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Case Report

From a Congenital Defect to Cancer: A Case of Squamous Cell Carcinoma in a Neglected Myelomeningocele

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Neural tube defects are common congenital anomalies, typically presenting early due to visible swelling and/or neurological deficits. Rarely, cystic swellings are neglected until adulthood, with only 14 cases of malignancy developing in an untreated meningomyelocele reported to date. We describe the case details of a 26-year-old Indian woman with this rare complication. Magnetic resonance imaging revealed a low-lying spinal cord with spinal dysraphism, cord herniation, and a cystic lesion. The biopsy confirmed a well-differentiated squamous cell carcinoma. Malignant transformation in an untreated myelomeningocele is rare, with chronic irritation and infection as proposed causes. Early biopsy and treatment are crucial for its management.

Key words: squamous cell carcinoma, meningomyelocele, occult spinal dysraphism

Neural tube defects (NTDs) are common congenital anomalies, with a reported incidence in India ranging from 0.5 to 18.2 per 1,000 total births [1]. Patients with an NTD typically present early due to visible swelling and/or neurological deficits. It is unusual for cystic swelling on an individual's back to go unnoticed and untreated until adulthood. Our literature search identified only 14 adult cases of squamous cell carcinoma that had developed in an untreated myelomeningocele, worldwide [2-15]. Here we describe the case of an adult female who experienced this uncommon complication of a common congenital abnormality.

Case Report

Patient history. A 26-year-old Indian woman presented to the neurosurgery outpatient department with a longstanding ulcerated midline swelling in her lower back (Fig. 1). This lemon-sized swelling had been present since birth and was skin-covered and roughly round; it intermittently discharged yellowish-watery fluid from a small opening. The patient had a history of bilateral congenital talipes equinovarus with limb-length discrepancy since birth (Fig. 2), with no evidence of trophic ulceration. She had experienced urinary incontinence since the age of 10. Despite medical advice to undergo surgery for this swelling, she declined it. Over the years, small, painful ulcerations occasionally appeared on the swelling but healed with local wound care. During the 2 years before her presentation, the ulcer stopped healing, expanded in size, spread beyond the swelling's margins, and began producing a purulent discharge. It gradually became painless.

Physical examination. On examination, a 5×5 cm ulcer was observed over the lumbosacral region in the midline, with everted edges and a base covered in yellowish slough. The surrounding skin was darkened and slightly indurated. Notably, the ulcer was painless and showed no cerebrospinal fluid (CSF) leakage.

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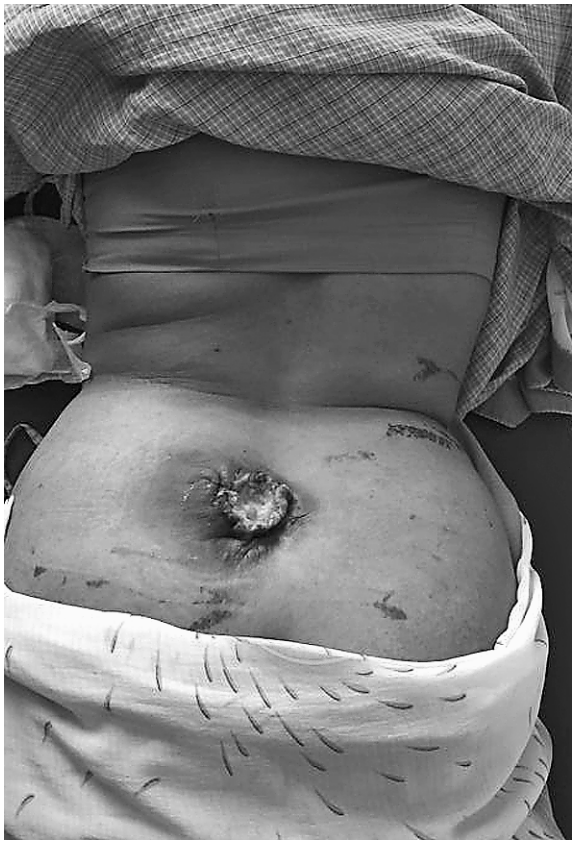

Fig. 1 The ulcer over the patient's lumbosacral region in the midline, with everted edges. The ulcer's base was covered in yellowish slough with dark and indurated surrounding skin.

