Early detection of pilonidal sinus cancer and approach of suspected cases: rationale and recommendations

Firas Safadi (1), Marius Dettmer (2), Dietrich Doll (3)

REVIEW

(1) Department for Visceral, Thoracic and Vascular Surgery, University Hospital Carl Gustav Carus, Technische Universität Dresden, Dresden, Germany.
(2) Dept. of Trauma Surgery and Orthopaedics, Reconstructive and Hand Surgery and Burn Medicine, German Armed Forces Central Hospital Koblenz, Koblenz, Germany.
(3) Dept. of Procto-Surgery & Pilonidal Sinus, St. Marienhospital Vechta, Academic Teaching Hospital of the MHH Hannover, Vechta, Germany.

REVIEWERS: UK, CK

Corresponding author: Firas Safadi. Department for Visceral, Thoracic and Vascular Surgery, University Hospital Carl Gustav Carus, Technische Universität Dresden, Dresden, Germany.
Email: doctor.safadi@gmail.com

Date of submission: 9/08/2023

ABSTRACT

Introduction: Chronic and neglected cases of pilonidal sinus disease may be complicated by malignant transformation, which is more common than reported. Carcinoma of the pilonidal sinus disease is considered an aggressive tumor with a higher stage at diagnosis, higher recurrence rate, and worse prognosis when compared to primary skin tumors. Till now, there are no clear recommendations for the early detection of this disease in patients with pilonidal sinus.

Methods: The available literature about the incidence, treatment, and prognosis of pilonidal sinus carcinoma was reviewed to develop a list of recommendations for the early detection of the disease.

Results: Three sets of recommendations are suggested for the early detection of carcinoma in patients with pilonidal sinus disease. A recurrence or non-healing of the chronic disease should be considered a malignant transformation until proven otherwise.

Conclusion: Since treatment in the early stages of the disease is associated with a better prognosis, surgical practitioners should know and endorse the recommendations for early detection of pilonidal sinus carcinoma.

Keywords: pilonidal sinus, pilonidal sinus carcinoma, squamous cell carcinoma. pilonidal surgery, early detection of cancer

ISSN: 2207-7537
DOI: 10.5281/zenodo.10627954

Pilonidal sinus disease (PSD) is one of the most encountered pathologies in surgical practice. Pilonidal sinus disease carcinoma (PSDCA) is a rare complication of the longstanding disease, which may arise in about 0.1% of patients with pilonidal disease [1]. PSDCA is characterized by higher recurrence and mortality rates compared with primary skin cancers [2]. The advanced tumor stage at diagnosis contributes to this worse prognosis [3], which underlines the importance of early detection of the disease. Till now, there are no explicit recommendations for the early detection of PSDCA. This review aims to draw the surgeons' attention to this potential diagnosis in every PSD patient. The importance of early detection will be highlighted and specific recommendations will be framed based on the available evidence.

Why is the detection of PSDCA important?

Higher incidence than expected

Carcinoma of the pilonidal sinus was long considered a very rare complication of neglected or prolonged PSD. Although the incidence was estimated at 0.1% of all PSD cases [1], some works showed a higher rate. The incidence reached even 1.6% among 123 PSD patients in one series, although the study was underpowered by the small sample size [4]. We illustrated in previous work that the calculated worldwide incidence of all reported cases equaled 0.17% based on the study of 12,887 patients with PSD [5].
Due to underreporting, among other causes such as the absence of PSDCA as a discrete type in the cancer registries, the actual incidence may be at least 0.2%, if not even more [5]. Of course, one should acknowledge that the available data about the incidence of the disease is very limited and it is not underpinned by robust epidemiological studies. Due to this fact, it is also not possible to determine the rate of diagnostic error in pilonidal sinus carcinoma based on the retrospective data.

Although minimal, the incidence of PSDCA may reach considerable significance in countries with particularly common pilonidal disease. Duman et al. found changes consistent with pilonidal sinus in about 6.6% among more than 19,000 examinees [6]. An increased incidence of pilonidal sinus disease was recently documented in many countries. For example, the total number of PSD inpatients showed a one-third increase within 13 years in Germany [7]. Therefore, it is expected that the incidence of PSDCA would also increase worldwide parallel to that of pilonidal disease. The literature confirms these findings, as more than half of the reported PSDCA cases were published in the last twenty years [5].

