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# Urachal abnormalities in the adult population. Is physical activity involved in clinical manifestation? Literature review

1. Joanna Męczyńska [JM]

   St. John Paul II Mazovian Provincial Hospital in Siedlce, Poland, Księcia Józefa Poniatowskiego 26, 08-110 Siedlce, Poland
   https://orcid.org/0009-0002-0292-8032
   joanna.meczynska@gmail.com

2. Aleksandra Mazurkiewicz [AM]
   Ludwik Rydygier Specialist Hospital in Kraków, Złotej Jesieni 1 Estate, 31-826 Kraków, Poland
   https://orcid.org/0009-0008-9427-9378
   aleksamazurkiewicz@gmail.com

3. Wojciech Mądry [WM]
   Ludwik Rydygier Specialist Hospital in Kraków, Złotej Jesieni 1 Estate, 31-826 Kraków, Poland
   https://orcid.org/0009-0003-9434-0478
   madry.mw@gmail.com

4. Justyna Marcicka [JM]
Ludwik Rydygier Specialist Hospital in Kraków, Złotej Jesieni 1 Estate, 31-826 Kraków, Poland
https://orcid.org/0009-0003-1766-7397
justyna.marcicka@gmail.com

5. Magdalena Kołodziej [MK]
Brothers Hospitallers Hospital in Kraków, Trynitarska 11, 31-061 Kraków, Poland
https://orcid.org/0000-0002-6597-1559
magdalenakolodziej502@gmail.com

6. Nazarii Saiuk [NS]
Ludwik Rydygier Specialist Hospital in Kraków, Złotej Jesieni 1 Estate, 31-826 Kraków, Poland
https://orcid.org/0000-0001-6722-0751
nazarii.saiuk@gmail.com

7. Tomasz Seredyński [TS]
St. Lucas Provincial Hospital Tarnów, Lwowska 178A, 33-100 Tarnów, Poland
https://orcid.org/0009-0000-7806-0220
tomasz.seredynski98@gmail.com

8. Adriana Wojciechowska [AW]
The Medical University of Lublin, Aleje Racławickie 1, 20-059 Lublin, Poland
https://orcid.org/0009-0006-9946-8448
adaw357@gmail.com

9. Michał Andrzej Kozicz [MAK]
Brothers Hospitallers Hospital in Kraków, Trynitarska 11, 31-061 Kraków, Poland
https://orcid.org/0009-0001-4254-4087
kozicz.michal@gmail.com

10. Weronika Salasa [WS]
Stefan Wyszynski Specialist Hospital in Lublin, Krasnicka 100, 20-718 Lublin, Poland
https://orcid.org/0000-0002-8683-2582
weronikasal@gmail.com

ABSTRACT

**Introduction**

The urachal anomaly is a developmental defect, a remnant of fetal life consisting of a patent connection between the dome of the urinary bladder and the umbilicus. It can take various forms. Most commonly it occurs in males, very rarely in adults. The main symptoms associated with this condition are: foul-smelling discharge from the umbilicus, abdominal pain, hematuria, or recurrent urinary tract infections. Imaging plays a crucial role in the diagnosis of the persistent urachus.

**Aim of the study**

The aim of the study is to sentize doctors' attention that patent urachus is the rare but possible diagnosis of acute abdomen pain in adults population and bring the proceedings.

**Materials and methods:**

An analysis of papers avaliable on Pubmed and Google Scholar was performed. The following key words: urachal anomalies, adults, abdomen pain, urachal remnants, urachal carcinoma were used.

**Results:**

Lately, thanks to modern precise imaging diagnostics, more valuable studies have been conducted, allowing to characterize residual ureteral remnants, which are most often asymptomatic in population of adults. However the manifestation of symptoms is mostly associated with infection of the mentioned area or a tumor transformation. There are at least two case reports which report that infection of urachal remnants could manifestate after intensive physical activity. It is worth in-depth interviewing. Studies indicate the necessity of surgical removal of persistent urachal anomalies, as they carry the risk of rapid growth and

unfavorable prognosis of cancerous spread.

