and periumbilical skin findings.
- We provide an update of malformations, inherited and acquired disorders, infections, neoplasms, and signs of systemic disease affecting this anatomic region.
- Knowledge of these clinical entities places the dermatologist in a unique position to aid in patient care.

Prune belly syndrome, a triad of hypoplastic abdominal musculature, cryptorchidism, and urinary tract abnormalities, is named for the characteristic finding of wrinkled abdominal skin (Fig 1, *D*). Multiorgan system malformations are present and multidisciplinary care is critical.[5]

Other umbilical and periumbilical dysplasias

A congenital umbilical hernia is caused by incomplete fascial closure around the umbilical ring. Spontaneous closure is typical by 5 years of age (Fig 1, *B*).[11] The hernia may uncommonly become incarcerated, strangulated, or rupture, necessitating surgical consultation. Associated conditions include the following: low birth weight, African American ethnicity, Ehlers–Danlos syndrome (dermatospraxis type),[12] Beckwith–Wiedemann syndrome,[13] mucopolysaccharidoses,[14] cutis laxa,[15] and congenital hypothyroidism.[16]

An umbilical granuloma, or granulation tissue that develops after cord separation,[17] is the most common cause of an umbilical mass in neonates. It manifests as a glistening, red umbilical papule and is treated with topical silver nitrate; if there is no improvement, obtaining a biopsy specimen is indicated. Surgical referral is indicated if ectopic tissue, such as pancreas or liver, is noted on the biopsy specimen.

Polythelia (supernumerary nipple) presents along the embryonic milk lines passing on each side of the umbilicus from the inguinal folds to the axillae. It appears as a skin-colored or pigmented umbilicated papule.

ACQUIRED ANATOMIC UMBILICAL ABNORMALITIES

Acquired umbilical hernias are more common in women in the fourth to sixth decades of life[18] and present as a soft protuberance of tissue at the umbilicus. Predisposing conditions include obesity, ascites, multiple pregnancies with prolonged labor, and large intraabdominal tumors. Surgical treatment is necessary because umbilical hernias in adults do not spontaneously involute.[19]

Inherited diseases

Leukocyte adhesion deficiencies (LADs) types I to III are autosomal recessive diseases caused by defects in leukocyte adhesion or activation and, in type III, platelet activation defects.[20] Types I and III present in neonatal life with omphalitis and delay in separation of the umbilical stump.[21] Other symptoms include mucositis, periodontitis, and delayed wound healing in LAD type I. Immunology referral is critical.[20]

Fabry disease is an X-linked disorder caused by mutations in the galactosidase alpha (GLA) gene resulting in alpha-galactosidase A (AGA) deficiency. It typically presents before puberty with signs and symptoms of abdominal pain, acroparesthesias, angiokeratoma corporis diffusum, corneal opacities, hearing loss, hypohydrosis, renal impairment, and cardiovascular and neurovascular disease. The abdomen and umbilicus are common sites for the angiokeratomas (Fig 2),[22] which may be arranged as "periumbilical rosettes."[3] The diagnosis can be confirmed by GLA gene sequencing.[23]

Pseudoxanthoma elasticum, caused by mutations of the ABCC6 gene, is inherited in an autosomal recessive fashion but may demonstrate pseudodominance. It presents in the second decade of life and is characterized by fragmentation and calcification of elastic fibers leading to cutaneous, ocular, and vascular sequelae.[24] Skin findings include yellow to orange papules on the lateral aspect of the neck and flexures, giving a "plucked chicken skin" appearance. Rarely, these papules are localized to the periumbilical region with or without systemic manifestations.[25] The typical patient without systemic manifestations is a multiparous,