Desmoid tumor (DT) is a benign and slow-growing soft tissue tumor of intermediate grade with potential for local recurrence and invasive growth without metastatic potential [1–3]. It is rare with the annual incidence of 2–4 cases per million population in the United States [4]. The tumor is thought to originate from musculoponeurotic or fascial structures [5]. In pediatric patients, most common tumor sites are head and neck and extremities; abdominal locations are rare [6,7]. Because of its infiltrative nature, the tumor frequently affects adjacent organs and its clinical presentation usually is determined by the organs it invades. We report a 14 year old boy with Asperger syndrome, who presented with rapidly worsening left upper quadrant abdominal pain, secondary to splenic infarction by effects of large abdominal desmoid tumor.

1. Case report

A 14 year old male developed progressively worsening left-sided abdominal pain, and nausea and vomiting 3 days prior to admission. Because of the worsening pain he was taken to our Emergency Department where acute surgical abdomen was suspected and was hospitalized. The pain was not alleviated by over-the-counter non-steroidal anti-inflammatory drugs (acetaminophen and ibuprofen).

He was an otherwise healthy child with high functioning Asperger syndrome. The patient and the mother denied any history of abdominal surgery or trauma. The family history was negative for any gastrointestinal cancers but was positive for lymphoma in an uncle.

On examination, following administration of morphine for his pain, he was in acute distress with mild dehydration. The findings consisted of tender and full abdomen more on the left side. A firm mass measuring approximately 10 × 17 cm was felt in the left upper abdomen. The upper border of the mass was not clearly defined on palpation. Spleen could not be palpated separately from the mass. A computerized tomography of the abdomen revealed a large hypo-intense mass occupying most of the left upper abdomen measuring 11 × 12 × 22 cm with internal necrosis (Fig. 1A and B). Hypo-intense...
area was also seen in the spleen, suspicious for asplenic infarct. Serum α-fetoprotein, CEA (carcinoembryonic antigens), and β-HCG (human chorionic gonadotropin) were negative. Because of the large size of the tumor it was decided to perform an incisional biopsy first to rule out malignancy. The biopsy showed proliferation of myofibroblastic cells characterized by vesicular nuclei and stellate basophilic cytoplasm arranged within the fibromyxoid stroma suggestive of aggressive fibromatosis. Unfortunately immunohistochemistry for β-catenin was retrospectively falsely negative. Considering the large size of the mass, we thought the biopsy findings (particularly negative β-catenin) not representative of the true tumor histology.

Subsequent laparotomy revealed a giant tumor located in the left upper quadrant of the abdominal cavity; it was movable and not fixed to the posterior abdominal wall but showed adhesions to pancreas, colon, spleen and greater curvature of stomach. A un block excision of tumor with the adherent spleen and omentum and partial resections of adherent stomach and pancreas was successfully performed. The resected area along the greater curvature of the stomach was stapled and sutured over. The patient tolerated procedure well and was discharged subsequently with no significant symptoms.

2. Pathology findings

The tumor weighed 2880 g and measured 28.5 × 17 × 11 cm. It was adherent to spleen which showed areas of geographic necrosis (infarction; Fig. 2A and B). The sectioning of the mass revealed solid rubbery consistency and a central area of cystic degeneration. Microscopically, the tumor was composed of uniform spindle cells set in collagen stroma containing prominent vessels sometimes with perivascular edema. The cells were arranged in sweeping bundles (Fig. 2C). The mitotic rate was negligible and no significant necrosis, except for the central area of degeneration was seen. The tumor was covered by peritoneum except for the areas where it infiltrated splenic hilum with enveloping of splenic vessels, pancreas and gastric wall. No infiltration of omentum or involvement of multiple reactive mesenteric lymph nodes was seen. Resection margins were negative. The tumor cells showed immunoreactivity with caldesmon and smooth muscle actin as well as strong nuclear and some cytoplasmic β-catenin staining (Fig. 2D). ALK (anaplastic lymphoma kinase), HHF-35 (a monoclonal antibody that reacts with muscle actin), desmin, myogenin, S-100, CD34, CD117, NFP (neurofilament protein), and WT-1 (Wilms tumor-1 protein) immunostains were negative. Ki-67 proliferative index was approximately 5–10%. The morphology and immunophenotype were diagnostic of desmoid-type fibromatosis. Genetic testing for APC was negative.

3. Discussion

Desmoid-type fibromatosis, also known as aggressive fibromatosis, Musculoaponeurotic fibromatosis or desmoid tumor is a rare soft tissue neoplasm characterized by infiltrative growth and a tendency toward local recurrence, but an inability to metastasize. The term “desmoid” is of Greek origin and means “band-like” [4]. Reitamo et al. [4] published a significant observation that up to 48% of the patients had radiological findings of various skeletal abnormalities consisting of cortical thickening, exostosis, cystic areas of translucency, and compact islands of the femur, alone or in combination. Furthermore, these skeletal abnormalities were found in the relatives of the patients. They concluded that the skeletal abnormalities were transmitted in an autosomal dominant fashion [4]. It may be that this constellation of findings represents a unique syndrome. Prior surgery or trauma alone could be the other possible trigger for the development of DT and association with estrogen exposure, in particular in young females is also commonly described in literature [8–11]. Our patient had no history of skeletal abnormalities or abdominal trauma or previous surgeries.

DT itself is a very rare tumor in pediatric age group and only 5–7% of tumors in children are abdominal (mesenteric) [6,7]. Most of the abdominal DTs remain asymptomatic until complications due to pressure or invasion occur. Intestinal obstruction, hydro-nephrosis or even cachectic symptoms have been reported [12–15]. DT is a slow growing neoplasm and acute onset of abdomen presentation such as in this case is unusual. The pain in our patient was likely attributable to the splenic infraction. Our patient also had Asperger syndrome. Children with Asperger syndrome have problems with sensory modulation and socialization [16–18]. It is possible that he did have chronic indolent pain, but did not verbalize any pain until it became intolerable.

Currently adequate surgical resection with negative margins is the treatment of choice, except cases when radical surgery would be mutilating or associated with considerable function loss [19]. Postoperative radiotherapy, chemotherapy, or combination of the two has been used in cases of positive margins with variable successes [20]. Debunking chemotherapy or radiotherapy has also been employed in cases of initially unresectable tumors [7]. Because of radiation-related morbidity and complications, radiotherapy should be avoided in cases with negative tumor margins. In our case, complete resection with tumor-free margin was possible, and no additional therapy was given. It would be prudent to perform testing for APC mutation for all patients with this tumor, since patients may develop colorectal cancer which would be more life-threatening than desmoid tumor. Literature review showed no
differentiating features, either clinically or pathologically the tumor associated with APC gene mutation from those without. Fortunately our case had negative APC mutation.

4. Conclusions

Desmoid tumor is a rare, locally aggressive tumor with frequent recurrences without metastasis. It invades adjacent organs and anatomic structures causing symptoms. Mesenteric origin like in our case represents only 5% of pediatric desmoid tumors. If possible, total resection of the tumor with free margin gives the best prognosis without any adjuvant therapy. Unresectable or partially resectable tumors and recurrent tumors have been treated with local radiation and/or adjuvant chemotherapy with variable successes. Though randomized clinical trials are needed to evaluate benefits of radio-, and chemotherapy and other adjuvant therapy such as estrogen and non-steroidal anti-inflammatory drugs (NSAIDS), the rarity of the tumor in children may preclude such studies.

Conflict of interest statement

All the authors declare that there are no conflict of interests regarding any personal or financial relationship with other people or organizations that could inappropriately influence (bias) their work.

References