Keywords: urachal abnormalities, urachal anomalies, adults, abdomen pain, urachal remnants, urachal carcinoma were used

**Introduction:**

Urachal anomalies are rare congenital developmental defects, which consists of a partially closed or completely patent channel between the dome of the bladder and the umbilicus. [1,2] In the majority of the population, this channel obliterates around mid-pregnancy, during the perinatal or infantile period. [3] In case of non-closure, in adults, a patent urachal channel usually does not present symptoms, but if they occur, they are manifested as an infection or neoplastic proliferation in the mentioned area. [4] Due to the rare occurrence of urachal abnormalities in the adult population, the literature mainly presents descriptions of clinical cases. [5] The aim of this review is to draw attention to the problem of persistent urachal anomalies as a rare, often overlooked developmental defect, that can be a cause of abdomal pain and soft tissues infection in adults patients, as well as to indicate the course of action.

**Epidemiology**

The frequency of urachal anomalies is difficult to determine because most of these defects are asymptomatic. [6] The majority of them manifest during early childhood, according to some research, around 80% are revealed by the age of 2 years. [7] This is undoubtedly a rarely recognized defect. Depending on the sources, in the population of children under 15 years old, it occurs in approximately 1.6%, and around 0.063% in adult population. [3,8,9] These defects are more commonly found in men, with a ratio of 2 men to 1 woman with the defect. [1,10,11,12,13] The age is raging from 20 to 40 years old. [2] The most common form described in the literature, occurring in about 50% of cases, is a persistent urachus. [11,14] Alternatively,

according to other sources, it can be a urachal cyst. [5,8,15] The most common benign manifestation of urachal remnants is infection. [15] The risk of neoplastic transformation in adults with persistent urachal anomalies is very high. According to the literature, nearly 50% of anomalies diagnosed in adulthood are neoplastic malformations. [7] However, other articles, state that the risk of malignancy is significantly lower than previously described. [3,8,16] According to the study by Joseph M. Gleason et al., to prevent a single adenocarcinoma in adulthood, the resection of 5,721 asymptomatic urachal remnants in children should be performed. [17]

**Patophysiology**

During embryonic development, the fetal bladder is connected to the umbilicus through a canal, which is used to empty the fetal bladder. This connection, called the urachus, originates from two structures: the cloaca and the allantois.[18] Around mid-pregnancy, as the urinary bladder descends into the pelvis, the canal dilates and then begins to obliterate. [10] After it closes up, it doesn't serve any function, which is why an unobliterated urachus is termed as anomaly. [19] From a histological standpoint, the urachus consists of three layers. The inner layer is typically composed of a transitional epithelium, the middle layer is a connective tissue, and the outer layer is a muscle, which forms a continuity with the detrusor muscle. [10,13] During the perinatal period, at the latest by 6 months of life, the urachus obliterates, forming a thin fibrous cord, which is called the median umbilical ligament. Once the lumen is obliterated, these layers are indistinguishable. [8,10,12] Delayed or incomplete closure associated with the quantity and location of tissue, results in one of the four forms of remnants: persistent urachus, urachal cyst, urachal sinus, or vesicourachal diverticulum.[7] Some sources also distinguish a fifth form known as the alternating sinus.[10] The urachus is located in the retroperitoneal space, between the transverse fascia and the parietal peritoneum, known as the space of Retzius. Its length varies from about 3 to 10 cm, and its diameter is approximately 8-10 mm.[9,10,13]

**Clinical presentation**

Urachal remnants in adulthood do not present typically clear symptoms for a long time. The first signs of abnormalities usually manifest before the age of 50.[16] There are four forms: persistent urachus, urachal cyst, urachal sinus, or vesicourachal diverticulum, optionally an alternating sinus.[10] While the persistent urachus is considered as a congenital defect, the other

forms may reopen after previous closure, for example, under the influence of infection of surrounding tissues. Some sources recognize these anomalies as acquired rather than congenital.[13] The most common symptoms of persistent urachal remnants include: abdominal pain, foul-smelling discharge from the umbilicus, urinary leakage from the umbilicus, tenderness, redness, palpable mass in the umbilical area, and hematuria. [19] Macro or microscopic hematuria may occur when the orifice to the dome of the urinary bladder is permeable.[9] Symptoms may vary, depending on the amount of faulty tissue and its location.[6,16] Redness, swelling, and tenderness around the umbilicus, both in children and adults, should raise suspicion of inflammation of persistent urachal remnants. Discharge from the umbilicus is not necessary for diagnosis. [2] We may observe it with a persistent urachus. However the most common symptom for vesicourachal diverticulum is hematuria. [12,14] Moreover in case of inflammation, high fever is often present. Diagnostic challenges arise when urachal anomalies manifest only with abdominal pain. [20] Physical examination may reveal palpable masses in the umbilical area. If there is uncertainty of whether the umbilical discharge contains urine, the creatinine level in urine can be measured. [6] In laboratory blood tests, leukocytosis and elevated inflammatory markers such as erythrocyte sedimentation rate (ESR) can be observed.[13]

