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Mesenchymal Hamartoma Liver Showing Malignant Transformation to Undifferentiated Embryonal Sarcoma- A Case Report

Dr. Veena Vijayan[1], Dr. Priya V S[2], Dr. Lilarani Vijayaraghavan[3]

[1]Junior resident, Department of Pathology, Government Medical College, Thiruvananthapuram, Kerala, India

[2]Assistant professor, Department of Pathology, Government Medical College, Thiruvananthapuram, Kerala, India

[3]Professor and Head of the Department, Department of Pathology, Government Medical College, Thiruvananthapuram, Kerala, India

ABSTRACT

Mesenchymal hamartoma of liver is the third most common hepatic tumor in pediatric age group. Typically, it presents as a large multicystic mass in children. Gradually they get increases in size and some can undergo incomplete spontaneous regression and very rarely, few have shown malignant transformation to undifferentiated (embryonal) sarcoma. Here, we report a 6 year old female child with no relevant personal and family history, who presented with abdominal distension and vomiting. Radiology revealed a multiloculated large cystic lesion in the right lobe of liver with a possibility of hemangioendothelioma on ultrasonography and CT abdomen was suggestive of mesenchymal hamartoma of liver. Following worsening of symptoms, partial excision and marsupialisation of the cyst was performed. Histologically, it was diagnosed as mesenchynal hamartoma with undifferentiated embryonal sarcomatous component and immunohistochemical confirmation done.

KEYWORDS: Mesenchymal hamartoma, Undifferentiated embryonal sarcoma.

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INTRODUCTION

The mesenchymal tumors of liver in neonates, infants and young children are hemangioma, mesenchymal hamartoma, infantile hemangioendothelioma, inflammatory myofibroblastic tumors and rarely undifferentiated embryonal sarcomas.[1] Among them, infantile hemangioma and mesenchymal hamartoma of liver are the most common.[2] They may be identified incidentally or may present with an alarm as large, rapidly growing abdominal mass. Liver mesenchymal hamartoma(LMH) is third most common neoplasm of liver in children after hepatoblastoma and infantile hemangioma. Pathogenesis of the lesion is still unknown, although association has been seen with abnormal mesenchymal congenital anomaly.[1] Very few cases of mesenchymal hamartoma liver have shown malignant transformation to undifferentiated sarcoma. Diagnosis is made based on clinical signs and imaging, but usually a biopsy or surgical resection is required to reach an accurate diagnosis. Tumor is managed in various modalities like enucleation, marsupialisation and liver resection.[3]

CASE REPORT

We here describe a case of mesenchymal hamartoma liver showing malignant transformation to undifferentiated embryonal sarcoma.

A 6 year old female patient with no relevant personal and family history, was presented with vomiting and abdominal distension of having a duration of 4 hrs. There was no other symptomatology like jaundice and loose stools. On physical examination, afebrile and anicteric with good general condition, good skin colour, but having distended abdomen with a large palpable mass measuring 10x10 cm in the right hypochondrium, epigastrium and left hypochondrium, which moves with respiration and was hard in consistency. Laboratory studies at the time of first presentation showed in table 1.

Table 1. Laboratory studies of the child at the time of presentation.

CBC : Hb – 10.9 gm%, WBC- 8000 cells/mm^3, Platelet count – 1.6 lac
LFT : Total S. Bilirubin/direct bilirubin- 1.2/0.4 mg/dL, SGOT/PT- 157/133 U/L, S. ALP- 532 U/L, GGT – 328 U/L
RFT : Urea/creatinine – 14/0.5 mg/dL, Total protein/albumin- 6.8/3.9 g/dL, Na/K – 135/3.7 mmol/L
TFT : S.T3- 116 ng/dL, S. T4- 12.7 ng/dL, S. TSH- 3.78 mIU/L
Coagulation studies : PT/INR : 1.06, aPTT – 29.9 sec, BT and CT – WNL
Viral markers : NR

