

Research Article

A Clinicopathological Analysis of Umbilical Lesions in Adults

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Abstract

Background: The umbilicus is a location of clinical interest as several lesions are known to affect it. It may herald the existence of an internal malignancy.

Materials and Methods: Records of umbilical biopsies from January 2009 to December 2023 were retrieved and studied.

Results: There were 44 umbilical biopsies during the period of 15 years(2009- 2023) made up of 11 males and 33 females with a M:F ratio of 1: 3. The ages of the male patients ranged from 28-74 years with a mean age of 50 years, while that of the females ranged from 22-63 years with a mean of 38 years. The histology of the umbilical lesions revealed that benign lesions were 48% and the malignant lesions 52%.The colon was the most common primary site in 40% of the metastatic lesions.

Conclusion: Umbilical lesions were infrequent, most common in the females with the malignant lesions as the most prevalent. The malignant lesions were metastatic. Clinicians are urged to be aware of the significance of the umbilicus in view of the ease of clinical and histological evaluation.

Keywords: Adults; Benign, Malignant, Metastasis; Umbilical Biopsies

Introduction

The umbilicus may become a focal point of clinical interest as several lesions are known to affect it [1,2]. It is a depressed scar composed of a cushion with slightly inclined border and a cicatrix with an irregular base [2,3]. This scar immortalizes the site of prenatal attachment of the fetus to the mother [2]. Umbilical lesions are rare and can be non-neoplastic and neoplastic(benign or malignant) [4]. These lesions can also be caused by inflammation, infection or primary and metastatic neoplastic conditions [3].

Benign umbilical tumors can be classified as epidermal or dermal neoplasms [3]. The umbilicus is the most common cutaneous site of endometriosis the most common benign umbilical lesion [2]. This was first described by Dr Villar in 1886 and it is also called Villar nodule [1]. Cutaneous metastasis arising from carcinoma though rare are important findings in clinical practice. It may herald the existence of an internal malignancy [5,6]. The type of histological pattern can be a clue to the organ of origin [6]. The most common neoplastic lesion of the umbilicus is metastatic adenocarcinoma [7]. It is eponymously called Sister Mary Joseph nodule(SMJN) and often a signpost of an advanced primary abdominopelvic malignancy with the warning of a bad consequence. It is rare

and represents about 10% of cutaneous metastasis [2]. The benign appearance of umbilical lesions may be responsible for late presentation, furthermore the clinician may fail to diagnose the lesion due to its infrequent occurrence [8]. The differential diagnosis of umbilical lesions are diverse and may present a diagnostic challenge [1,2]. Imaging studies are not conclusive for diagnosis [1]. Histopathology with Immunohistochemistry(IHC) when relevant constitutes the gold standard for diagnosis including the location of the primary malignancy [1-6]. Due to the prognostic significance of umbilical lesions, accurate and correct diagnosis is important [2]. We present this study to highlight the demographic, clinical and pathological attributes of umbilical lesions towards the enhancement of the diagnostic resolution. Furthermore, to renew the interest of clinicians in the value and prognostic significance of umbilical lesions.

Materials and Methods

Records of umbilical biopsies with histologic diagnosis from January 2009 to December 2023 were retrieved and studied. The materials obtained were reprocessed to confirm the histology. Data analyzed included the patients' age, gender, clinical features, date of biopsy, histology, and site of the primary tumour where relevant. These were presented using tables and pie charts.

Results

There were 44 umbilical biopsy specimens reviewed during the period of 15 years(2009- 2023) made up of 11 males and 33 females with a M:F ratio of 1: 3, (Figure 1a), (Table 1). This translated to about 3 biopsies per year. The ages of the male patients ranged from 28-74 years with a mean age of 50 years, while that of the females ranged from 22-63 years with a mean of 38 years, (Table 1).