*Patent Urachus*

The form of a persistent urachus indicates a completely patent channel. It is usually detected during childhood, according to some reports, even in the newborn period.[7,11,21] Sometimes the lumen of a persistent urachus is extremely narrow, so patients may remain asymptomatic.[10] In the physical examination, urine leakage from the umbilicus along with swelling of this area, granulomas can be observed. Also delayed healing process after cutting the umbilical cord may occur.[11]

*Urachal cyst*

An urachal cyst forms when the ends of the persistent urachus are obliterated into a ligament and beyond the middle section, most commonly the lower 1/3 it forms a cyst. It does not connect either to the urinary bladder or to the umbilicus.[10,11] This form rarely gives symptoms due to its size. Therefore, diagnosis mainly occurs in late childhood or adulthood as an infection or incidental finding during the diagnosis of other anomalies. [11]

*Umbilical-urachal sinus*

This sinus forms when the umbilical end of the persistent urachus does not obliterate and persists as a spindle-shaped structure, just beneath the umbilicus. It is characterized by a lack

of communication with the bladder.[10] Patients typically report tenderness in the umbilical area, with periodically appearing purulent discharge and a sensation of chronically wet umbilicus.[11,12]

*Vesico-urachal diverticulum*

This is the rarest form of ureteral remnants, accounting for 3% to 5% of cases.[22] It constitutes incomplete closure of the urachal canal on the side of the dome of the urinary bladder. A diverticulum, a bulging, appears on the anterior dome of the urinary bladder. Most commonly, due to its large opening and good communication with the urinary bladder, it does not cause any symptoms and is detected incidentally. In case of a narrow opening, there can be the formation and accumulation of stones, which can lead to frequent urinary tract infections.[13] Diagnosis mainly occurs in adulthood.[10,11]

*Alternating sinus*

The definition of an alternant sinus is considered as a cystic dilation of the urachus, which periodically drains into the bladder or the umbilicus. It is often not included in literature. [10]

**Diagnosis**

Due to the various clinical presentations and fact that these anomalies are frequently asymptomatic, urachal malformations are a significant diagnostic challenge.[8] Modern imaging, including cross-sectional views, allows for much more frequent diagnosis, even when it presents no symptoms. [3,11,12] The definitive diagnosis can only be made after prior surgical intervention and obtaining confirmatory histopathological results.[1,14] Ultranosography is considered as the first-line imaging modality for diagnosing urachal remnants.[1,5,8,15,22] These structures are located extraperitoneally, moreover, they are situated near the urinary bladder. The estimated percentage value of detecting a urachal cyst in ultrasonographic examination is 79-83%. [5] When ultrasonographic examination fails to visualize urachal remnants, computed tomography can be used. In specific cases requiring even more detailed imaging, magnetic resonance imaging or fistulography, cystoscopy can be performed. [11,12,15] In ultrasonographic examinations, the most commonly visualized findings include a canal with a hypoechoic wall and anechoic content extending from the dome of the urinary bladder to the umbilicus, or a homogeneous fluid-filled structure between obliterated ends adjacent to the umbilicus and the dome of the bladder. Less frequently thickening and spindle-shaped blind dilation of the ureter at the umbilical opening, without connection to the urinary bladder, which indicate vesicourachal diverticulum is observed. [11] In CT scans, periumbilical inflammatory

foci can be observed.[7] CT scans also can provide more detailed information on the severity of inflammation and its depth, including involvement of muscles and fascia.[2] In case of neoplastic growth, ultrasonographic examination may reveal a heterogeneous, hyperechoic change with abundant vascularity in the examined area.[15,23] Examinations such as CT, and especially MRI, allow for the assessment of the tumor's stage, the presence of metastases in lymph nodes, and further structures. In CT imaging, a cystic mass with solid or mixed consistency which often contains calcifications may be visible.[14] Calcifications are considered to be the pathognomonic signs of urachal tumors.[24] CT has limited sensitivity regarding the extent of tumor invasion, reaching about 50%.[15] The diagnosis of a tumor in MRI imaging typically involves visualizing a heterogeneous mass with areas of high intensity, which enhance depending on the projection.[14] PET imaging is not the preferred diagnostic method, however, it can reveal the presence of soft tissue with scattered calcifications, mildly avid for FDG tracer.[25] The study by C. Niedworok et al. indicates the presence of elevated tumor markers RHAMM, IMP3, Ki67, and p53. However, it notes that the studies were conducted on 26 patients and should be further expanded to a larger, more diverse group of patients.[26]