Aggressive behavior

The available data which included all reported cases of PSDCA confirm that the disease can show aggressive behavior with a poor prognosis [8]. First, the disease tends to spread and advance locally; an invasion of the sacral bone, sometimes with extension to the spinal canal and rectal wall, was described on first diagnosis in many cases [9]. Second, an advanced stage on presentation is the rule, with more than 90% of the tumors being in stage III and IV [8], although it is not possible to know if the late diagnosis is due to delayed medical care or simply due to patients' neglect of their chronic condition. Third, the overall survival rate of PSDCA is around 60% [2], which is significantly lower than the reported survival rate of 98% in primary squamous cell carcinoma of the skin [10]. Fourth, the prognosis is far worse in advanced stages compared with early disease, with a reported 5-year survival of 47.8% in stage IV versus 70.8% for stages I and II [8]. Fifth, the recurrence rates that exceed 40% are also considerably higher than cutaneous squamous cell carcinoma, which shows a total recurrence rate of less than 5% [3,8,10]. Finally, the treatment of the advanced cases of the disease is not without challenges. These often require radical resection, complex surgical reconstruction, adjuvant therapy, long hospitalization, and high costs [11,12].

Although all the available survival and recurrence rates are derived from potentially limited retrospective data that included less than 150 reported patients so far [8], these facts show that PSDCA is an aggressive disease that can originate on a background of a common pathology. As the summary in Table 1 shows, surgeons must consider this disease in every PSD patient. Therefore, the need for clear recommendations for the early detection of PSDCA cannot be underestimated.

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• High frequency of PSD in some populations • Potential underestimation of PSDCA incidence in PSD patients • Increasing incidence of PSD and PSDCA in the last decades • Advanced stage at the first diagnosis of PSDCA • Poorer prognosis of PSDCA compared with primary skin carcinoma • Therapeutic challenges in advanced PSDCA

Table 1. The rationale of the early detection of PSDCA.
PSD, pilonidal sinus disease; PSDCA, pilonidal sinus disease carcinoma

Recommendations for the early detection of PSDCA

Table 2 presents our suggested recommendations for the early detection of PSDCA. These can be considered in three different stages: on the initial approach of any patient with pilonidal sinus disease, in the phase of operative planning, and after the surgical resection of PSD. In the next paragraphs, we discuss the available evidence for these recommendations.

General recommendations for all PSD patients
• Consider PSDCA in every pilonidal sinus even in young patients or early disease. • Ask carefully about the development of symptoms and the duration of the disease. • Critically look for the clinical signs of malignancy. • Examine the inguinal lymph nodes and the anal canal. • Warn patients with a neglected disease about potential malignant transformation.
Detection before PSD resection
• Consider a preoperative tissue biopsy in every patient with clinical suspicion. • Obtain multiple deep biopsies with favoring the margins and avoiding the necrotic areas. • Consider simultaneous FNA in the case of inguinal lymphadenopathy. • If the findings of FNA or punch biopsy were negative despite clinical suspicion, repeat the biopsy or consider a surgical incisional biopsy. • If malignancy is confirmed, do not proceed to resection before a complete staging.
Detection after PSD resection
• Submit all PSD specimens for histological examination regardless of the patient's characteristics. • Pathologists should conduct a critical macroscopic and microscopic examination of PSD specimens. • Consider every PSD recurrence or nonhealing as a potential PSDCA until proven otherwise.

Table 2. Recommendations for the early detection of PSDCA in patients with pilonidal sinus disease.
FNA, fine needle aspiration; PSD, pilonidal sinus disease; PSDCA, pilonidal sinus disease carcinoma

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General recommendations for every PSD patient

The surgeon should think about PSDCA in every patient who presents with pilonidal disease. In the history taking, the duration of the disease and the previous operations, if any, should always be investigated: the longer the duration of the disease, the higher the rate of malignant transformation [13]. Many alarming symptoms and signs were described in the literature, including continuous discharge, severe pain, rapid growth of the lesion, overgrowth beyond the level of the skin, superficial ulceration with friability, or external bleeding [14,15].

As described by Marjani, the typical findings on the physical examination include "… an indurated, friable, fungating mass with ulceration in the center, and multiple sinus openings with purulent discharge" [16]. The mobility of the ulcer or mass on the underlying structures, especially the sacrum, should be assessed [17]. Palpation of the inguinal lymph nodes and rectal digital examination should be a routine part of the examination in every patient who presents with pilonidal disease. Positive findings should be followed by further investigations such as inguinal ultrasonography or rectoscopy [18].

Additionally, the absolute need for treatment with the goal of complete healing should be discussed with every patient who presents with manifested PSD. Many reported PSDCA patients did not take the treatment of their pilonidal disease seriously, neglected their disease, refused radical operations, or did not show up for follow-up visits [19,20]. Therefore, negligent patients should be explicitly warned that aggressive cancer may develop if they do not get their chronic disease properly treated.

Detection before PSD resection

When the malignant transformation of the chronic disease is suspected clinically, histological diagnosis and tumor staging should be completed before any surgical intervention [3]. Multiple surgical or punch biopsies should be taken from the edges of the ulcerated lesion, as obtaining tissue from the necrotic areas can yield negative results [21]. The pathologist should be provided with adequate information about the high clinical suspicion so that the specimen is cut and examined properly. Negative findings or the presence of hyperplastic or dysplastic changes should prompt repeating the biopsy in highly suspected cases [3].

