Japanese Journal of Gastroenterology and Hepatology

Case Report

ISSN: 2435-1210 | Volume 8

Intestinal Congenital Fibrosarcoma Revealed by Intestinal Occlusion in A Three Months Old Child: Report of an Exceptional Case

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Received: 19 Feb 2022
Accepted: 28 Feb 2022
Published: 07 Mar 2022
J Short Name: JJGH

Citation:

Azzakhmam M, Intestinal Congenital Fibrosarcoma Revealed by Intestinal Occlusion in A Three Months Old Child: Report of an Exceptional Case. Japanese J Gstro Hepato. 2022; V8(9): 1-4

Keywords:

Congenital Infantile Fibrosarcoma; Intestinal; Rare

1. Abstract

The congenital infantile Fibrosarcoma (IFS) is a rare non rhabdomyosarcoma malignancy diagnosed early at bird or first years of new born life. The intestinal location of this malignancy is exceedingly rare.no specific clinical or radiological nor histological signs can lead to diagnosis. The ETV6-NTRK3 fusion transcript is characteristic molecular signature of this tumor, but not always present. Herein, we report an exceptional case of intestinal IFS revealed by intestinal occlusion and which the diagnosis been assessed by molecular biology demonstrating the presence of ETV6-NTRK3 fusion transcript in SAINT JUDE pediatric hospital.

2. Introduction

Congenital infantile fibrosarcoma (IFS) is a non-rhabdomyosarcoma soft tissue malignancy. This condition is exceptional as it occurs in less than five new cases per million children [1]. this rare malignancy represents 5 to 10% of soft tissue sarcoma before 1year old [1,2,3]. 40%of these tumors are present at birth, but the reported age of diagnosis is three months [2]. Few cases of IFS involve the gastrointestinal tract (as only 10% occurs in the abdomen) [3]. The IFCS develops usually in the limbs (approximal 70%). But many ectopic cases were reported and some of them are located in the abdominal region and deriving from the intestine [3,4-7]. Herein, we report an exceptional case of IFS in a three years old child revealed by an intestinal occlusion, giving suspicion first to a stromal tumor, then undifferentiated sarcoma, which have been finally confirmed in SAINT JUDE-hospital, as having the ETV6-NTRK3 fusion-positive mutation sarcoma confirming the diagnosis of IFS of the intestine.

3. Case Presentation

The emergency of a general hospital received a three years old boy plaining of abdominal distension and vomiting. The physical examination found a dehydrated new born with abdominal distension. Medical history was insignificant; unique infant of his family coming from a first-degree blood related parent the echography examination described a polylobed mass of 60mm under hepatic which let distinguish invagination or lymphoma. The new born underwent a surgery for intestinal strangulation; segmental intestinal resection involving the mass.

4. Gross Examination

The intestinal resection measured 13.5 cm and carried a polylobed tumor limited by a capsule. the tumor measured 5,5x3x2 cm. the cut surface was fleshy grey with hemorrhagic and necrosis areas. The tumor was located at 2,5cm from the ileocecal valve, and 3,5 cm from the nearest margin. There was an appendix measuring 3,5 cm. the lymph node dissection found five lymph nodes.