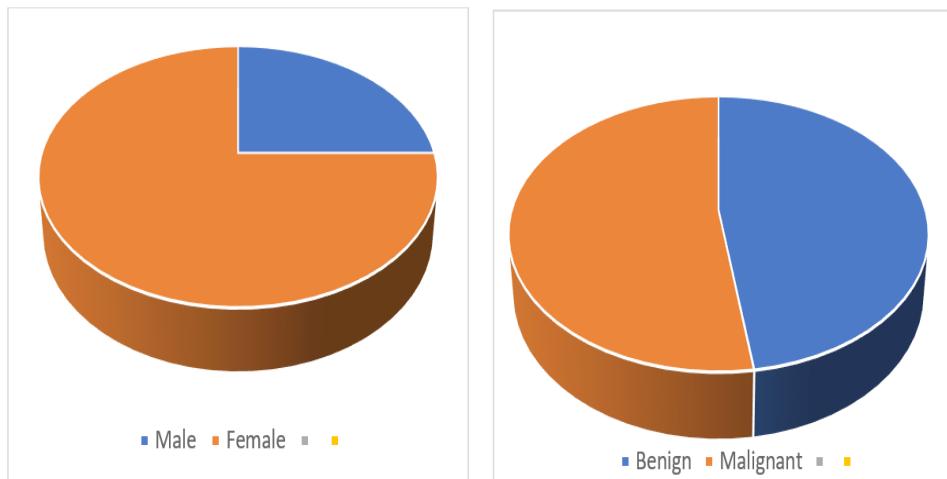


Figure 1: Figure 1a. Gender distribution of umbilical lesions- Male- 11(25%), Female- 33(75%). Figure 1b. Distribution of umbilical lesions. benign- 21(48%) and malignant lesions- 23(52%).

Age Range(yrs.)	Male	Female	Total	Percentage(%)
21-30	2	12	14	31.8
31-40	3	8	11	25
41-50	-	7	7	15.9
51-60	2	5	7	15.9
61-70	3	1	4	9.1
71-80	1	-	1	2.3
Total	11	33	44	100

- Males- Age range 28-74years, mean 50 years
- Females- Age range 22-63years, mean 38.0years

Table 1: Age/Sex Distribution of umbilical lesions.

All the patients presented initially with umbilical nodule or mass. The information on age, sex, clinical presentation were sufficient in some of the patients for appropriate clinical diagnosis, however, the definitive diagnosis was by histology of the umbilical biopsies. The history of cyclical painful umbilical swelling in catamenia was suggestive of endometriosis. These lesions were hyperpigmented and firm, (Figure 2a). Cyclical painful umbilical swelling in catamenia was found in a patient with a swelling on the umbilical scar of a patient that followed a previous umbilical hernia repair in the previous 2 years. The local features of the umbilical nodule were non-tender, firm, fissured, ulcerated, with occasional discharge, exuberant or exophytic mass. These findings in addition to other clinical findings pointed to the likelihood of metastatic umbilical nodule and the possible primary site. Some patients presented with a history of weight loss and intraabdominal mass located in the stomach or large bowel. A patient diagnosed with ovarian cancer on chemotherapy developed metastasis to the umbilicus, (Figure 2b). Three patients presented with metastatic umbilical nodules without evidence of the primary site of the malignancy. Figure 5 showed the algorithm for clinical evaluation of umbilical lesions.



Figure 2: Figure 2a- Clinical photograph of endometriosis. Figure 2b- Clinical photograph metastatic nodule on a distended abdomen with ascites.

