An 18-year-old college student was referred from the University Clinic for having low hemoglobin (Hg 4.7 g/dl).
He was completely well till one month back when he presented with easy fatigability, postural dizziness, palpitation and dyspnea.
He gave history of 30 kg weight loss over the past 6 months.
On physical examination, he looked pale, not jaundiced or cyanosed.
His vital signs were all within normal limits, except for an elevated heart rate (98/min).
Cardiovascular, respiratory and abdominal examinations were unremarkable.
Primary investigations in our hospital revealed low hemoglobin level (5.1 g/dl), low iron (<10 ug/dl), and a positive occult blood test.
A provisional diagnosis of microcytic hypochromic anemia for further workup was made.
Bone marrow aspirate and trephine biopsy revealed normocellular marrow, depicting normal trilineage hematopoiesis.
Upper GI endoscopy showed erosive antral gastritis with patchy ulcerative inflammation.
Abdominal ultrasound showed a slightly enlarged liver with a rounded echogenic lesion in the anterior wall of the right lobe suggestive of hemangioma.
The spleen was slightly enlarged and normal in echogenicity with no focal lesions.
Computed Tomography (CT) scan of the abdomen and pelvis showed a fairly large well defined soft tissue mass located in the anterior upper pelvis and engulfing jejunal loops causing bowel wall thickening.
The patient underwent an exploratory laparotomy and excision of the jejunal mass.
Macroscopic examination revealed an 11 × 8 × 2 cm annular mass located within the jejunal wall ulcerating through the mucosa and extending to the serosal surface.
Microscopic examination revealed a tumor situated in the muscularis propria and extending to the mucosa and the serosa.
The neoplastic cells were arranged in predominantly pseudoalveolar pattern (Fig.1a,b).
They were polygonal in shape with variable amount of eosinophilic to clear cytoplasm (Fig.1c).
The nuclei were oval with vesicular chromatin and inconspicuous nucleoli.
Scattered osteoclast-like multinucleated giant cells were also identified (Fig.1b,c).
Frequent mitotic figures and necrosis were noted.
All lymph nodes were not involved by tumor.
Immunohistochemically, the neoplastic cells were strongly and diffusely positive for S-100 protein (Fig.1d).
They were also positive for vimentin.
Melanocytic markers (HMB45 and Melan A), neuroendocrine markers (chromogranin A, synaptophysin and CD56), smooth muscle actin, desmin, CD34, CD117, cytokeratin and LCA were all negative in the neoplastic cells.
Fluorescein in situ hybridization (FISH) analysis for EWSR1 break apart probe on paraffin-embedded tumor showed evidence of a 22q12 rearrangement in 197 out of 205 (96%) of interphase nuclei scored.
The native state of EWSR1 break apart probe will be seen as two adjacent or fused (overlapping) red/green (yellow) signals.
However, EWSR1 gene rearrangement presented as one red and one green separated signal (Fig.1e).
The patient did not receive chemotherapy or radiotherapy.
During the clinical follow up, the patient remained disease free for 3 years until he presented with local recurrence.
The treatment plan was surgical resection but the patient sought medical advice in a different institution where he died of the disease a year later.