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DESMOPLASTIC TUMOR OF SMALL ROUND CELLS, A DIFFICULT TO DIAGNOSE NEOPLASM

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INTRODUCTION

Desmoplastic Small Round Cell Tumor (TDPCR) is a rare aggressive neoplasm, belonging to the group of soft tissue sarcomas. It predominantly affects young men, with a mean age of 21 years, mainly involving the retroperitoneum, pelvis, omentum and mesentery. It has a reported survival rate of 15% in 5 years, with an average of 17 months, and is most often diagnosed at an advanced stage due to non-specific clinical presentation, contributing to its difficult diagnosis. From this, there is interest in the clinical study about TDPCR, aiming to understand and contribute to the brief existing literature.

OBJECTIVE

Describe epidemiological data, clinical manifestations, diagnosis, treatment and prognosis of TDPCR, aiming to help the medical and academic community to characterize this malignancy early, in order to seek better perspectives on the prognosis of the disease.

METHODOLOGY

This is a narrative review, carried out through the PubMed database using "desmoplastic small round cell tumor" as descriptors. From this search, ten articles were selected that best addressed the proposed theme.

RESULT

TDPCR is a highly aggressive primary peritoneal tumor that occurs rarely, with a poor prognosis when diagnosed. It mainly affects young adults, between 18 and 25 years old, predominantly in males.

Described for the first time in the literature in 1989 by Gerald and Rosai, as a malignant intra-abdominal desmoplastic small cell tumor with immunocytochemical differentiation of epithelial, neuroendocrine and musculoskeletal cells. The signs and symptoms of TDPCR are nonspecific, and diagnosis in advanced stages is common. Most often, a large intra-abdominal or retroperitoneal/pelvic mass is found with multiple nodular implants and sites of metastasis to lymph nodes, liver and lungs. Furthermore, symptoms may include abdominal pain, weight loss, ascites, constipation and hepatomegaly. Although it mainly affects the abdominal cavity, extra-abdominal cases have been reported. In addition to the nonspecific clinical presentation, another challenge to diagnosis is its morphological appearance similar to other small round cell tumors. However, the characteristics of TDPCR are well defined. Histopathological analysis showing small round blue cells separated by abundant desmoplastic stroma, associated with immunohistochemical study, showing positivity for epithelial markers cytokeratin and epithelial membrane antigen; mesenchymal markers desmin and vimentin; Neuron-specific neural marker enolase and, S100 protein, are useful in reaching the diagnosis. In questionable cases, verification of the specific chromosomal translocation is indicated t(11;22),(p13;q12), which involves fusion of the EWS gene with the WT1 gene (EWS-WT1), also present in Ewing Sarcoma and Wilms, respectively. Currently, even though there are several treatment modalities, there are still no clinical trials supporting efficient therapeutic strategies. Despite the therapeutic regimen for Ewing's sarcoma, volume reduction surgery, abdominal radiation and high doses of chemotherapy with autologous stem cell transplantation, there was no significant improvement in the disease's evolution. Total tumor resection can improve survival, especially in non-metastatic TDPCR. However, this method is often impossible when the disease has already

progressed. Therefore, despite the limited literature regarding this tumor, it is concluded that it is essential to recognize it as a differential diagnosis in young patients, with non-specific abdominal complaints, when disseminated peritoneal disease without visualization of a primary lesion is identified. In these cases, raising a high degree of suspicion can provide early diagnosis and therapeutic planning, and may be a determining factor in the prognosis of the disease.

COMPLETION OF WORK

Through this review, it was possible to compare the clinical, epidemiological and laboratory characteristics and the great difficulty in establishing an effective treatment/diagnosis to contain the evolution of the pathology and avoid unfavorable outcomes. In view of this, due to its rarity, the disease continues to be a great challenge and, for this reason, its study becomes extremely important, having positive repercussions on the life of the affected patient.

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