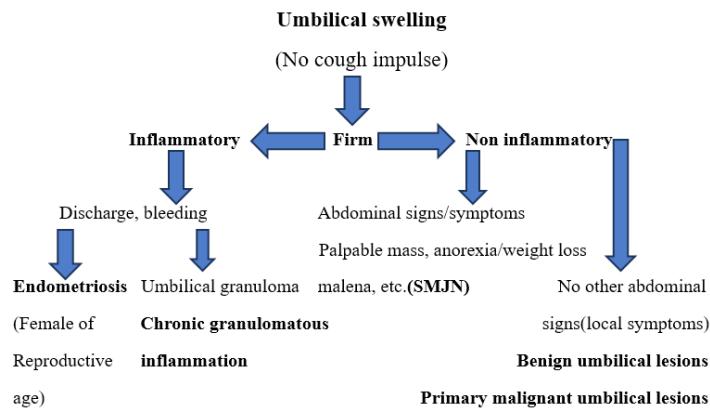


Figure 5: Algorithm for the clinical evaluation of umbilical lesions.

The histology of the umbilical lesions revealed that the benign lesions were 21(48%) and the malignant lesions 23(52%), (Figure 1b). The distribution of the benign lesions revealed the most common as endometriosis 11(52%), others were nonspecific inflammation 2(10%), chronic granulomatous inflammation 4(19%), trichilemmal cyst 1(5%), vitelline duct remnant 1(5%) and desmoid tumor 2(10%), (Figure 3a,4a,4b). A total of 21 patients comprised 4(19%) males and 17(81%) females with a M: F ratio of 1: 4.3 presented with benign umbilical lesions. In the male category their ages ranged from 28- 74 years with a mean age of 47 years, while the ages of the females ranged from 22-48 years with a mean age of 40 years. Endometriosis ranked first among the benign lesions with the peak age in the 3rd decade followed by the 4th decade, 55% and 36% respectively, (Table 2).

Age Range(yrs.)	Number	Percentage(%)
21-30	6	54.5
31-40	4	36.4
41-50	1	9.1
Total	11	100

Endometriosis, primary-10, secondary-1

Table 2: Age distribution of umbilical endometriosis.

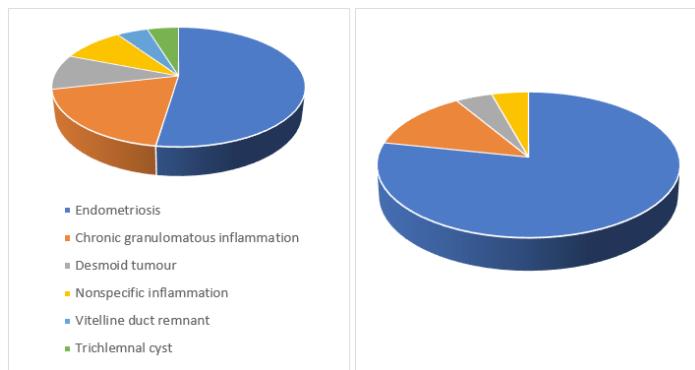


Figure 3: Figure 3a- Distribution of benign umbilical lesions- Endometriosis 11(52%), chronic granulomatous inflammation 4(19%), desmoid tumour 2(10%), nonspecific inflammation 2(10%), vitelline duct remnant 1(5%), Trichilemmal cyst 1(5%).

Figure 3b-Distribution of malignant umbilical lesions

- Metastatic adenocarcinoma 18(78%)
- Lymphoma 3(13%)
- Olfactory neuroblastoma 1(4%)
- Gastrointestinal stromal tumour 1(4%).

