Polish Annals of Medicine

Journal homepage: https://www.paom.pl

Case Report

Localized amyloidosis of the uterine cervix: A case report

Natalia Ewa Brzezińska

[1] *Department of Obstetrics and Gynecology, Oncological Gynecology and Gynecological Endocrinology, Faculty of Medicine, Medical University of Gdansk, Poland*

Article info

Article history
Received: December 26, 2024
Accepted: February 7, 2025
Available online: November 5, 2025

Keywords
amyloidosis
uterine
cervix
reproductive tract

Doi
https://doi.org/10.29089/paom/201131

Abstract

Introduction: Amyloidosis is a disease characterized by the deposition of misfolded proteins in tissues, which aggregate into insoluble fibrils, leading to progressive organ damage. It presents with a wide spectrum of clinical manifestations depending on the location. Localized amyloidosis of the uterine cervix is exceedingly rare and may pose diagnostic challenges. Gynecological amyloidosis can be asymptomatic or present with symptoms such as abdominal pain, postmenopausal bleeding, or menorrhagia.

Aim: This case report aims to highlight the occurrence of amyloidosis in gynecological practice and to describe the clinical, radiological, and histopathological features, along with the management approach leading to successful treatment.

Case study: We report the case of a 52-year-old woman who presented with isolated abdominal pain. Transvaginal ultrasonography revealed a tumor at the cervico-uterine junction. A biopsy confirmed the diagnosis of localized cervical amyloidosis. Systemic involvement was excluded following whole-body radiological evaluation.

Results and discussion: Laparotomy with radical hysterectomy and bilateral adnexectomy was conducted. Complete excision of the amyloid deposits was achieved without complications. Surgical treatment successfully alleviated the presenting symptoms.

Conclusions: Localized amyloidosis of the uterine cervix represents a distinct clinical entity. Early diagnosis is crucial for effective management and symptom resolution. Surgical excision remains the preferred treatment for localized amyloidosis in gynecological sites, with histopathological confirmation required for diagnosis. This case underscores the importance of considering amyloidosis in the differential diagnosis of unusual gynecological presentations.

Corresponding author: Natalia Ewa Brzezińska; Department of Obstetrics and Gynecology, Oncological Gynecology and Gynecological Endocrinology, Faculty of Medicine, Medical University of Gdansk, Smoluchowskiego 17, 80-214 Gdańsk, Poland; E-mail address: nataliabrzezinska@gumed.edu.pl

1. INTRODUCTION

Amyloidosis is a rare and heterogeneous group of disorders characterized by mainly extracellular deposition of misfolded protein fibrils, which disrupts normal tissue architecture and function leading to the inflammatory response.[1] The disease can be systemic, affecting multiple organs, or localized, confined to a specific organ or tissue. Thus far, 36 proteins have been identified as amyloidogenic in humans, with at least 17 capable of causing systemic disease.[2] Systemic amyloidosis affects multiple organs, with the kidneys, heart, liver, gastrointestinal tract, nerves, and soft tissues being the most frequently involved. As a result, its clinical presentation is diverse and often nonspecific, making diagnosis challenging. Common symptoms include unexplained weight loss, fatigue, chronic diarrhea, congestive heart failure, arrhythmias, peripheral edema, hepatomegaly, orthostatic hypotension, and proteinuria. In approximately 15% of cases, more distinctive signs, referred to as 'red flags', are observed, such as macroglossia, submandibular edema, and 'raccoon eyes' (periorbital purpura).[3] In systemic amyloidosis, the amyloidogenic protein is synthesized in one location (e.g., bone marrow or liver) and deposited in distant sites (e.g., heart or kidneys). Conversely, localized amyloidosis arises when amyloid production and deposition occur at the same site. Localized amyloid deposits are often associated with endocrine disorders and originate from hormones or local protein precursors.[2] Protein aggregation may result in a wild range of symptoms of the disease, depending on the location of the deposits and their amount. The only definitive method for diagnosis of amyloidosis is tissue biopsy. The deposits are identified by their binding to Congo red and their characteristic yellow-green birefringence when observed under polarized light, regardless of the protein type.[4] To distinguish systemic from local disease whole body imaging should be performed. Cases involving the reproductive tract are extremely rare. According to the literature, amyloid deposits have been identified in the vagina, cervix, uterus, and vulva.[5–15] Most of the reported cases of diagnosed amyloidosis in gynecological practice present with either postmenopausal bleeding or menorrhagia, leading to malignancy concerns and unnecessary stress. The clinical presentation is highly variable, often with nonspecific symptoms leading to diagnosis concerns and late treatment.[4–14]

