

The symptoms of SMD vary depending on the age of the patients<sup>(1)</sup>, the principal symptoms being as follows: limited postnatal growth; rhizomelic shortening of the limbs in early childhood evolving to shortening of the trunk by the age of 10 years; thoracic hypoplasia, which causes respiratory problems in the neonatal period and increases susceptibility to respiratory tract infection<sup>(4)</sup>; scoliosis with dorsal kyphosis; abnormalities of the metaphyses and pelvis<sup>(5)</sup>; odontoid hypoplasia; and valgus of the knees and claudication<sup>(6)</sup>, the latter typically being the first sign of the disease<sup>(2)</sup>. There might be little or no ossification of the cervical vertebrae, leading to cervical instability and swan neck deformity<sup>(7)</sup>.

A review of the literature revealed that there are currently 10 recognized subtypes of SMD. However, there is no consensus in the medical literature regarding those subtypes, because they are based on characteristics that are minimally different. Some subtypes are based on reports of only one case, and others can be diagnosed only after years of follow-up, which is difficult. For example, the longest follow-up period in a report of Sedaghatian-type SMD was 161 days. Therefore, there is no acceptable standard for subclassifying the disease.

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Multiple primary malignancies: synchronous urothelial carcinoma of the bladder and adenocarcinoma of the colon

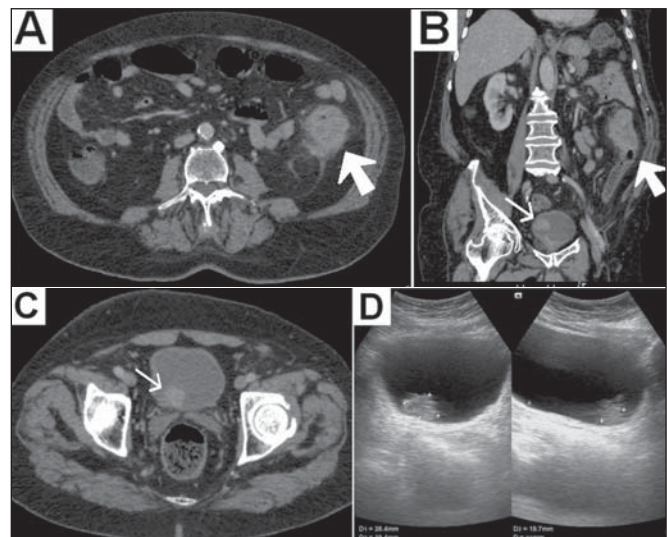
Dear Editor,

A 75-year-old White male presented with a three-month history of pain in the left hypochondrium. The patient also reported experiencing an episode of gross hematuria six months prior. He had quit smoking 20 years prior, having previously smoked 30 cigarettes/day for 30 years. He had also undergone surgery for a gastric ulcer 20 years prior. He reported no other comorbidities.

Computed tomography of the abdomen showed a solid, irregular, concentric mass, which was expansive and stenotic, in the middle third of the descending colon (Figures 1A and 1B). The mass showed heterogeneous uptake of the intravenous iodinated contrast medium and increased density of adjacent fat tissue, suggesting that it had expanded through the serosa. In addition, a vegetative lesion, with irregular borders and showing contrast enhancement, was observed in the right posterolateral wall of the bladder (Figures 1B and 1C).

Colonoscopy with biopsy of the intestinal mass led to a histological diagnosis of moderately differentiated adenocarcinoma of the colon, and the patient was therefore submitted to segmental colectomy with colostomy. The anatomopathological study revealed a hard, annular tumor, which was ulcerative and vegetative, infiltrating the intestinal wall and surrounding fat, thus confirming the result of the microscopy study of the biopsy. Subsequently, ultrasound of the urinary tract confirmed bladder nodulation (Figure 1D), with no perceptible flow on color Doppler. Complete transurethral resection of the nodulation was performed, and histopathological analysis of the resected specimen led to a diagnosis of superficial low-grade papillary urothelial carcinoma (World Health Organization grade I). A subsequent computed tomography scan of the abdomen and pelvis, for staging, showed no suspicious lesions. The final diagnosis was multiple, synchronous primary malignancies, probably secondary to smoking.

Colon cancer is the fourth most common malignancy in men, accounting for 90% of the cases that occur after the fifth decade of life, adenocarcinoma being the most common type<sup>(1)</sup>.



**Figure 1.** A,B: Computed tomography scan of the abdomen, obtained in the portal phase after intravenous administration of contrast medium, in an axial view (A) and oblique coronal reconstruction (B), showing a solid, irregular, concentric mass, which was expansive and stenotic, in the descending colon, presenting heterogeneous enhancement, together with increased density of the adjacent fat tissue (large arrow). Note also the vegetative lesion, with irregular borders and showing contrast enhancement (small arrow in B). C: Axial computed tomography slice, obtained in the portal phase after intravenous administration of iodinated contrast medium, showing the vegetative lesion, with irregular borders, located in the right posterolateral wall of the bladder (arrow). D: Abdominal ultrasound, confirming the lesion in the bladder wall.

In 5–10% of cases, adenocarcinoma is associated with hereditary syndromes (e.g., familial adenomatous polyposis, hereditary non-polypoid colorectal cancer, etc.), especially in young adults<sup>(1)</sup>. It is related to obesity, a sedentary lifestyle, a diet low in fiber, and inflammatory bowel diseases<sup>(1–4)</sup>. Smoking and alcoholism can also play roles<sup>(2–4)</sup>.

