CASE REPORT

Desmoplastic small round cell tumor: a case report

Tumor desmoplásico de pequenas células redondas: relato de caso

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DOI: 10.5935/2238-3182.20140061

ABSTRACT

The desmoplastic small round cell tumor (DSRCT) is a neoplasm of rare occurrence, described in 1987 by Sesterhenn *et al.* Currently, there are about 101 cases described in the consulted literature. The authors report a case of intra-abdominal DSRCT in a 53-year-old patient, carrier of a palpable mass in epigastrium, asymptomatic. The propaedeutics showed multiple expansive lesions of varying dimensions compromising the peritoneal cavity. The patient was referred to exploratory laparotomy; only cytoreductive surgery was possible. The intraoperative findings are described, tomographic, and macroscopic and immunohistochemical aspects. The patient was followed up at the Oncology Service. He died months after surgery.

Key words: Desmoplastic Small Round Cell Tumor; Abdominal Neoplasms; Immunohistochemical.

RESUMO

O tumor desmoplásico de pequenas células redondas (TDCPR) é uma neoplasia de ocorrência rara, descrita em 1987 por Sesterhenn et al. *Atualmente existem cerca de 101 casos na literatura consultada. Os autores relatam um caso de TDCPR intra-abdominal em paciente de 53 anos de idade, portador de massa palpável em epigástrio, assintomático. A propedêutica evidenciou múltiplas lesões expansivas de dimensões variadas comprometendo a cavidade peritoneal. Foi submetido à laparotomia exploradora, sendo possível apenas a cirurgia citorredutora. São descritos os achados intraoperatórios, tomográficos, bem como os aspectos macroscópicos e imuno-histoquímicos. O paciente manteve acompanhamento no Serviço de Oncologia. Faleceu meses após a cirurgia.*

Palavras-chave: *Tumor Desmoplásico de Pequenas Células Redondas; Neoplasias Abdominais; Imuno-Histoquímica.*

INTRODUCTION

The desmoplastic small round cell tumor (TDPCR) is a neoplasia of rare occurrence, described in 1987 by Sesterhenn et al. [1] and in 1989 by Gerald and Rosai[2]. There are currently close to 101 cases described in the consulted literature[3]. There is a predominance of the male gender, occurring with a higher incidence in children and adolescents. Until now, there is no proven effective therapy for the treatment of TDPCR. The diagnosis is usually done at an advanced stage of the disease, when a large mass is most often detected with dissemination intra-abdominally. Unlike other undifferentiated neoplasias in the abdomen, surgical resection is indicated when

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Submitted: 2010/05/01
Approved: 2014/04/23

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possible. The evolution of the disease is unfavorable, with an average survival of 17 months and variations between 3 and 72 months.[4]

CASE REPORT

P.A.M., 53 years old, male, sought medical assistance due to a palpable abdominal mass in the mesogastric with no other symptoms. On physical examination, a mesogastric mass was palpable, voluminous, and painless. CT showed multiple expansive lesions of varying dimensions compromising the peritoneal cavity, exuberantly impregnated and with heterogeneous contrast, being the largest dimension of approximately 18 cm in the cranium-caudal axis, located in the median plane of the abdominal cavity, stretching from the mesogastric to the hypogastric region (Figures 1 and 2).

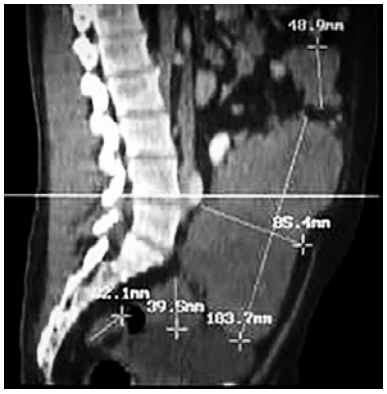

Figure 1 - Computed tomography of the abdomen. Presence of intra-expansive abdominal lesion and multiple implants.

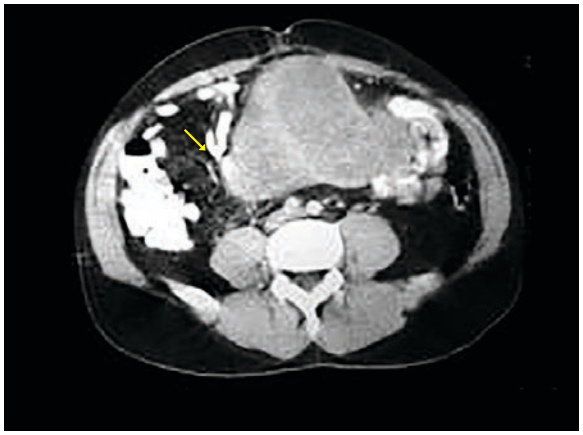

Figure 2 - Computed tomography of the abdomen. The arrow indicates the intra - abdominal lesion.