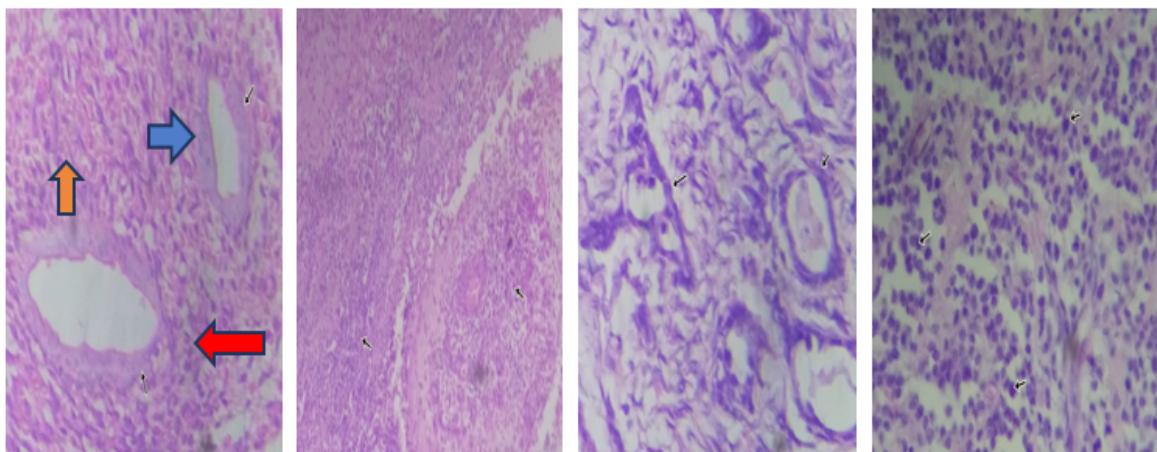


Figure 4: Figure 4a- Endometriosis, H&E x 100. Endometriosis- showing endometrial glands(blue arrow) and stroma(red arrow) in fibrocollagenous tissues(brown arrow). Figure 4b- Chronic granulomatous inflammation, H&E x 100. Chronic granulomatous inflammation showing intense mixed inflammatory infiltrates consisting of lymphocytes, plasma cells and a few polymorphs with areas fibrosis and necrosis. Figure 4c- Metastatic adenocarcinoma- H&E x 100, showing malignant irregular shaped glands infiltrating the stroma, lined by malignant epithelial cells. Figure 4d- non-Hodgkin lymphoma H&E x 100, section showed small mature lymphocytes scattered within the parenchyma. The cells have round nucleus with clumped chromatin and scanty cytoplasm.

The distribution of the malignant umbilical lesions revealed the most common as metastatic adenocarcinoma 18(78%). Others were lymphoma 3(13%), olfactory neuroblastoma 1(4%) and gastrointestinal stromal tumour(GIST) 1(4%), (Figure 3b,4c,4d). All the malignant umbilical tumours were secondary. Out of the 23 patients, 7(30%) were males while 16(70%) were females with a M: F ratio of 1: 2.3. Their ages ranged from 23- 68 years with a mean age of 47 years. The ages of the 16(69.6%) females ranged from 23- 63 years with a mean age of 44 years . In the male category 7(30%), their ages ranged from 36- 68 years with a mean age of 53 years, (Table 3). Table 4 shows the distribution of metastatic cancer and their primary organ in both male and female. Colon was the most common site of the primary tumour in both male and female and accounted for 7(39%). The stomach 4(23%) ranked second in both sexes. The ovary was the primary site in 4(22%) patients. The primary source of umbilical metastasis was not detected in 3(17%) of the patients(1- male,

2-females). There were 4 males and 7 males with benign and malignant lesions respectively. In the females, 17 lesions were benign and 16 malignant lesions, (Table 5).

Type	Male	Female	Total(%)
Metastatic adenocarcinoma	5	13	18(78.2)
Lymphoma	1	2	3(13)
Olfactory neuroblastoma	1	-	1(4.4)
Gastrointestinal stromal tumour	-	1	1(4.4)
Total	7	16	23(100)

*23 patients, age range- 23-68 years, mean age- 47 years

16 females, age range- 23-63 years, mean age- 44 years

7 males, age range- 36-68 years, mean age- 53 years

Table 3: Gender distribution of malignant umbilical lesions.

Site	Male	Female	Total(%)
Stomach	2	2	4(22.2)
Colon	4	3	7(38.9)
Ovary	-	4	4(22.2)
Uterus	-	-	-
Unknown primary	1	2	3(16.7)
Total	7	11	18(100)

Table 4: Gender/site distribution of umbilical metastatic adenocarcinoma.