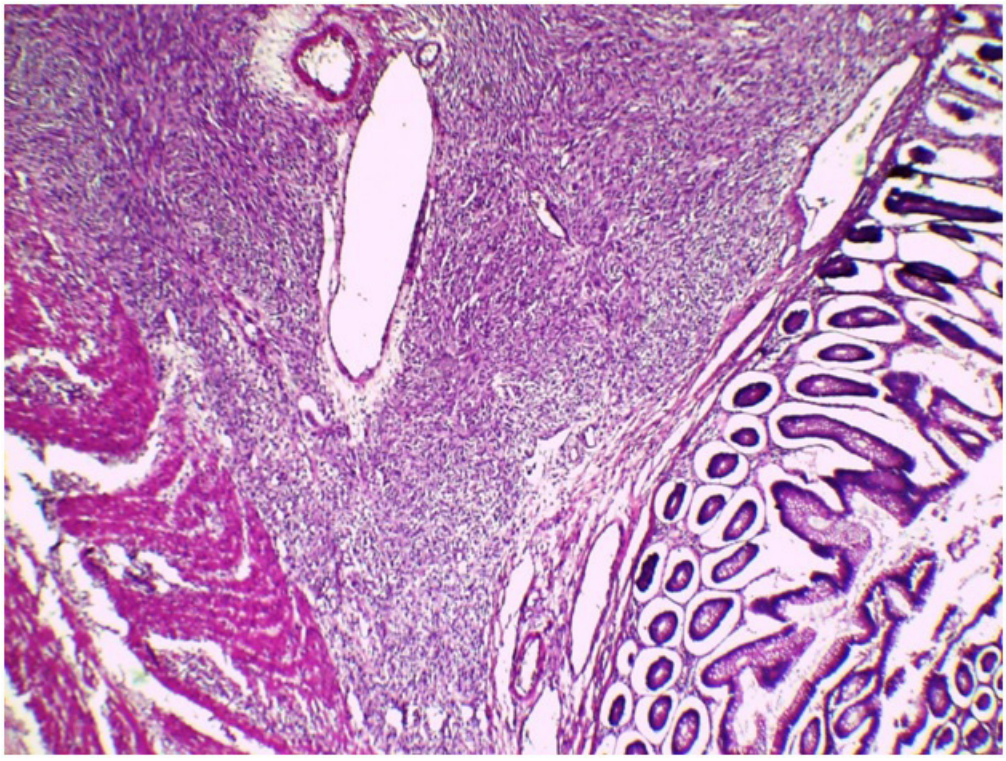

Figure1: microscopy [HEx10]; low power showing tumoral proliferation of spindle shaped cells most extensive in the submucosa.

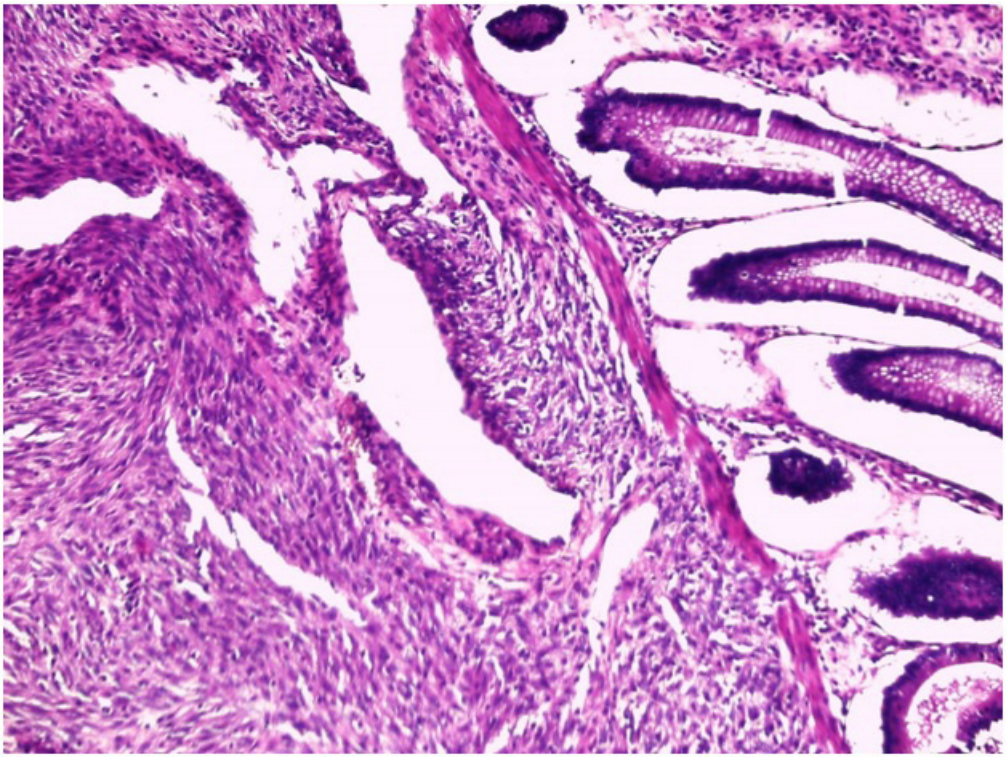

Figure2: microscopy [HEx20] medium power showing herringbone pattern made by interlacing bundles and crisscrossing fascicles.