Bladder cancer, which is the most common type of malignant neoplasia of the urinary tract, affects individuals 55–60 years

of age, 75–80% of whom are men, urothelial carcinoma being the predominant form<sup>(5,6)</sup>. Urothelial carcinoma can be multifocal/multicentric, can occur in the upper or lower urinary tract, and is often recurrent<sup>(5)</sup>. Smoking is implicated in 50–65% of all cases in men and in 20–30% of all cases in women<sup>(4)</sup>. Other, less common causes include chemotherapy, exposure to aromatic or heterocyclic amines, radiotherapy, and chronic infection<sup>(2,4–6)</sup>.

Multiple primary malignancies are defined as those that are confirmed, independent, and of non-metastatic origin<sup>(7)</sup>. They are classified as synchronous if they are identified within the first six months after the appearance of the first lesion or as metachronous if they are identified thereafter<sup>(7)</sup>.

The overall prevalence of multiple primary malignancies is 0.7–11.7%, increasing proportionally with patient age<sup>(2,3,7,8)</sup>. It is estimated that 75% of cases occur in individuals over 50 years of age<sup>(7)</sup>. These values are on the rise due to the effectiveness of treatments, the variety of therapeutic techniques now available, the improvement of diagnostic methods, the increased longevity of the population, and contemporary lifestyles<sup>(3,7)</sup>. Hayat et al.<sup>(2)</sup> reported a probability of developing a second malignancy, depending on the primary tumors diagnosed, ranging from 1% (history of hepatic neoplasia) to 16% (previous bladder tumors)<sup>(2)</sup>. Braisch et al.<sup>(4)</sup> observed that 1.2–2.5% of cancer patients who were smokers developed another distinct malignant lesion within the first year of follow-up.

In smokers, multiple primary malignancies can affect several organs, notably the lungs, upper aerodigestive tract, and kidneys, as well as the upper and lower urinary tract. Other potential sites include the thyroid gland, stomach, colon, rectum, and pancreas<sup>(4,6,8)</sup>.

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Dural fistula with bilateral arterial supply, mimicking a brainstem tumor

Dear Editor,

A 73-year-old woman presented with a history of at least four episodes of deep vein thrombosis. In the last five months, she had experienced severe ataxia, difficulty in swallowing, bilateral tinnitus, and symptoms related to intracranial hypertension, such as nausea and vomiting. Magnetic resonance imaging (MRI) revealed a hyperintense signal on T2-weighted images and an enlarged brainstem, the swelling extending to the thalamus, cerebellar peduncles, and to the cervical portion of the spinal cord (Figures 1A and 1B). The images could erroneously indicate a diagnosis of brainstem tumor, glioma in particular, due to the infiltrative pattern of the lesion and the increased organ volume. However, thorough evaluation with advanced imaging techniques, such as magnetic susceptibility-weighted sequences, demonstrated an extensive network of dilated peripheral veins, together with pronounced collateral circulation. Cerebral angiography showed a dural arteriovenous fistula (DAVF) with bilateral arterial supply via branches of the maxillary arteries. Venous drainage was mostly through the rectum and galenic system (Figures 1C and 1D). Involvement of the brainstem and cervical spinal cord was due to venous congestive injury. The classical surgical approach was precluded by the deep, inaccessible location, whereas endovascular therapy was precluded by the extensive involvement and bilateral nature of the fistula-sustaining arterial supply. The patient underwent gastrostomy and was discharged to palliative home care.

Vascular lesions are often difficult to diagnose<sup>(1–8)</sup>. DAVFs, which are characterized by abnormal communication between

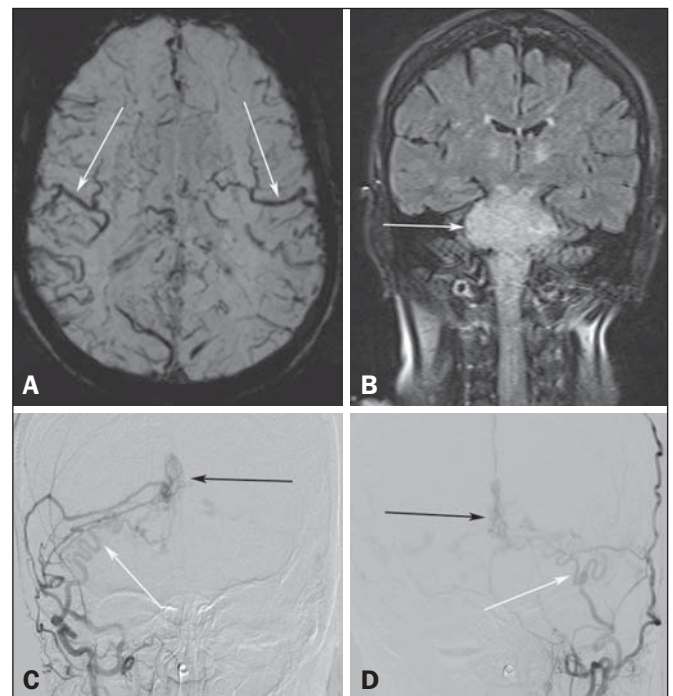


Figure 1. A: Axial slice in a susceptibility-weighted sequence showing numerous large-caliber superficial veins, representing venous congestion. B: Coronal slice in a fluid-attenuated inversion recovery sequence showing a hyperintense signal and increased brainstem volume, mimicking a brain tumor. C,D: Digital angiography with subtraction technique, revealing the bilateral nature of the arterial supply (white arrows) and the nidus (black arrows) formed by the fistula.