Case Report

Intra-abdominal desmoplastic small round cell tumor

Zeeshan-ud-din, Nausheen Yaqoob, Turab Pishoroi, Muhammad Zafar Rafique, Ali Kamran
Department of Pathology and Microbiology, The Aga Khan University, Karachi.

Abstract

Intra-abdominal desmoplastic small round cell tumor (IDSRCT) is a unique, highly aggressive neoplasm that chiefly affects male adolescents and young adults and most frequently presents as a large abdominal mass with widespread peritoneal involvement at the time of diagnosis. We present two cases of IDSRCT in a young male and a female. Both typically presented with diffuse peritoneal involvement.

Introduction

Intra-abdominal desmoplastic small round cell tumor (IDRSCT) is an uncommon neoplastic condition that predominantly occurs in young adult men and usually diffusely involves the abdominal and/or pelvic peritoneum.[1] IDSCRT has distinctive clinical, histologic and immunophenotypic features. Histologically it is typically composed of nests of small, undifferentiated round or oval hyperchromatic cells with abundant desmoplastic stroma. Immunohistochemically it is reactive for epithelial markers; keratin, epithelial membrane antigen (EMA), neural (neuron-specific enolase; NSE) and muscle markers (desmin).[1] Molecular studies have identified a translocation t (11; 22) (p13; q12) as being unique to IDSRCT.[2] We describe two cases of IDSRCT in a young male and a female.

Case Report

Case No. 1

A 32 year old well built male presented with complaints of pain in the left upper quadrant, weight loss and constipation for 6 months. He also noticed a mobile mass in the left upper quadrant. There was no significant past medical history and he did not have any family history of cancer. On examination there was a non tender mobile mass palpable in the left upper quadrant of the abdomen. On ultrasound examination of the abdomen, multiple deposits in the liver and spleen were observed. A barium enema revealed extrinsic compression of the colon near the splenic flexure due to a mass and a CT scan confirmed the above findings. A colonoscopic as well as needle biopsy of the mass was done which at that time revealed an undifferentiated neoplasm. He was seen in the outpatient clinic and was referred by the gastroenterologist for a surgical opinion considering this as colon cancer. A diagnostic laparoscopy was performed, multiple peritoneal deposits of tumour were found one of which was taken for biopsy. Histopathology of the specimen showed a neoplastic lesion composed of clusters of small tumor cells with scanty cytoplasm. Nuclei were of small size and pleomorphic with inconspicuous nucleoli. Marked desmoplastic response was identified around the tumor cell clusters (Figure 1). No glycogen positivity was seen on special stains. Immunohistochemical studies showed strong Cytokeratin AE1/AE3 (Figure 2), Cytokeratin Cam 5.2, CK 7, epithelial membrane antigen positivity. Globoid dot positivity of Desmin was seen along

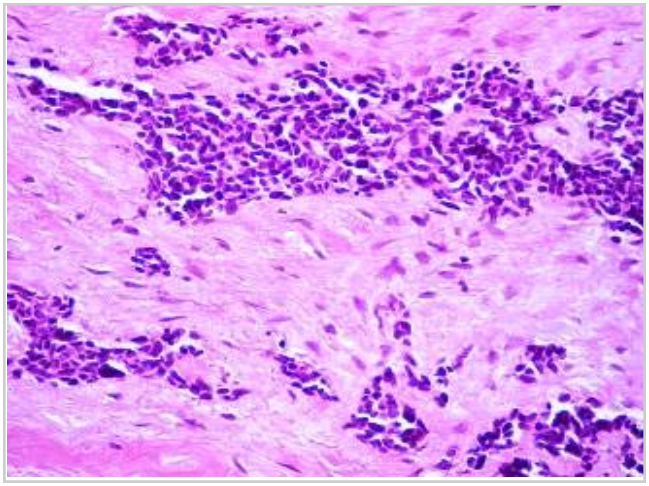

Figure1. H & E section of IDSRCT showing nests of small tumor cells surrounded by dense desmoplastic stroma. 20X.

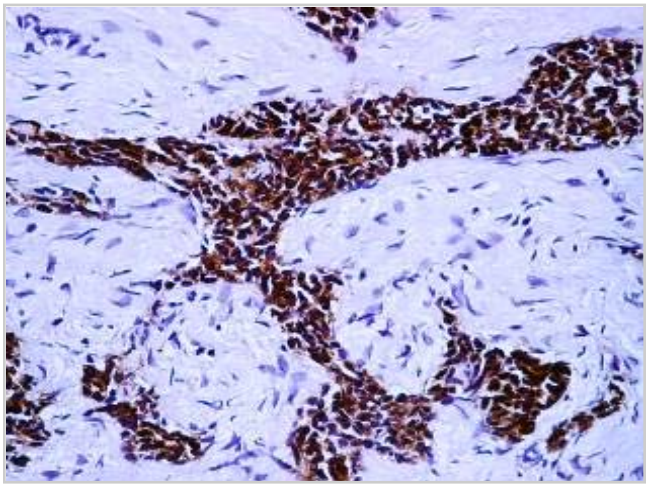

Figure 2. The tumor cells of IDSRCT show strong positivity for CK AE1/AE3. 20X.

with focal positivity of Vimentin and Neuron specific enolase. The tumour cells were negative for S-100, Mic-2, ASMA, Chromogranin and synaptophysin. Based on morphological and immunohistochemical features, a diagnosis of Intra abdominal desmoplastic small round cell tumor was made.