Median laparotomy was performed for the resection of the mass and other implants, being found lesions on the diaphragmatic right dome, inter-aortic-cava, and peritoneal implants. Because the advanced stage of the disease and unresectable character found during surgery, some of these lesions were addressed for anatomo-pathological study, which revealed homogeneous architecture and well-defined cell aggregates with fibrous desmoplastic stroma. Immunohistochemistry analysis showed overexpression of protein S-100 (Figure 3), positivity for epithelial markers (epithelial membrane antigen – Figure 4), mesenchymal such as WT-1 (Figure 5), and desmin (Figure 6). The microscopic study with hematoxylin-eosin staining (Figures 7A and 7B) identified islets of hyperchromic tumor cells with rounded nuclei, barely visible nucleoli, and scanty cytoplasm well delimited by abundant fibrous stroma.

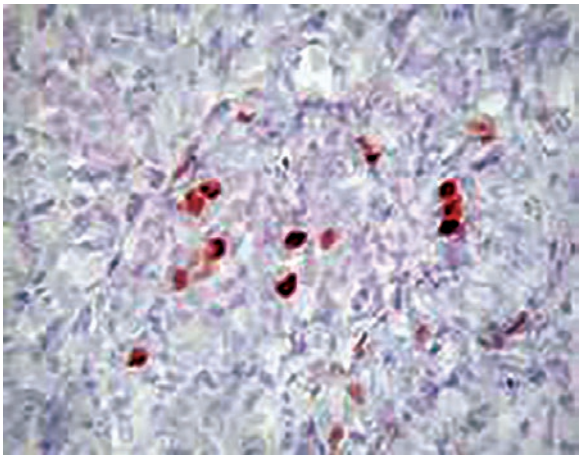

Figure 3 - Protein S 100.

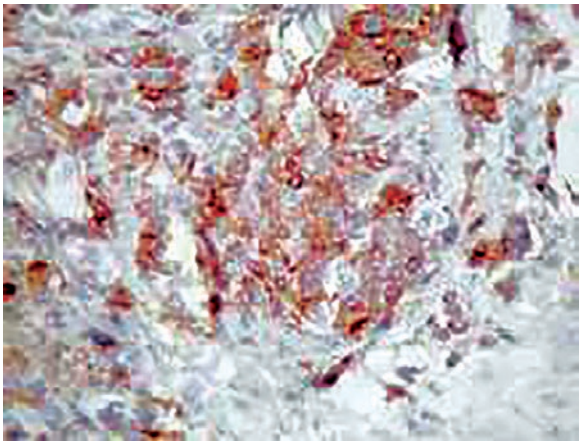

Figure 4 - Membrane Epithelial Antigen.

The patient was discharged after two weeks of surgery. Remained in followup at the Oncology Service and died months after surgery.

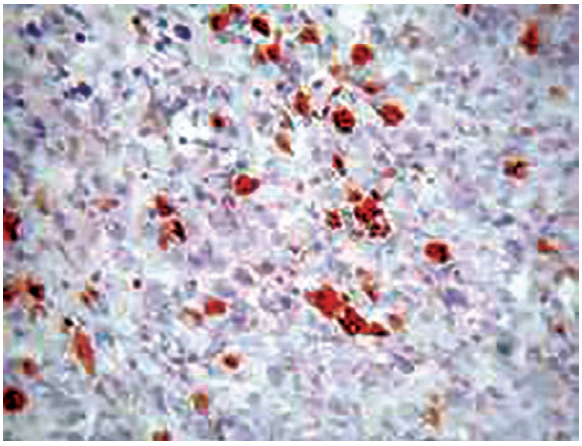

Figure 5 - WT-1 Marker. Product of the tumor suppressing gene.

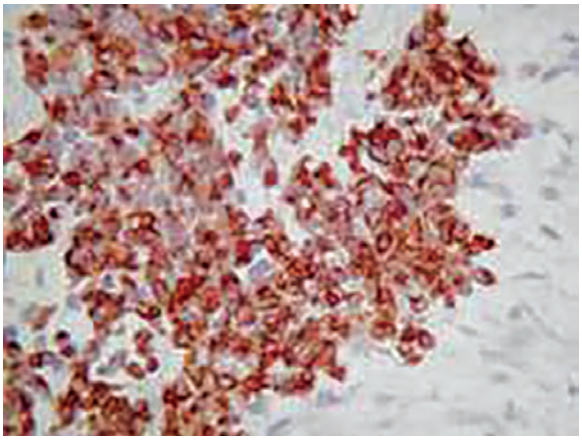

Figure 6 - DESMINA Marker. Paranucleous globular pattern ultra-structurally corresponding to paranuclear aggregates of intermediary filaments.