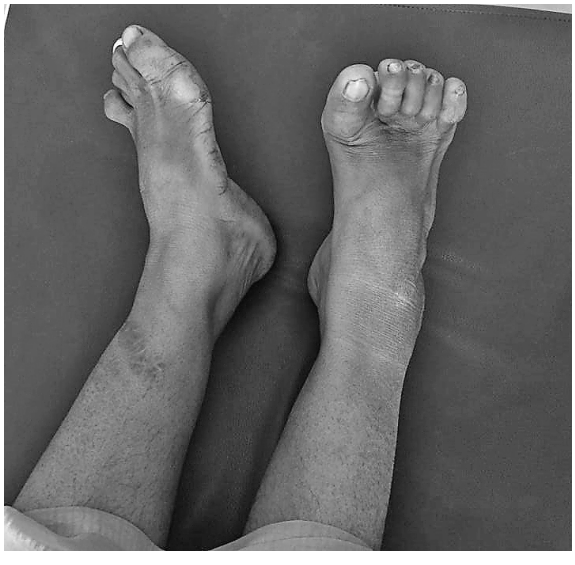

Fig. 2 Bilateral congenital talipes equino varus with limb-length discrepancy in the patient, a 26-year-old woman.

Neurological examination. A neurological examination revealed significant weakness in the foot dorsiflexors and plantar flexors and reduced sensation in the 5th lumbar (L5) and sacral 1 to 3 (S1-S3) dermatomes. Anal tone and perineal sensation were also diminished.

Imaging findings. Magnetic resonance imaging (MRI) revealed a low-lying spinal cord with spinal dysraphism at the S1 vertebra and cord herniation, with CSF observed through this defect, which was approx. 8.8 mm long (Fig. 3). A well-defined oval cystic lesion approx. $3 \times 2.2 \times 1.9$ cm in size and exhibiting CSF signal intensity reached the subcutaneous plane at the S1-S2 level. This lesion appeared hypointense on T1-weighted images, hyperintense on T2-weighted images, and showed no contrast enhancement. Additionally, mild widening of the sacral canal was noted.

Beyond the myelomeningocele, an ill-defined, heterogeneously enhancing soft tissue lesion measuring 5.18×3.54 cm was observed in the subcutaneous plane, extending to the skin with associated ulceration. No signs of calcification or hemorrhage were present. The MRI findings suggested sacral spina bifida, cord tethering, and a posterior myelomeningocele protruding into the subcutaneous plane at the S1 level. The nature of the enhancing soft tissue lesion raised questions regarding a potential neoplastic or inflammatory etiology.

Biopsy findings. The patient underwent a biopsy of the ulcer's edge. The histological examination of the tissue sections stained with hematoxylin and eosin (H&E) revealed tumor cells arranged in sheets and nests, with keratin pearl formation, which is consistent with a well-differentiated squamous cell carcinoma (SCC) (Fig. 4). Under high magnification, the tumor cells were notably enlarged, displaying a high nuclear-to-cytoplasmic ratio and prominent nucleoli. These characteristics strongly indicated the presence of squamous cell carcinoma.

Treatment plan. The patient was scheduled for a metastatic evaluation, lesion excision, and excision of the entire dermal sinus tract followed by adjuvant therapy. However, she did not consent to the proposed interventions and was subsequently lost to follow-up.

Discussion

A meningomyelocele is a congenital abnormality characterized by the protrusion of the spinal meninges (which carry CSF) together with the protrusion of neu-