In the presence of inguinal lymphadenopathy at the initial presentation with a suspected PSD, a fine needle biopsy (FNA) should be obtained from the enlarged lymph nodes to differentiate reactive inflammatory lymph nodes from metastatic cancer. Compared with surgical biopsy, FNA is considered easier, cheaper, and safer [22]. Negative findings indicate reactive lymphadenopathy due to chronic inflammation, but the biopsy should be repeated in cases of high clinical or ultrasonographic suspicion [3].

Detection after PSD resection

Many authors challenged the traditional recommendation to submit all resected PSD specimens for histological examination. Boulanger et al. found no cases of malignancy after the examination of 731 PSD specimens. Consequently, they questioned the value of the routine pathological examination of all resected specimens. Alternatively, they recommended submitting the specimen to histology only in the presence of atypical clinical findings, prolonged disease, or patients older than 50 years [23]. Similarly,

Otutaha et al. did not detect any case of carcinoma in 320 PSD specimens. Based on these results, they also regarded the routine examination of specimens as unnecessary in patients younger than 50 years [24].

However, these studies provide a sense of security rather than suspicion. Dettmer et al. pointed out that more than 20% of all PSDCA patients are younger than 50 years, some are even in their twenties or thirties [25]. Additionally, the time interval from the first diagnosis of PSD until the development of carcinoma may not be necessarily long, as more than 18% of PSD carcinomas developed in less than ten years from the onset of the disease [5]. Moreover, many cases were discovered incidentally on histology and without any prior clinical suspicion or characteristics typical for malignant degeneration. This was explicitly described by Esposito et el. who reported a patient who "… did not clearly show these characteristics so that it was difficult to diagnose a carcinoma before the surgery" [26].

Therefore, the literature clearly shows that it is not possible to stratify the patients or determine when to perform a pathological examination based on characteristics like the age of the patient, duration of the pilonidal disease, clinical symptoms and signs, or preoperative suspicion. Consequently, we recommend sticking to the traditional surgical standards and submitting the resected PSD for histological examination in all cases to avoid missing unsuspected carcinomas. This practice should be stressed in developing countries that do not have healthcare insurance systems, where surgeons try to cut down costs by sparing the measures that are deemed unnecessary. Additionally, crisis-related shortcomings in medical care, as described in a recent report of 8 PSDCA cases [27], are one other reason for the increase in PSDCA due to prolonged exacerbated disease progression.

Lastly, practitioners should bear in mind that pilonidal disease recurrence or nonhealing may be one manifestation of missed or newly-developing carcinoma and the literature includes many such examples. García et al. were able to diagnose PSDCA after the third operation for recurrent PSD disease [28]. Alarcon-Del Agua et al. found cancer in a lesion that appeared only three months after the removal of a pilonidal cyst [29]. Michalopoulos et al. reported poor healing only 6 months after the resection of a chronic pilonidal sinus complicated by an anal fistula, where a subsequent radical surgical resection revealed the development of squamous cell carcinoma [11]. Accordingly, every PSD recurrence should be considered a potential PSDCA until proven otherwise.

CONCLUSION

Carcinoma originating on the background of pilonidal sinus disease is an aggressive tumour. While the prognosis is relatively poor in advanced stages, adequate therapy can be curative in early-detected cases. Due to the prevalence as well as the increasing incidence of pilonidal sinus disease, malignant transformation should be considered and excluded in every patient.

Based on the available literature, we suggested recommendations that can be applied on the initial approach of PSD patients, before PSD resection, and after PSD resection. We believe that every surgical resident and practitioner should know and endorse these recommendations, although more data is needed for validation. We also encourage the reporting of every case of PSD carcinoma to improve the understanding of this disease.

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Most of the listed recommendations are supposed to be feasible and applicable, while some limitations may arise in countries with limited resources. For example, insurance coverage may still limit the ability to submit all PSD-specimens for histological examination. Some diagnostic techniques, such as FNA, may not be available, but they can still be replaced with incisional or excisional biopsy. Due to the limited available data about pilonidal sinus carcinoma, future prospective research is absolutely needed for the critical validation of the recommendations.

ACKNOWLEDGMENTS
N/A

MULTIMEDIA
N/A

ETHICS
This study was conducted in accordance with the principles outlined in the Declaration of Helsinki, the analyses conducted in this study did not involve any interventions that could potentially cause harm to human participants. Written approval was obtained from the patients.

CONFLICT OF INTEREST
All authors declare that they have no conflicts of interest, and there are no relevant or minor financial relationships between relatives or next of kin and external companies.

FUNDING
No funding or grants were received from any other funding agencies in the public, commercial, or not-for-profit sectors.

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