2. AIM

We present a case of localized amyloidosis confined to the uterine cervix, an exceptionally rare occurrence. This case report aims to raise awareness of the potential for this condition in gynecological practice, illustrating its clinical, radiological, and histopathological manifestations. Additionally, it seeks to provide guidance for effective diagnosis and management strategies.

3. CASE STUDY

A 52-year-old female patient was admitted to the gynecology department with a tumor located at the cervico-uterine junction which was diagnosed by transvaginal ultrasound (Figure 1). The ultrasound examination was prompted by nonspecific, persistent lower abdominal pain that had persisted for several months. The patient denied experiencing any abnormal vaginal bleeding, weight loss, or fatigue. On physical examination a blood pressure was 118/68 mmHg and a heart rate of 55 bpm, with no palpable abdominal masses detected during the physical examination. Pelvic examination findings were unremarkable. Laboratory evaluations, including hemoglobin (14.6 g/dL), platelets (149 G/L), serum creatinine (0.66 mg/dL), sodium (147 mmol/L), potassium (4.1 mmol/L), D-dimers (264 ng/mL), clotting time, inflammatory and hepatic markers, were all within normal ranges. The patient's obstetric history was notable for a gravida of six and a parity of five (all spontaneous vaginal deliveries) and one miscarriage in the first trimester. She underwent menopause at the age of 51. Her past medical history included hearing loss, cleft palate, and a prior appendectomy. She was not on any regular medications.

During a hospitalization a loop electrosurgical excision procedure (LEEP) biopsy was performed on the suspicious lesion following the endometrial curettage, with no complications. Histopathologic analysis of the biopsy specimen revealed amyloidosis, with immunohistochemical staining positive for amyloid deposits, CD68 negativity, and paCK negativity. Further imaging with a contrast-enhanced magnetic resonance imaging (MRI) of the pelvis showed a tumor mass occupying the vaginal vault, measuring 61 mm (cranio-

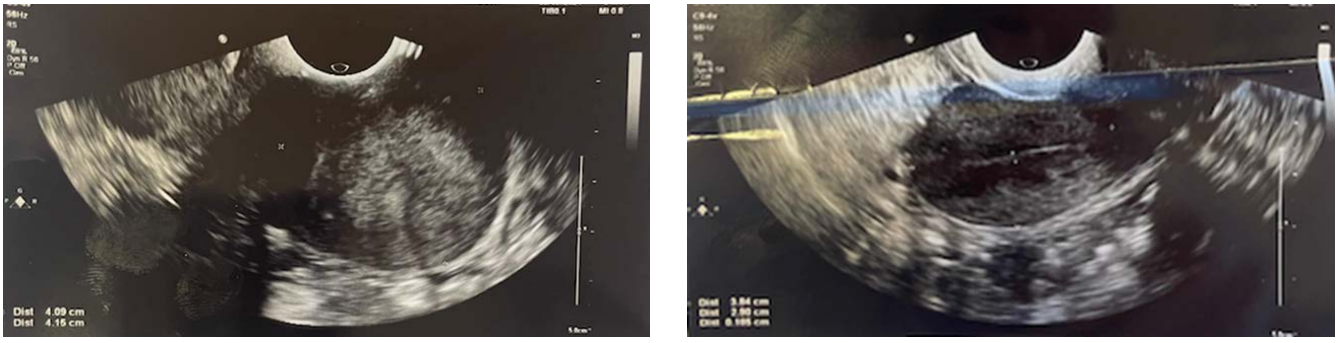

Figure 1. Transvaginal ultrasound image shows a mass measuring 46 × 44 × 54 mm with variable echogenicity located at the cervico-uterine junction.

