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CASE STUDY

Small round cell desmoplastic tumour. Atypical morphology in the sub-maxillary gland

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KEYWORDS
Desmoplastic small round cell tumour;
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Abstract
Desmoplastic small round cell tumour (DSRCT) is a rare disease usually affecting young males. There are no other articles with a sub-maxillary location. The tumour consists of nests and masses of undifferentiated small round cells embedded in a desmoplastic stroma. The co-expression of epithelial, muscular and neuronal antigens distinguishes this entity from other small round cell tumours. The t(11;22) (p13;q12) translocation is a recurrent characteristic of this type of tumour.
We report a case of desmoplastic small round cell tumour of the sub-maxillary gland, with an evolution of 8 months, affecting a 36-year-old male. He suffered chronic lymphatic leukaemia 5 years ago and needed a bone marrow transplant. There was a 4x3 cm tumour. There were no signs of malignancy on the CT scan. A right submaxillectomy was performed. The pathology analysis gave a diagnosis of DSRCT. Post-surgical radiotherapy was given.
The definitive diagnosis was reached using immunohistochemical techniques, such as polyphenotypical differentiation (epithelial, mesenchymal, and neural), and by demonstration of translocation (11;22) (p13;q12). Sub-maxillary location is very rare.

PALABRAS CLAVE
Tumor desmoplásico de células pequeñas y redondas;
FISH;
Translocación;

Tumor desmoplásico de células pequeñas y redondas. Morfología atípica en la glándula submaxilar

Resumen
El tumor desmoplásico de células pequeñas redondas (TDCPR) es una neoplasia infrecuente que afecta a varones jóvenes y se localiza en el abdomen principalmente. La localización submaxilar es extremadamente infrecuente y no hemos encontrado ningún caso en la literatura. La

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histología típica se caracteriza por nidos de células tumorales embebidas en una estroma fibromixoide densa. Su principal característica diagnóstica es la coexpresión de marcadores epiteliales, mesenquimales y neurales y la translocación recíproca t(11;22) (p13;q12).
Presentamos el caso de un varón de 36 años con antecedente de leucemia linfática crónica y trasplantado de médula ósea con una tumoración submaxilar derecha de 8 meses de evolución. Exploración física: masa dura de 4 x 3 cm. La tomografía computarizada no muestra signos de malignidad. Se realiza submaxilectomía derecha. El análisis anatomopatológico reproduce la morfología y el inmunofenotipo característicos del TDCPR. Se completa el tratamiento con radioterapia.
El diagnóstico definitivo se basa en la demostración inmunohistoquímica de diferenciación polifenotípica (epitelial, mesenquimal, neural), y en la demostración citogenética de la translocación (11;22)(p13;q12). La localización submaxilar es excepcional.

Introduction

Desmoplastic small round cell tumour (DSRCT) is a rare neoplasm affecting the abdominal cavity of children and young adults, especially males. It was described as a separate clinical-pathological entity by Gerald et al. in 1991. In 1989, Rosai carried out its anatomical-pathological description.[1]

It has been described in other locations with a much lower frequency (peritoneum, pleura, ovary, testis, kidney). About 150 cases of DSRCT have been reported in the medical literature.[2]

Clinical report

Male patient of 36 years of age, with a history of chronic lymphocytic leukaemia which had required bone marrow transplant 5 years earlier, with right submaxillary tumour lasting for 8 months. He presents with recent pain in the area and an increase in size.

Examination revealed a tumour in the right submaxillary area, not rolling and adhered to deep planes.

Cervicofacial CT scan: tumour in the right submaxillary of 5x4 cm in diameter. It does not infiltrate adjacent areas and displaces neighbouring structures, with no signs of malignancy.

Right submaxillectomy is carried out and 2 adjacent nodes are removed, the largest measuring 3.5 cm and the smaller, 3 cm. The tumour was greyish with yellow foci surrounded by lobes of salivary gland. It had a firm consistency and focally infiltrative edges (Figure). No post-operative complications appeared.

Figure Pathological specimen of right submaxillectomy.

Pathology report: malignant DSRCT. Focally infiltrating adjacent soft tissue. Reactive lymphadenitis in periglandular nodes. Presents large nests of round cells, with areas of central necrosis.

Immunohistochemical study: positive phenotype for tumour cells, intense and paranuclear for CAM 5.2, EMA, vimentin and desmin; diffuse nuclear expression for progesterone receptors, EGFR+, focal ENE+ and middle-high proliferative activity (MIB1); P53+ NM23+ in cytoplasm and membrane, C-Kit, weak cytoplasmic B-catenin+, Cerb2-, actin-, CD99-, Cytoq 20-, S100-, 34 BE12-. t11;22 is confirmed.

Images of nuclear interface with fracture of EWS (60%).

The final molecular confirmation came through the detection of EWS-WT1 fusions by later RT-PCR.

Ten months after the surgery, the patient presented no local recurrence or metastasis.

Discussion

DSRCT is uncommon. Enziger classified it histopathologically as a sarcoma in miscellaneous B-type malignant tumours. It represents a new translocation, affected by chromosomes 11 and 22, with different break points from other diseases [t(11;22) (p13;q12)].[3]

The term "small round cell tumour" describes very aggressive malignant tumours composed of small and undifferentiated cells with round or oval hyperchromatic nuclei, non-evident nucleoli and scant cytoplasm.[4]

This group includes: Ewing's sarcoma, peripheral neuroepithelioma, peripheral neuroblastoma, rhabdomyosarcoma, desmoplastic small round cell tumours, lymphoma, leukaemia, small cell osteosarcoma, small cell carcinoma, and malignant melanoma.[5]

It appears between ages 3 and 48, with an average age of 18-20 years, predominantly in males (4:1).[6] Patients present non-specific symptoms such as pain and palpable masses, as in this case. It normally affects the abdominal or pelvic peritoneum diffusely. Other locations of much lower frequency are the tunica vaginalis, paratesticular region, retroperitoneum and prostate area, pleura, central nervous system, liver, ovary, etc.[7] We have not found any case in the literature with a submaxillary origin.

Cytogenetic studies show translocation t(11;22) which differs from the translocation of Ewing's tumour and seems to be specific for this entity.[6]

Three very useful markers are used to distinguish sarcomas, carcinomas and lymphomas: pan-leukocyte antigen CD45 (specific for lymphomas), cytokeratins (present in tumours of epithelial origin and sarcomas), and vimentin (present in mesenchymal tumours and some carcinomas).

The differentiation of this tumour is achieved by immunohistochemistry for epithelial (keratin antigen of the cytoplasmic membrane of epithelial cells), neuronal (neurospecific enolase), and muscular (desmin) markers. Similarly, the reciprocal translocation t(11;22) (p13;q12) which involves the product of the *EWS1* and *WT1* genes is a recurring and specific feature of this type of sarcoma.[8,11]

Treatment can include chemotherapy and/or radiation therapy and/or aggressive surgical resection when possible. In any case, patients have a poor prognosis with an average survival of approximately 17 months.[10]

Conclusions

DSRCT is a type of sarcoma with defined histochemistry and translocation t(11;22) (p13;q12). Sub-maxillary location is extremely rare. Treatment is surgical accompanied by chemotherapy and radiation therapy. Nonetheless, it has a very bad prognosis.

Conflict of interests

The authors have indicated there is no conflict of interest.

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