Histological type	Male	Female	Total(%)
Benign		4	17
Malignant		7	16
Total	11		44(100)

Table 5: Histological type/Gender distribution of umbilical lesions.

Discussion

Umbilical lesions are rare. In this study, a total of 44 umbilical biopsies were evaluated in 15 years with an average of 3 biopsies per year. This is in keeping with the report from Onyishi and Okafor [2] from Nigeria. In another study, Yan et al reported 99 umbilical lesion in 23 years with an average of 4.3 biopsies per year [4]. Our study revealed that the benign lesions were 48% and the malignant lesions 52%. A similar result(benign- 47% and malignant- 53%) was reported by Das and Manda [9]. This was like the report from Yan et al [4] that showed 40(40.4%) as benign and 59(59.6%) as malignant [4]. Similarly, another report from Nigeria showed benign 25(37.3%) and malignant 42(62.7%) [2]. Benign tumours were commoner in the females with a M: F of 1: 4.3. In the male category, the mean age was 47 years and the female the mean age was 34 years. This was due to endometriosis that ranked first among the benign lesions and affecting females of reproductive age. However, in the report by Yan et al, epidermoid cyst was the most common benign lesion followed closely by endometriosis [4]. The authors further reported a M: F ratio of 1:3 with the mean ages 52 and 43 years in the males and females respectively [4]. The malignant lesions in our report were most common in the females with a M: F ratio of 1: 2.3. Yan et al reported a similar experience [4]. Contrarily, Onyishi and Okafor recorded almost an equal male to female preponderance [2]. A report from the Netherlands showed that umbilical metastasis in males were mostly of colonic origin whereas the females were ovarian and 15-30% of cases turn out to be metastasis from unknown primary [8]. The umbilicus is an anatomical site located at the center of the anterior abdominal wall that presents with a variety of diseases: developmental, endometriosis, benign neoplasms, primary and metastatic cancers among others [2,3]. This is because the umbilicus is located adjacent to the anterior peritoneum and it communicates with a variety of solid organs in the abdomen and pelvis through a myriad of vascular and lymphatic drainage pathways [3]. The umbilicus can be easily evaluated during clinical examination [3]. Umbilical lesions are classified as non-neoplastic(inflammatory or infective) and neoplastic.

The latter is further subdivided into benign and malignant. The malignant lesions can be classified as primary and metastatic. Benign lesions of the epidermis include epidermal cyst, epidermal naevi and verrucae vulgaris. Those of dermal origin comprised of dermatofibroma, hypertrophic scars, keloids, neurofibroma and fibroma [3]. Others include umbolith, umbilical granuloma, umbilical hernia, papilloma, miliary tuberculosis and endometrial deposits [8]. The pathophysiology of spontaneous or primary Umbilical Endometriosis(UE) is unknown. Multiple theories have been advanced but the most accepted are the implantation of endometrial tissue through retrograde menstruation(implantation theory), hematological or lymphatic dissemination of endometrial cells (Dissemination theory) and differentiation of pluripotential peritoneal progenitor cells into endometrial tissue(coelomic metaplasia theory) [1]. Symptoms characteristic of primary UE are cyclical pain and palpable mass in women of reproductive age related to catamenia. Imaging studies are not conclusive for the diagnosis. Definitive diagnosis is based on histology that demonstrated endometrial glands, stroma and hemosiderin pigments. The standard modality of treatment is surgery [1]. Benign lesions shown in this study revealed the most common as endometriosis 52%, others were nonspecific inflammation, chronic granulomatous inflammation, desmoid tumor, trichilemmal cyst, vitelline duct remnant.

