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Clear Cell Sarcoma-Like Tumor of the Gastrointestinal Tract: Case Report and Review of the Literature

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1. Abstract:

The rare and aggressive tumor known as gastrointestinal clear cell sarcoma (GICCS) mostly affects the digestive system. framework. There have been noticeably fewer cases of conventional clear cell sarcoma (CCS) of soft parts arising in the gastrointestinal tract as compared to GICCS. We discuss the case of a 28-year-old man who had liver metastases in addition to a big abdominal tumor. Although the diagnosis of this patient was originally complex and tough, the right diagnosis of GICCS was made by the appropriate use of immunohistochemistry (IHC) and molecular research. A review of the literature reveals that GICCS is also referred to by other names, such as gastrointestinal clear cell sarcoma/ gastrointestinal neuroectodermal tumor GICCS/GNET and clear cell sarcoma-like tumor of the gastrointestinal tract (CCSLGT). Often, it's challenging to tell if the rare and aggressive tumor known as gastrointestinal clear cell sarcoma (GICCS) mostly affects the digestive system.

There have been noticeably fewer cases of conventional clear cell sarcoma (CCS) of soft parts arising in the gastrointestinal tract as compared to GICCS. Due to their similarities, liver cases have been reported to represent one or the other of these tumors. It has been discussed if they could represent variations of the same entity, which is what we believe to be the case. We describe a case of a 28-year-old man who arrived with a big abdominal mass. Clear cell sarcoma of the gastrointestinal tract is referred to in this study as "CCSLGT," to distinguish it from other body regions. We go over and define the terms used to explain this. kind of sarcoma. Additionally, we describe the distinct presentation, diagnostic assessment, and therapeutic strategy of a patient with gastrointestinal clear cell sarcoma. This paper emphasizes the difficulties in recognizing this uncommon tumor and the significance of taking it into account in the differential diagnosis of gastrointestinal malignancies through a review of the literature. Enzinger et al. originally reported on clear cell sarcoma (CCS), formerly known as melanoma of soft parts. It is an uncommon, aggressive malignancy that typically appears in the deep soft tissues.

1. Clear cell sarcomalike tumors of the gastrointestinal tract (CCSLGT) is the new term for clear cell cancers that arise in the digestive system. There have been noticeably fewer cases of CCS developing in the gastrointestinal system as compared to CCSLGT. Ekfors et al. originally reported a case of CCS developing in the duodenum. Since then, compared to CCSLGT, significantly fewer reports of CCS developing in the gastrointestinal system have been documented.

2,3 Although CCSLGT lacks immunohistochemistry (IHC) reactivity for melanocytotic markers, it does have certain histological similarities. Attributes of CCS. Our understanding of the composition of CCS has improved due to recent advancements in molecular biology techniques. The majority of CCS patients, according to studies, have chromosomal translocations that include the fusion of the EWSR1 gene on chromosome 22q12 with the ATF1 or CREB genes on chromosome 12q13 or 8q34, respectively. The fact that these translocations are also seen in CCSLGT suggests that these two entities may be closely related.
A wide differential diagnosis that includes carcinoid, schwannoma, gastrointestinal stromal tumors, neuroectodermal neoplasms, metastatic melanoma, alveolar rhabdomyosarcoma, primary or metastatic conventional clear cell sarcoma, gastrointestinal stromal tumor (GIST), and synovial sarcoma is made more difficult by the rarity of CCSLGT. EWSR1-CREB1 gene fusions are also linked to CCSLGT, which frequently consists of large cells with variable degrees of by different intensities of anemia, nausea, vomiting, and weight weariness and loss.
4. anemia, bowel obstruction, and rectal the tumor may present with non-specific symptoms such as bleeding, nausea, widespread stomach discomfort, and weight loss.

5. Surgical resection is the principal therapeutic technique used to achieve full tumor excision in CCSLGT treatment modalities, which are still largely non-standardized.
6 The need for interdisciplinary treatments customized to each patient's unique traits and illness presentation is highlighted by the questionable success of adjuvant medicines, such as radiation therapy and chemotherapy. Despite receiving treatment, CCSLGT typically relapses in a variety of metastatic locations.
7. This paper attempts to provide a concise summary of CCSLGT, covering its prognostic implications, clinical presentation, difficulties in diagnosis, treatment approaches, and histological findings. By combining the body of current research and providing insight,

2. Case Presentation:

A 28-year-old man showed up at the emergency complaining of persistent stomach pain and vomiting in the room. The symptoms have developed over three month. Without symptom relief, he had cautious treatment with an antibiotic course. He was told to visit the emergency department because the abdomen discomfort had gotten so bad over the past two days. There was nothing notable about his medical history in his prior documents. His family history did not include any mention of cancer. An extensive 12 cm nodular mesenteric tumor originating from the small intestine and encroaching on adjacent tissues was shown by MRI investigations. Two suspicious liver lumps were also found, the bigger one measuring 4 cm and the smaller one measuring 2.8 cm. The homogenous T1-weighted MR scans showed muscle was isointense and slightly hyperintense, however the signal intensity on T2-weighted MR images was more varied and more heterogeneous. Both the big lump in the liver's right lobe and the mesenteric mass were subjected to fine needle aspiration (FNA) and core biopsies. Features from the two biopsies were quite consistent with CCSLGT. Spindle cells were the most common morphology, followed by clear cells and focused pleomorphic histomorphology. Spindle cell morphology ranged from bland to very unusual cells, and only small cores of enormous masses were studied; mitosis was common, averaging 12–14 mitosis/10 HPF. The two cores had significant necrosis that covered almost half of the tissue under examination, indicating that the tumor was high grade. (Figure

1 (A, B, and C).