Diffrent diagnosis

Persistent urachus can present clinical symptoms that closely resemble umbilical hernia. A study from North Shore University Hospital highlights the difference in imaging by indicating a lack of connection to the urinary bladder and the presence of fat and intestinal fragments in case of umbilical hernia.[7,21] In case of doubts arising from ultrasound examination, a computed tomography scan should be performed.[7] Furthermore urachal anomalies may resemble Meckel's diverticulitis, prostatitis, or appendicitis. [7,13,19,20,21] Sometimes it resembles pelvic inflammatory disease or urinary tract infection. [19] During the diagnostic process, it is also necessary to exclude hematoma, abscess, umbilical hernia, ureteral cancer, bladder cancer, and abdominal wall tumors.[7,20]

**Treatment**

Urachal remnants may resolve spontaneously in infants which are below 6 months of age.[9] However, due to frequent infections and the high risk of malignant transformation, urachal anomalies detected in the adult population should be treated surgically.[1,27] It is observed that around 30% of infections relapse after conservative tretment.[9] Systematic excision of urachal lesions is safer. In a study from Nuestra Señora de Candelaria University Hospital in Tenerife,

in the case of two patients, a lesion that appeared benign in preoperative diagnosis, turned out to be malignant in postoperative histopathological examination.[12] The recommended treatment for symptomatic infection urachal remnants posing a risk of peritonitis is to start with broad-spectrum antibiotic therapy, followed by surgical excision. .[2,7,13,20] It includes both Gram-positive and Gram-negative bacteria. Preoperative antibiotic therapy reduces the risk of complications and shortens hospitalization.[2] The excision of a urachal cyst can be performed in a one-stage procedure which is a complete removal of the cyst, or in a two-stage procedure, by incision and drainage of the infected cyst under ultrasound guidance, followed by its subsequent removal. [7] Drainage may take the percutaneous form or through the urinary bladder with the assistance of a JJ stent.[13] The traditional technique of removing a cyst involves obtaining transverse subumbilical access or a vertical incision along the midline. [11] Mostly laparoscopic techniques are used. The removal of urachal anomalies may be accompanied by other surgeries such as cholecystectomy, umbilical hernia repair, or oophorectomy. [8]

In patients without other accompanying pathologies, the preferred surgical technique is three-port left-sided laparoscopy with preservation of the umbilicus and urinary bladder.[8] The first port can be inserted about 3-5 cm above the umbilicus, with subsequent ports placed to the left of the umbilicus, ensuring optimal visibility.[20] According to some researches, if the anomalies extend to the dome of the bladder, surgical treatment requires segmental resection of the bladder.[9,14] In a significant number of cases, umbilical resection is not required. Studies show that there are no recurrences of infections or malignant transformations after conservative surgery. It also allows to maintain a good visual effect, especially in young patients. [9] There are also studies showing that resection can be performed with satisfactory cosmetic results. A good example of this is LESS (laparoendoscopic single-site surgery), which involves making a Y-shaped incision around the umbilicus, followed by a three-flap plastic surgery. A study by Hakushi Kim et al. demonstrated better cosmetic outcomes and comparable therapeutic effects to conventional laparoscopy.[4] The intraoperative use of a Foley catheter is justified in cases of urinary bladder leakage and possibility of performing bladder cuff resection.[8] The removal of residual urachal remnants results in the complete resolution of symptoms.[3] The majority of patients do not experience any postoperative complications following sparing laparoscopic surgery[8] In case of malignant tumor detection, partial or radical cystectomy should be performed along with complementing treatment with chemotherapy. Studies conducted by Ashley and colleagues did not find any difference between the application of these forms of surgery and the average survival time of patients.[3]