DISCUSSION

TDCPR is a rare entity. There are close to 101 cases in the world literature, [1] being four in the consulted Brazilian literature. Is a neoplasia that shows preference for the masculine gender, in the ratio of 4:1, affecting predominantly young patients.

In most cases, the clinical findings are nonspecific and with gastrointestinal symptoms through the presence of an abdomen tumor, and in other systems by metastatic dissemination. The most found form of this type of lesion consists of a large abdominal mass, with multiple nodules and dissemination through the peritoneal surface. The main TDCPR site is intra-abdominal.

Pleura involvement, para-testicular region, bones, ovaries, and soft tissue have been also reported. In some circumstances, it is possible to establish the lesion site of origin.

The clinical presentation and TDCPR topography in the reported case is compatible with the most common location for this type of lesion. Macroscopically, the tumor displays firm, elastic, cambered, grayish-yellow to brown tissue, with none of these characteristics being pathognomonic tissue. The pathogenesis remains unknown.

However, there is a translocation [t(11;22)(p13;q12)] that involves the fusion of the EWS gene with the WT1 gene. The immunohistochemical profile of TDPCR features the positivity for the epithelial markers cytokeratin and epithelial membrane antigen; mesenchymal desmin and vimentin markers; and protein S100. The WT1 protein overexpression (Wilm's Tumor Protein) has been demonstrated in TDPCR, featuring a fusion of the EWS/WT-1 genes.

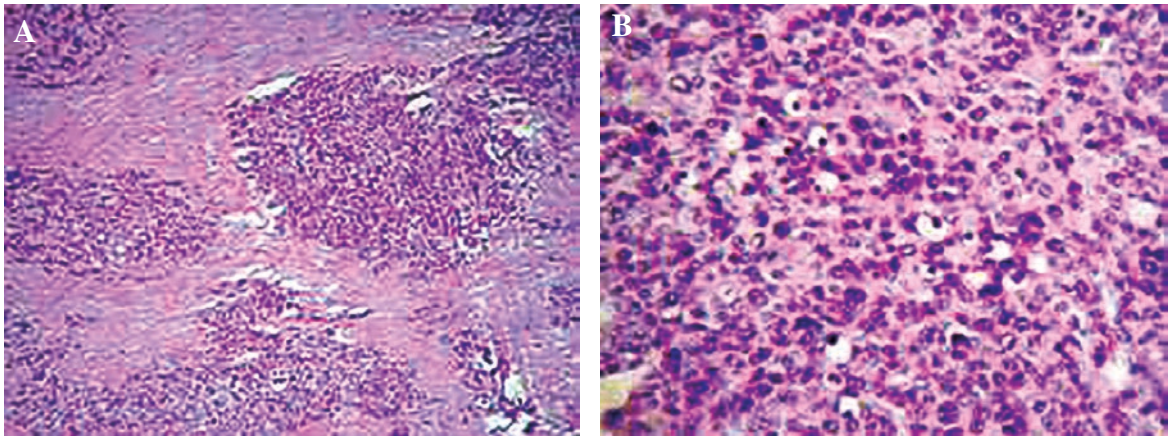

Figure 7 - Hematoxylin-Eosin. Islets of islets of hyperchromic tumor cells with rounded nuclei, barely visible nucleoli, and scanty cytoplasm well delimited by abundant fibrous stroma.

The clinical case showed alterations similar to those described for the immunohistochemical profile of TDPCR. The metastases occur most commonly in the peritoneum, liver, and lymphoid tissue.

Currently, there is no effective therapeutic against TDPCR and, unlike other undifferentiated neoplasias in the abdomen, surgical resections are indicated when possible. Reports of multidrug therapy have been observed, as well as autogenic bone marrow transplant, however, without changing the survival of these patients. Radiation therapy also proved ineffective.

The evolution of the disease is unfavorable, with average survival of 17 months, with variations between 3 and 72 months.

ACKNOWLEDGEMENTS

The authors are thankful to Dr. José de Souza Andrade, a pathologist from the Felício Rocho Hospital for his contribution in the diagnosis and publication of this case.

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