The case was submitted for consultation after immunohistochemistry investigations were completed at the external institution. Both the liver nodules and the mesenteric mass biopsies revealed similar results: patchy positive for synaptophysin and SMA, positive for SOX-10, and positive for S-100 (diffuse strong nuclear and cytoplasmic reactivity). CD117, CD34, HMB-45, and CK AE1/AE3 are all negative. The liver biopsy revealed sporadic benign-appearing glands that were positive for cytokeratin but negative for S-100; this was regarded as non-malignant bile duct glands that were entrapped rather than as a component of a tumor that had undergone divergent differentiation (Figure 2 A, B, C, D). The general sarcoma panel, additional melanocytic markers, neuroendocrine markers, and other cytokeratin markers were among the additional IHC investigations that were conducted for additional assessment. The tumor cells had vimentin and spotty positivity. CD56 and synaptophysin, but negative for CD31, EMA, CK7, CAM 5.2, HMW-CK 903, DOG1, β-catenin, CD3, and CD20; NSE, Chromogranin, Melan-A, Desmin, and Myogenin. Other differential diagnoses were not supported by the IHC profile. Negative desmin and myogenin did not support alveolar rhabdomyosarcoma; negative HMB45 and negative MelanA did not support primary or metastatic conventional clear cell sarcoma; negative CD117 and DOG1 did not support gastrointestinal stromal tumor (GIST); and negative cytokeratin did not support synovial sarcoma. The tumor was diagnosed as a gastrointestinal tract metastatic clear cell sarcoma-like tumor, most likely originating from the small bowel. Fluorescence in situ hybridization analysis was used to identify gene rearrangements in the Ewing sarcoma breakpoint region 1 (EWSR1), validating the diagnosis of CCSLGT.

3. Discussion:

A uncommon soft-tissue cancer, clear cell sarcoma-like tumor of the gastrointestinal tract (CCSLGT) typically develops in the gastrointestinal tract, particularly the stomach and small intestine. The absence of melanocytic differentiation in a tumor originating from the GI tract and the presence of multinucleated osteoclastic giant cells and S-100 positive are required to distinguish this tumor from ordinary clear cell sarcoma (CCS). FISH analysis can be used to confirm the predicted rearrangements of the Ewing sarcoma breakpoint region 1 (EWSR1) gene for additional confirmation.
7. It was eventually identified as a separate creature and described using various names to correspond with its appearance. Young individuals are most frequently affected with CCSLGT, with a female predominance. It is often misdiagnosed due to its rarity and similarities to other malignancies.
8 CCSLGT originates in the gastrointestinal tract and is also present in the liver, mesentery, and retroperitoneum. The average age at diagnosis is 35 years old, although cases can arise at any stage of life.
9.The exact cause of CCS is unknown, however recent research has suggested that a specific chromosomal translocation, t(12;22)(q13;q12), causing an EWSR1-ATF1 fusion gene, may be the cause. Currently, there does not appear to be a disturbance in any specific gene or chromosome explicitly predisposing someone to the disease.
10A review of the literature has revealed that the development of CCSLGT is not influenced by any specific risk factors.

Nonetheless, a small number of studies suggest a link between radiation exposure and certain substances such as arsenic, vinyl chloride, and dioxin.

11 Regretfully, the supporting data the proof for these linkages is weak since the supporting evidence is not strong. Moreover, genetic factors might possibly be involved, as demonstrated by CSS cases in people with neurofibromatosis type 1 (NF1). A detailed examination is warranted by the correlation. The EWSR gene is a typical finding of gastrointestinal clear cell sarcoma (CCSLGT), which is distinguished from melanoma by the absence of BRAF gene alterations. This information is obtained through the use of Fluorescence-in-situ hybridization (FISH) to search for gene discrepancies in CCSLGT.

12, 13 The most typical cellular patterns in which CCSLGT appear microscopically are those of epithelioid and spindle-shaped cells divided by thin fibrous septa in the form of a nest, pseudopapillary, or pseudoalveolar arrangement. These tumors are macroscopicallylobulated.

14, 15 In CCSLGT, multinucleated giant is a frequent sighting. Multinucleated giant cells (CCSLGT) are seen throughout the tumor and are made up of osteoclastic-type cells.

12–14, 15 CCSLGT tumor cells stain positively for CD56, positive for S100 protein in the cytoplasm and nuclei,

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