**Complications and prognosis:**

The presence of persistent forms of the urachus in the adult population most commonly leads to the formation of granulomas around the umbilicus, recurrent urinary tract infections, or tumor transformations.[11,14] The most common bacteria causing recurrent infections are Staphylococcus aureus, Escherichia coli, Enterococcus, Citrobacter, Klebsiella, and Proteus. [2,11] Complications such as intestinal obstruction, urinary tract damage, and bleeding have also been noted. Untreated infection can lead to the formation of an abscess, which gradually enlarges and may rupture, ultimately leading to peritonitis and sepsis or necrotizing fasciitis [10,15] Tumors can originate from the epithelial lining, as well as from the mesenchymal tissue.[28] Moreoften they take on a malignant form than a benign one.[11] Among the benign forms, there are adenomas, fibroids, adenofibromas, leiomyomas, and hamartomas. They often mimic malignant tumors.[10,27] Frequently they manifest as fatty infiltration in the Retzius' space.[27] While adenocarcinoma in the mentioned area has never been reported in pediatric population, the malignancy was present in a significant number of cases of urachal remnants in adults,. Due to the negligible symptoms, local tumor progression or extensive metastases are usually noted at the first appointment.[15] Among primary tumors, they are mainly diagnosed in the fourth-fifth-sixth decade of life. [10,23,24] They include less than 1% of bladder tumors. The most common type is adenocarcinoma with a mucinous subtype, according to some sources up to 80%, [23] followed by unspecified and signet ring cell type. Their extraperitoneal location means that, they do not cause any symptoms for a long time.[10] The most common symptom of a tumor transformation is hematuria. [24,26] In histopathological examination, mucinous, intestinal, and signet ring cells are present. [6] Due to the paucity of symptoms, the prognosis for patients with urachal cancer is unfavorable.[24] This cancer rapidly infiltrates the peritoneum and metastasizes to bones, lungs, and liver.[28] According to studies, the 10-year survival rate reaches approximately 49% of patients after surgical treatment for the tumor and palliative chemotherapy offers a chance of approximately 10% for patients with metastases.[23] Another researches indicate a 5-year survival rate of around 50%.[24,26] Some studies indicate KRAS mutation and microsatellite instability, which tends to occur often in urachal tumors as the chance of biological treatment and better prognosis. [29] An extremely rare complication of mucinous urachal carcinoma is dissemination to the peritoneum in the form of pseudomyxoma peritonei which should also be considered as a couse of abdominal pain [30,31]

**Tie-up between urachal remnants and physical activity**

There are at least three case reports which indicate that infection of urachal remnants appeared after intensive physical activity. In one of them a 24 year-old healthy athlete started to feel unusual pain in of abdomen after vigorous exercise in this area. [5] The second case report describes 22-year old football player with no previous medical history of periumbilical pain who had been hit in the lower abdomen while playing football. He started to feel minor discomfort which increased over time.[32]

In the third case a 14-year old also healthy football player with a 2-year experience started to feel a strong discomfort after a session of training which included abdomen exercises. [33] The periumbilical pain was so severe that each of them was looking for help in the emergency room. [5,32,33] After intensive physical effort or a hit the overlayers in abdomen area are taut. This promotes bacteria to grow and cause inflammation when urachal remnants are present. [5]

**Conclusions**

The literature review unequivocally indicates the presence of persistent urachus not only in children but also in the adult population as well. When it manifests in adulthood, it carries a number of risks, often with a component of malignancy. To prevent complications, especially the spreading of the tumor process, a diagnosis should be made as soon as possible. It is undoubtedly important to examine patients presenting with nonspecific abdominal pain. It can be helpful if abdominal pain is accompanied by objective symptoms such as foul-smelling discharge or urine leakage from the umbilicus, swelling and pain in this area, muscle guarding, fever, and hematuria. Intensive physical effort could be a precious indication taken from the interview to consider urachal remnants as the cause of the abdomen pain. Ultrasonography is used as the first-line imaging diagnostic, and in case of visualization problems, the patient should be referred for CT or MRI. The therapeutic pathway for urachal remnants should begin with early antibiotic therapy followed by surgical removal.

**Author's contribution**

Conceptualization, Joanna Męczyńska, Aleksandra Mazurkiewicz, Wojciech Mądry; Methodology, Magdalena Kołodziej; Software, Justyna Marcicka; Check, Justyna Marcicka, Nazarii Saiuk and Michał Andrzej Kozicz; Formal Analysis, Adriana Wojciechowska and Magdalena Kołodziej; Investigation, Weronika Salasa and Adriana Wojciechowska; Resources, Joanna Męczyńska Aleksandra Mazurkiewicz; Data Curation, Tomasz Seredyński; Writing -

Rough Preparation, Nazarii Saiuk; Writing - Review and Editing, Michał Andrzej Kozicz and Weronika Salasa; Visualization, Tomasz Seredyński; Supervision, Weronika Salasa; Project Administration, Joanna Męczyńska; Receiving Funding, Tomasz Seredyński.

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