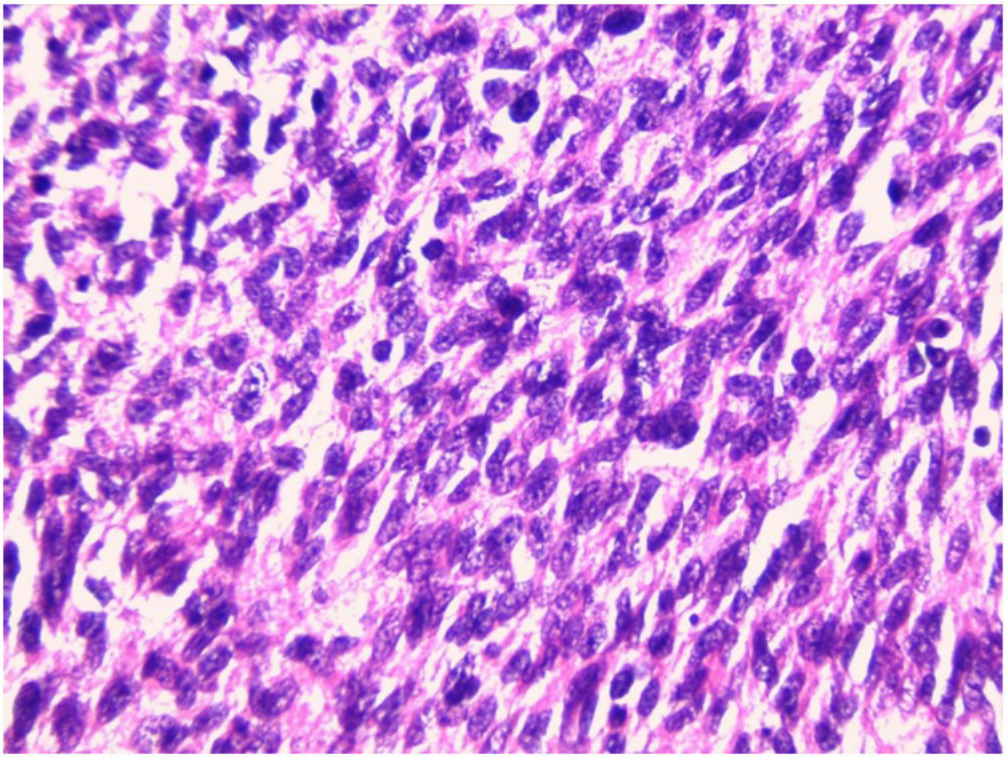

Figure 3: High power amplification[HEx40] demonstrated hyperchromatic nuclei with little atypia, and some mitotic activity (6 mitotic figures per 50 HPF). there is no necrosis and no apoptosis.