Case No. 2

A 23 year old female presented with the complaint of constipation and abdominal pain for the last two months. On examination masses were palpable in right and left pelvic regions. Per-vaginal examination showed hard irregular masses in the pouch of Douglas. Laparotomy was done; peroperatively hard irregular nodules were present in the pouch of Douglas alongwith bilateral ovarian masses. No para-aortic lymphadenopathy was present. Bowel and mesentry were normal. Total abdominal hysterectomy and bilateral salpingo-opherectomy with omentectomy was done along with removal of masses from pouch of Douglas. Grossly right ovary was 9 x 5 cms and left ovary was 7 x 6 cm in size. Cut surface showed entire ovary replaced by grey white firm lesion. Uterus and cervix were grossly unremarkable. Multiple sections were taken from the right and left ovaries, omentum and masses from pouch of Douglas. Histological and immunohistochemical findings were consistent with intra abdominal desmoplastic small round cell tumor.

Discussion

IDSRCT usually involves the abdominal and/or pelvic peritoneum but extra-abdominal location has also been described. Cases are reported in the central nervous system, bone, extremity, kidney, salivary gland, paratesticular region and pleura.[1] The histogenesis is uncertain.

Common clinical presentations are pain, abdominal distension and a palpable abdominal, pelvic and scrotal mass, sometimes with associated ascites. Histologically uniform closely packed small cells are distributed in a background of desmoplastic stroma. Differential diagnosis includes lymphoma, Ewing sarcoma/PNET, neuroblastoma, alveolar rhabdomyosarcoma, malignant mesothelioma. Cytologically, the tumor cells consist of small, round to oval cells with a scant amount of light blue cytoplasm. Cytogenetic and molecular analysis have revealed a typical t(11,22)(p13,q12) translocation. This translocation involves fusion of the Ewings sarcoma gene on chromosome 22 with the Wilm's tumor 1 gene (WT1) on chromosome 11. This gene fusion is postulated to produce an oncogenic chimeric protein with the zinc finger DNA-binding domains of WT1 with the transcriptional regulatory domain of the Ewings Sarcoma gene. Ordonez[1] studied 39 cases which demonstrated spindle cells, signet ring cells, Homer Wright rosettes, insular pattern, tubules and epithelioid cells. Ultrastructural features in some cases include presence of intermediate sized cytoplasmic filaments. An elevated serum CA 125 concentration is found in some patients with IDSRCT clinically.[3] Elevated CA-125 levels were also noted in both our cases.

Ovarian involvement by IDSRCT has been frequently reported. According to a study conducted by Zaludek et al[4], ovarian involvement was seen in 30% of the cases. This frequency was observed by him in a series of 17 cases of IDSRCT in young females. Three cases of ovarian involvement by IDSRCT, at the time of initial diagnosis were also reported by Young et al.[5] Another case is reported by Slomovitz et al[6] who reported ovarian involvement by IDSRCT in a young female. Reich et al[7] have described a IDSRCT with an unusual age of presentation mimicking a metastatic ovarian neoplasm. We have also seen a case of IDSRCT in a 23 year old female with bilateral ovarian masses and widespread peritoneal involvement. The patient also had raised CA-125 levels. Similar case of IDSRCT with raised CA-125 levels with ovarian, omental and hepatic involvemnt in a young female was reported by Parker et al.[8] DSCRCT may mimic an ovarian primary tumor hence it should be added to the differential diagnosis of unusual gynaecologic malignancies in elderly as well as younger females.

The differential diagnosis in cases of IDSRCT involving the ovary is that of ovarian small cell carcinoma of hypercalcemic type (OSCCHT). Histologically both tumours differ significantly. OSCCHT lacks the desmoplastic stroma characteristic of IDSRCT and when present it is diffuse and irregular in distribution. Also OSCCHT exhibits follicle like spaces lined by neoplastic cells and IDSRCT contains degenerative spaces that result from tumor necrosis. WT1 is characteristically positive in IDSRCT using an antibody against the C-terminal. OSCCHT is also usually WT1 positive but with an antibody against the N-terminal.[9] Patients with IDSRCT present with a short duration of nonspecific symptoms, and the disease is fatal almost uniformly, regardless of the treatment modality used. It is a chemosensitive tumor, generally with short-lasting response and poor survival gain from systemic chemotherapy.[10] Both of our cases were lost to follow up, therefore we can not assess the fatality of disease in these patients.

References

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