caudal) × 48 mm (antero-posterior) × 62 mm (transverse). The tumor demonstrated a heterogeneous, predominantly intermediate signal on T2-weighted sequences, with heterogeneity on diffusion-weighted imaging (DWI) and irregular enhancement post-contrast on T1-weighted imaging. The posterior-superior vaginal wall adjacent to the tumor, over a 25-mm area suggested invasion of the vaginal wall. The cervix and isthmus appeared to have a preserved signal, with a blurred boundary observed between the mass and the cervical vaginal portion. The lower two-thirds of the vagina remained uninvolved. Adjacent pelvic organs showed no evidence of invasion, and no pathological lymphadenopathy was detected. A minor amount of free fluid was noted in the pouch of Douglas. To exclude the systemic amyloidosis, whole-body evaluation with contrast-enhanced computed tomography (CT) of the head, chest, abdomen, and pelvis was conducted. Only a localized enhancement at the cervico-uterine junction, measuring 82 × 63 mm, was noted, with no signs of additional amyloid deposits elsewhere. Given the localized extent of the disease and significant symptomatic burden, the patient was deemed a candidate for surgical intervention. She subsequently underwent a laparotomy with radical hysterectomy and bilateral adnexectomy. The surgical procedure proceeded without complications, and the patient was discharged after 4 days with plans for outpatient follow-up. Final pathological examination of the surgical specimen confirmed amyloid deposits within the cervical myometrium, with the largest focus measuring 48 mm. The ectocervical and serosal layers appeared histologically normal, and the endometrium was atrophic. At the 6-month follow-up, the patient remains in good general health, with no reported symptoms or evidence of disease recurrence.

4. RESULTS AND DISCUSSION

Gynecological amyloidosis, both localized and systemic form, is an extremely rare condition, with only view cases described in the literature.[5] The underlying predisposing conditions include monoclonal gammopathy, multiple myeloma, or other lymphoplasmacytic disorders known to result in production of monoclonal immunoglobulins, persistent uncontrolled inflammatory diseases such as autoinflammatory disease and a family history of amyloidosis.[1] The described in literature causes of gynecological amyloidosis included primary systemic amyloidosis, multiple myeloma, rheumatoid arthritis, familial Mediterranean fever inducted amyloidosis.[5] Although amyloidosis is a benign disease it can lead to the potentially life threating complications, such as a vaginal hemorrhage.[5] The increased bleeding is considered to occur due to the reduced activity of factor X, vascular infiltration with amyloid, and abnormal liver function due to amyloid deposition.[17–19] That is why early diagnosis can avoid extended area of vessel involvement and implement proper treatment before hemorrhage occurrence. In our case the protein accumulations were isolated to the myometrium, without blood vessel deposition and without signs of post-menopausal bleeding. According to the Copeland et al., amyloid infiltration of corpus uteri compromises uterine contractions, eventually promoting prolonged bleeding events during menstrual periods.[12] Our patient didn't experience abnormal bleeding and only abdominal pain was noted. Therefore, uterus amyloidosis may be more common than anticipated, but misdiagnosed on the early stage of the disease. Previously reported cases of amyloid deposition of the cervix were associated with an invasive squamous cell carcinoma in the same site. In this case there was no evidence of an underlying abnormality.[9]

5. CONCLUSIONS

(1) Localized amyloidosis confined to the uterine cervix is extremely rare in gynecologic practice.
(2) Diagnosis is made through histopathological evaluation and exclusion of systemic amyloidosis.
(3) Amyloid deposition in the female reproductive organs may be more common than previously reported in the literature. The absence of bleeding symptoms may lead to overlooked examinations of the reproductive tract.

Conflict of interest
None declared.

Funding
None declared.