5. Microscopic Examination

Histological examination of the resected tumor showed a spindle cells tumor. It was densely cellular most extensive in the ileal intestinal submucosa. This tumor was well circumscribed by a thin capsule, focally infiltrated. the tumor cells are organized in crisscrossing fascicles giving a herringbone pattern, tangled fascicles and some whorls. The cells have oval hyperchromatic nuclei and blurred cytoplasmic limits. they demonstrated little atypia, and some mitotic activity (6 mitotic figures per50 HPF) There were no apoptosis nor necrosis and no heterotopia in favor of Meckel diverticulum. No hemangiopericytoma pattern was observed. Tumor resection was complete. Therefore, this morphology suggested strongly a malignant stromal tumor. We performed an immunohistochemistry study on paraffin sections: the tumoral cells did not express Desmin, MyoD1 and Myogenin, excluding a rhabdomyosarcoma as the most frequent sarcoma of the childhood. They also did not express stromal tumor marquers (CD34, CD117, DOG1), nor Neurofilament and GFAP, neither SML and CKAE1AE3 or ALK. Only the vimentin was strongly expressed and a patchy weak expression of PS100. Thus, the diagnosis of an undifferentiated sarcoma giving rise to suspicion of congenital fibrosarcoma. Therefore, the need for a molecular testing been mandatory. The paraffin block has been sent to the department of pathology of the SAINT JUDE HOSPITAL (Memphis USA). The diagnosis of infantile congenital fibrosarcoma was supported by break apart fluorescence in situ hybridization performed at ST JUDE hospital showing rearrangement of ETV6 gene (ETV6-NTRK3). This is a very rare entity with few reports in literature.

6. Discussion

The IFS represent a very rare condition clinically and an exceptional mesenchymal tumor histologically. This tumor has a less aggressive behavior and it's characterized by a particular molecular alteration. The IFS affects males more than females with a sex ratio M/F: 3/1-4/1.the IFS may represent 24% of soft tissue sarcomas, but it accounts only for 5 to 10% of soft tissue sarcomas before 1 year [1-4]. The time of diagnosis is defined during the first year of life; however, cases are diagnosed before three months of life. Finally, about third cases are diagnosed at birth [5,7-9]. The majority of IFS (69.2%) have the same diagnostic chromosomal translocation (12;15) (p13; q25) than mesoblastic nephromas, that resulting in fusion of ETV6 (TEL)gene or chromosome 12 with the neurotrophin-3 receptor NTRK3(TRKC). on chromosome 15. These make the IFS more prevalent with the kidney [10-13].

Demonstrating ETV6/NTRK3 transcript needs RT-PCR or FISH on paraffin embedded tumor tissue [14]. Virtually, the IFS may occur anywhere of the body whereas the gut is an exceedingly rare location: only 17 cases of IFS of the gut have been reported till now [4-11]. majority of them occurred in the small intestine. The differential diagnosis of intra-abdominal-located -IFS are all intra-abdominal tumors, any remainder malignant mesenchymal tumors or sarcomas [2] hemangioma or infantile fibromatosis [2,3] and MECKEL's diverticulum [3,5]. Clinically, in majority of cases (almost 70%), IFS occurs in the limbs. However, some unusual ectopic cases have been reported as the trunk [4,5] and the neck [4]. The most reported complication was the acute abdominal perforation. Through BEREB-

BI-study cohort, it seems clearly that abdominal IFS are frequently associated with a digestive perforation (54%), or a bowel obstruction (30%) [3]. Diagnosis could be hided by a big mass that led to perforation and stenosis. In gross examination the measured diameter of the tumor ranged between 1 to 10cm [3,5]. Histological examination shows a characteristic herringbone pattern made by relatively-uniform spindle cells with elongated nuclei arranged in interlacing fascicles. In histological analysis; IFS shows a herringbone pattern made of layers of closely packed spindle-shaped cells arranged in fascicles and bundles. The nucleus is hyperchromatic and the mitotic activity is higher. Vascularization is rich. Sometimes, areas of hemorrhage, necrosis or inflammation can be observed [4,7,10].

The immunohistochemical study shows that tumor cells express only vimentin, while muscular markers are not expressed (such as desmin), as found in our case study [10]. The fusion transcript ETV6-NTRK3(FT) associated with the t (12;14) (p13; q25) translocation, is the best biomarker to establish diagnosis, and which allow also to differentiate IFS from infantile fibromatosis, adult fibrosarcoma and other spindle-cells-tumors [10,16-18]. After complete resection the patient is doing well during several controls extended on six months. The interest of this case lies in its uncommon intestinal location, and its diagnostic timeline and revealing symptoms, the tumor can develop in size without symptoms, or ongoing un acute complication like in our case. The authors declare that there are no conflicts of interest regarding the publication of this article.

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