The mechanism of metastasis is not fully understood but potential contributory factors included: extensive vascular connections with the upper and lower trunk of the body(venous, lymphatic and arterial); ligamentous remnants of embryological structures; and proximity to abdominal viscera. Also, likely to be important are the intrinsic tumor characteristics and the umbilical skin microenvironment might cooperate to encourage tumor homing [2]. Presentation is classified into the following settings: those that develop after the initial diagnosis of the primary lesion, those detected at the same time of diagnosis [5] and carcinoma of unknown primary site(CUPS). In the later, IHC, provides a useful adjunct in directing the search of the primary tumor. Three patients in our report presented as CUPS. SMJNs are firm to hard, irregular, painless mass fixed to the skin. The size may vary from a few centimeters to greater than 5-7cm. The surface may be fissured, ulcerated, exuberant without or occasional discharge [8]. Ulcerative changes on the surface of the mass may be an indication of necrotic degeneration from rapid growth in a malignant tumor [3]. Sometimes they may mimic umbilical hernia as the diagnosis may be missed by the clinician [9]. Abdominopelvic imaging may detect primary lesions and tissue biopsy is required for definitive diagnosis [8]. The algorithm for clinical evaluation of umbilical lesions based on the study is presented. The pattern of histology revealed that the malignant lesions were more likely to be found in the males, while the distribution in the females did not reveal any significance difference. Sister Mary Joseph Nodule(SMJN) refers to an umbilical nodule that represents a cutaneous metastasis where the primary lesion is found in the abdomen and pelvis. It is

found in 1-3% cases of abdominopelvic malignancies [8,9]. Our study revealed the most common malignant lesion as metastatic adenocarcinoma 78%. Lymphoma ranked second(13%), olfactory neuroblastoma 4% and gastrointestinal stromal tumour 4%.

The distribution of metastatic adenocarcinoma 78% showed that the colon was the most common site of the primary tumour in both male and female and accounted for 39% of the lesions. The stomach 22% ranked second in both sexes. The ovary 22% was the primary site in the gynecological tract. The primary source of umbilical metastasis was not detected in 17% of the patients. All the malignant tumours were secondary like the report from Yan et al [4]. In their report, gynecological cancer(48%) was the most common primary site. Others were gastrointestinal and pancreatic(32%), lymphomas(8%), breast, prostate and melanoma 4% each [4]. In a report by Onyishi and Okafor [2], gynecological organs were the most common primary source of metastatic malignancy to the umbilicus in the females, while the gastrointestinal tract was the most common in the males. Das and Manda reported that in SMJN, gastrointestinal malignancy 35-65% was the most common primary, genitourinary metastasis comprised 12-35% with primary umbilical tumors found in 3-6% of cases [9]. Furthermore, they reported gynecological ovarian cancer was the most common primary cancer to metastasize to the umbilicus 34% [9]. Skin metastasis in ovarian cancer is rare. Ovarian cancer is a very common primary malignancy in postmenopausal women aged 50-70 years [9]. Lymphoma ranked second 13%, metastatic tumor to the umbilicus referred to as SMJN is very rare from lymphoma. Lymphomas, historically seem to reject the umbilicus as a metastatic site. The first report of lymphoma presenting as SMJN was by Chagper and Carter [10]. Our study revealed a SMJN from GIST, this presentation is rare and had been earlier reported by Kundu et al as umbilical metastasis from an initial presentation of GIST diagnosed by cytology and IHC following the aspiration of the umbilical lesion [7]. All the malignant umbilical lesions in our report were metastatic. In another report with primary umbilical malignancy, the lesions were all skin cancers(melanomas, squamous cell carcinoma and basal cell carcinoma) [2].

Conclusion

Umbilical lesions were infrequent. The lesions were more common in the females than the males, in addition malignant lesions were the most common. In the benign lesions, umbilical endometriosis was the most prevalent in women of reproductive age. In the males, malignant lesions(SMJNs) were the most common umbilical lesions. All the malignant tumors were metastatic and the most common metastatic tumour was adenocarcinoma with the abdominopelvic region as the primary site . The gastrointestinal tract in both males and females and gynecological tracts in postmenopausal women were primary locations for SMJN. Clinicians are urged to be aware of the significance of the umbilicus in view of these attributes, ease of clinical and histological evaluation.

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