References
1 Picken MM. The Pathology of Amyloidosis in Classification: A Review. *Acta Haematol*. 2020;143(4):322–334. https://doi.org/10.1159/000506696.
2 Muchtar E, Dispenzieri A, Magen H, et al. Systemic amyloidosis from A (AA) to T (ATTR): a review. *J Intern Med*. 2021;289(3):268–292. https://doi.org/10.1111/joim.13169.
3 Nagajewski A, Muzolf M, Michniowska K, et al. Immunoglobulin light-chain amyloidosis – Diagnosed through electrocardiographic and echocardiographic features. *Pol Ann Med*. 2016;23(2): 156–160. https://doi.org/10.1016/j.poamed.2016.01.012.
4 Benson MD, Buxbaum JN, Eisenberg DS, et al. Amyloid nomenclature 2018: recommendations by the International Society of Amyloidosis (ISA) nomenclature committee. *Amyloid*. 2018;25(4):215–219. https://doi.org/10.1080/13506129.2018.1549825.
5 Cansu DÜ, Teke HÜ, Arik D, Korkmaz C. Menorrhagia due to uterine amyloidosis in familial Mediterranean fever: case-based review. *Rheumatol Int*. 2021;41(1):205–211. https://doi.org/10.1007/s00296-020-04721-2.
6 Takimoto Y, Kogiku A, Konda E, Samoto T, Ishihara M, Yamashita T. Primary localized amyloidosis of the

uterine cervix during pregnancy. *J Obstet Gynaecol Res*. 2020;46(5):787–790. https://doi.org/10.1111/jog.14218.

7 Tsang WY, Chan JK. Amyloid-producing squamous cell carcinoma of the uterine cervix. *Arch Pathol Lab Med*. 1993;117(2):199–201.

8 Pehlivanov B, Belovegdov V, Ivanov G, Ivancheva H. Primary localised amyloidosis of the vagina. *Aust N Z J Obstet Gynaecol*. 2008;48(1):120–122. https://doi.org/10.1111/j.1479-828x.2007.00812.x.

9 Gibbons D, Lindberg GM, Ashfaq R, Saboorian MH. Localized amyloidosis of the uterine cervix. *Int J Gynecol Pathol*. 1998;17(4):368–371.

10 Lee JA, Angus B. Amyloidosis of the uterine vessels: an unusual cause of menorrhagia. *Br J Obstet Gynaecol*. 1993;100(11):1056–1057. https://doi.org/10.1097/00004347-199810000-00012.

11 Taylor E, Gilks B, Lanvin D. Amyloidosis of the uterine cervix presenting as postmenopausal bleeding. *Obstet Gynecol*. 2001;98(5 Pt 2):966–968. https://doi.org/10.1016/s0029-7844(01)01504-6.

12 Copeland W Jr, Hawley PC, Teteris NJ. Gynecologic amyloidosis. *Am J Obstet Gynecol*. 1985;153(5):555–556. https://doi.org/10.1016/0002-9378(85)90472-7.

13 Jongen VH, Grond AJ, van Veelen H, Santema JG. Uterine amyloidosis in menopause. *Br J Obstet Gynaecol*. 1998;105(3):362–364. https://doi.org/10.1111/j.1471-0528.1998.tb10102.x.

14 Winkler DD, Emery JA, Alan CB. Amyloidosis of the endometrium: an asymptomatic presentation. *Obstet Gynecol*. 2004;104(5 Pt 2):1144–1147. https://doi.org/10.1097/01.aog.0000124985.48138.7e.

15 Yue CC, Lampman JH, Park CH, Ballou SP. Secondary amyloidosis: diagnosis from an endometrial biopsy. *Arthritis Rheum*. 1983;26(10):1295–1296. https://doi.org/10.1002/art.1780261026.

16 Kotru M, Chandra H, Singh N, Bhatia A. Localized amyloidosis in endometrioid carcinoma of the uterus: a rare association. *Arch Gynecol Obstet*. 2007;276(4):383–384. https://doi.org/10.1007/s00404-007-0357-x.

17 Yood RA, Skinner M, Rubinow A, Talarico L, Cohen AS. Bleeding manifestations in 100 patients with amyloidosis. *JAMA*. 1983;249(10):1322–1324.

18 Mumford AD, O'Donnell J, Gillmore JD, Manning RA, Hawkins PN, Laffan M. Bleeding symptoms and coagulation abnormalities in 337 patients with AL-amyloidosis. *Br J Haematol*. 2000;110(2):454–460. https://doi.org/10.1046/j.1365-2141.2000.02183.x.

19 Sucker C, Hetzel GR, Grabensee B, Stockschlaeder M, Scharf RE. Amyloidosis and bleeding: pathophysiology, diagnosis, and therapy. *Am J Kidney Dis*. 2006;47(6):947–955. https://doi.org/10.1053/j.ajkd.2006.03.036.