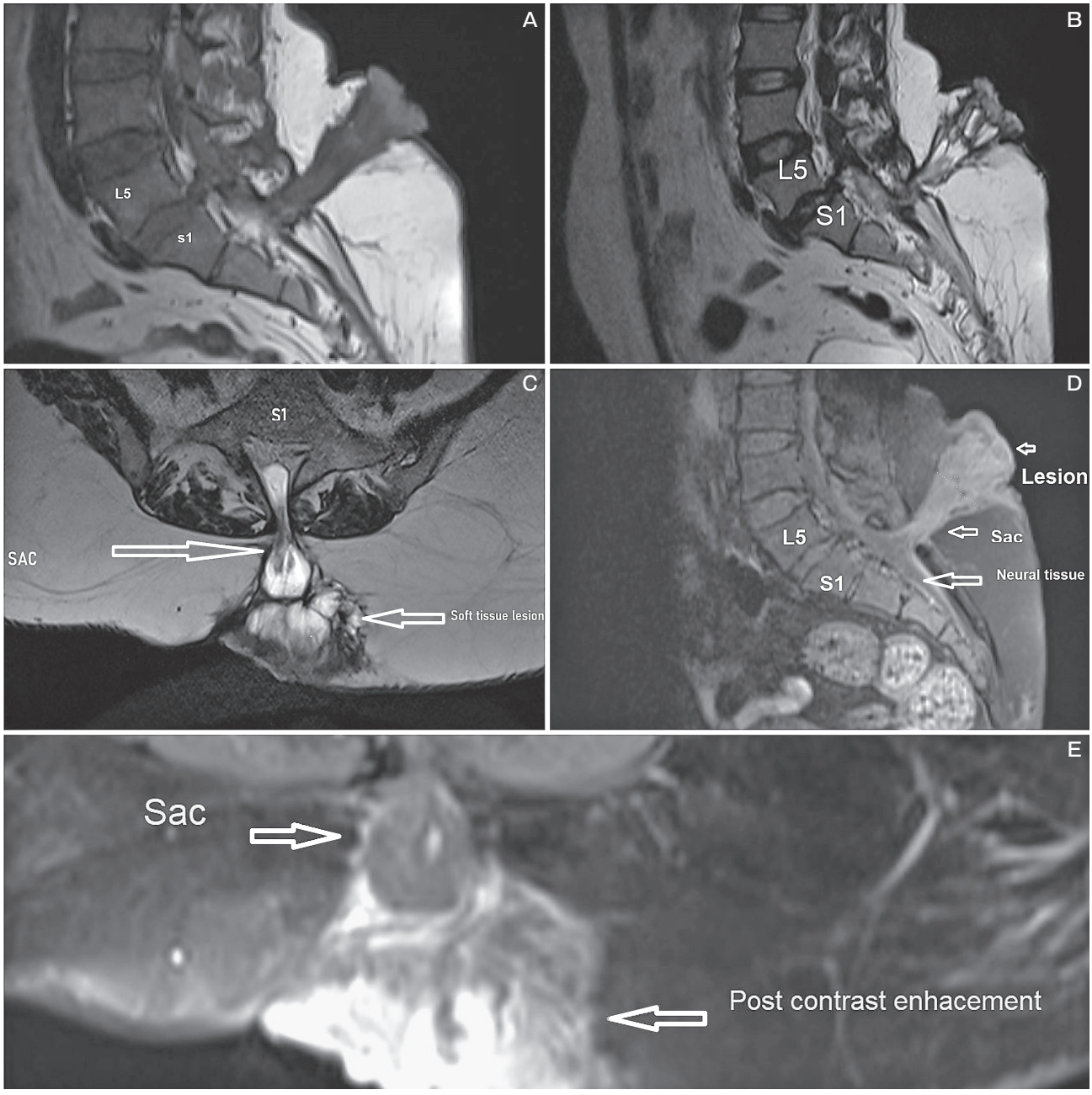

Fig. 3 **A**, T1WI sagittal section showing the spinal defect at the S1 vertebrae level with a cerebrospinal fluid (CSF) filled sac with meninges and neural tissue protruding into the subcutaneous plane; **B**, T2WI sagittal section showing the CSF sac protruding from the vertebral defect at the S1 vertebrae level; **C**, T2WI axial section at the S1 vertebrae level with the CSF-filled sac (yellow arrow) protruding into the subcutaneous plane with meninges and neural tissue. This hyperintense multiloculated lesion was observed in the subcutaneous plane, posterior to the sac (orange arrow); **D**, T1W1 sagittal contrast visualized the enhancing soft tissue lesion (orange arrow) in the subcutaneous plane; adjacent to it is the hypointense sac with meninges and neural tissue (yellow arrow). The spinal cord is low-lying and likely a tethered cord (blue arrow); **E**. T1WI post-contrast sections depicting the enhancing subcutaneous soft tissue lesion (orange arrow). However, the CSF-filled sac with meninges and neural tissue (yellow arrow) did not show any obvious post-contrast enhancement.

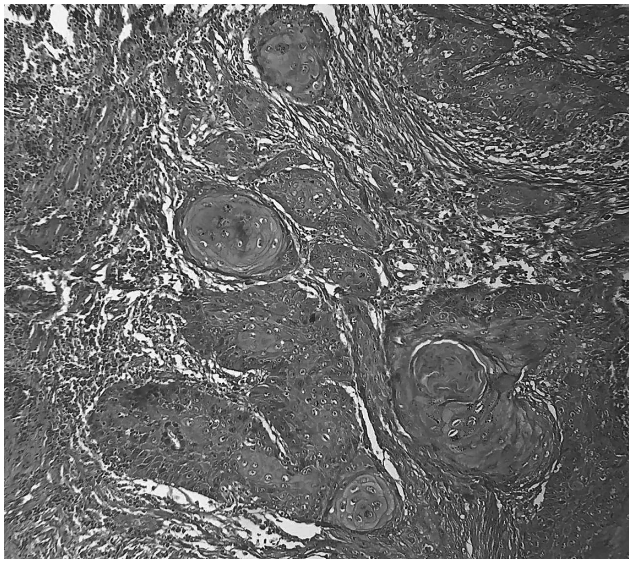

Fig. 4 Tumor cells grouped in sheets and nests with keratin pearl formation.

ral tissue. It occurs when the neural arches fuse in conjunction with an open NTD [11]. Dermal sinus and lipoma may both be present in conjunction [14].

The occurrence of skin carcinoma associated with a meningomyelocele is rare, with few cases in the literature. We identified 14 cases of squamous cell carcinoma that arose in adults with a meningocele or myelomeningocele (Table 1) [2-15]. Most of the reported cases described associated ulceration and discharge or fistula tracts at the site of the meningomyelocele defect. Malignancies at these sites may result from chronic mechanical irritation and infection. Among the 14 cases, 12 were squamous cell carcinoma, one was anaplastic carcinoma, and another was a mucin-secreting adenocarcinoma. Erhamamci *et al.* used fluorodeoxyglucose-positron emission tomography/computed tomography (FDG-PET/CT) for the detection and localization of the primary tumor and for metastatic evaluation [12]. Notably, Duan *et al.* reported the

Table 1 Summary of clinical presentations in reported cases of malignancy linked to myelomeningocele[15]

Author	Year	Age	Sex	Management	Histopathology	Outcome	Site	Sinus/Fistula
Thorp[2]	1967	26	M	Surgery + RT	Anaplastic Carcinoma	Expired 5 days after second surgery	Lumbar	+
Pope & Todorov[3]	1975	37	M	Surgery	SCC	Uneventful at discharge	Cervical	?
Banerjee[4]	1977	20	F	Biopsy + RT	SCC	Retroperitoneal metastasis	Lumbosacral	−
Saksun & Fisher[5]	1978	25	M	Antibiotics + RT	SCC	Expired due to meningitis	Lumbosacral	Purulent discharge
Marini et al.[6]	1979	41	M	Surgery + RT	Epidermoid Carcinoma with Inguinal lymph node metastasis	Recovered	Lumbar	+
Tintelen et al.[7]	1980	50	M	Surgery	Epidermal Carcinoma	Death in 9 weeks		?
Chadduk and Uthman[8]	1984	29	F	Surgery	SCC	Uneventful at discharge	Lumbar	Present for first 2 years
Lee and Jayakrishnan[9]	2008	62	F	Surgery	Mucin secreting adenocarcinoma	Uneventful at discharge	Sacral	−
Duan et al.[10]	2009	11	M	Surgery	SCC	Uneventful at discharge	Lumbosacral	Serosanguinous discharge
Wani et al.[11]	2016	60	M	Surgery + RT	SCC	Uneventful at discharge	Sacral	−
Erhamamci et al.[12]	2016	26	F	Biposy with FDG-PET CT	SCC		Lumbosacral	Foul discharge
Goyal et al.[13]	2018	21	M	Surgery	SCC	Uneventful	Lumbar	No
Kurt- ozkaya[14]	2019	22	F	Biopsy	SCC	Death in 20 days	Lumbar	Foul discharge
Mahindra et al.[15]	2020	18	M	Surgery + RT	Leiomyosarcoma	Uneventful	Cervical	Foul discharge
Our Case	2023	26	F	Biopsy	SCC		Sacral	Foul discharge

SCC, Squamous Cell Carcinoma; RT, Radiotherapy;
FDG-PET CT, fluorodeoxyglucose-positron emission tomography/computed tomography

youngest case of malignant transformation in an 11-year-old child [10].

In cases of an untreated meningomyelocele (especially in the presence of discharging dermal sinus), there is a risk of neoplastic change after years of mechanical irritation and chronic bacterial infection [16]. This appears to be a likely cause in the present patient. A defect that lacks a protective epithelial cover must be viewed with a high degree of suspicion, just like other chronic ulcers. To establish a histological diagnosis, a biopsy should be performed as soon as a change is detected. If carcinoma is discovered, a thorough search for metastases, lymph node involvement, and local invasion is necessary for proper staging and treatment.

Conclusion

The development of carcinoma in an untreated myelomeningocele is a very rare complication. Early recognition and surgical excision may be life-saving.

Patient Consent Declaration. The patient provided written informed consent for this report and the publication of the images.

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