An ultrasound of abdomen was performed as the initial imaging test, revealed a multiloculated cystic lesion with possibility of hemangioendothelioma. The plane and contrast enhanced CT abdomen showed a large multiloculated cystic mass with multiple enhancing internal septations, measuring 13x13x13 cm, with moderate enhancement in the venous plane in the right lobe of liver- segment VIII and with no calcification suggestive of mesenchymal hamartoma. Severe mass effect noted by displacement of pancreas and stomach towards left side. Also right heart ventricle compresses along the posterior margin. 1 week later, child developed severe acute abdominal pain, increase in the abdominal distension with respiratory distress. On physical examination, peritoneal fluid collection noted. CT abdomen repeated – a large multiloculated cystic lesion arising from the right lobe of liver, measuring 13x13x13 cm. Left lobe liver appears normal. Mass was displacing right kidney inferiorly. No significant internal vascularity noted within the mass. No evidence of calcification also. Image findings were suggestive of mesenchymal hamartoma liver(Fig.1&2).

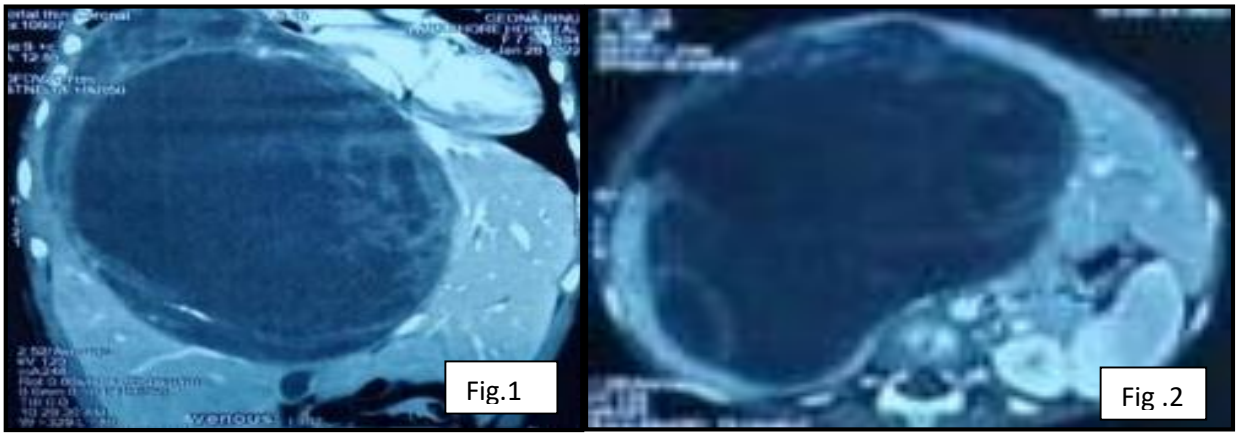


Fig.1 & 2. CT abdomen showing a large cystic lesions in the liver with internal multiseptations in the right lobe, with mass effect.

Surgical intervention was performed through a bilateral subcostal incision laparotomy. Identified a multiseptate cystic lesion measuring 15x15 cm with friable tissues and hematoma involving segments VII,VIII, IV,V extending to segment III, measuring 300x400 ml in volume. Tumor extends from upper part of retrohepatic IVC, hepatic vein and portal vessels seen adherent to wall of cyst. So partial excision of cyst with deroofing and marsupialisation was done. Intraoperative and post operative days were uneventful.

Gross - We received the whole specimens in multiple fragments of cyst wall and portions of liver tissue with areas of hemorrhage and slimy areas.

Histology - A cystic neoplasm composed of elongated and tortuous ductular structures surrounded by abundant loose fibromyxoid stoma was seen(Fig. 3&4). Also noted a cellular area adjacent to and intermingled with the above lesion and it composed of cells arranged in sheets(Fig. 5&6). Individual cells are plump spindly to epithelioid with pale eosinophilic and clear cytoplasm, oval plump vesicular nucleus and coarse chromatin(Fig.7&8). Mitotic figures- 30/10hpf noted in mitotically active areas. Atypical MF noted. Foci of extramedullary hematopoiesis were seen(Fig. 9). Scattered multinucleated giant cells was also seen.

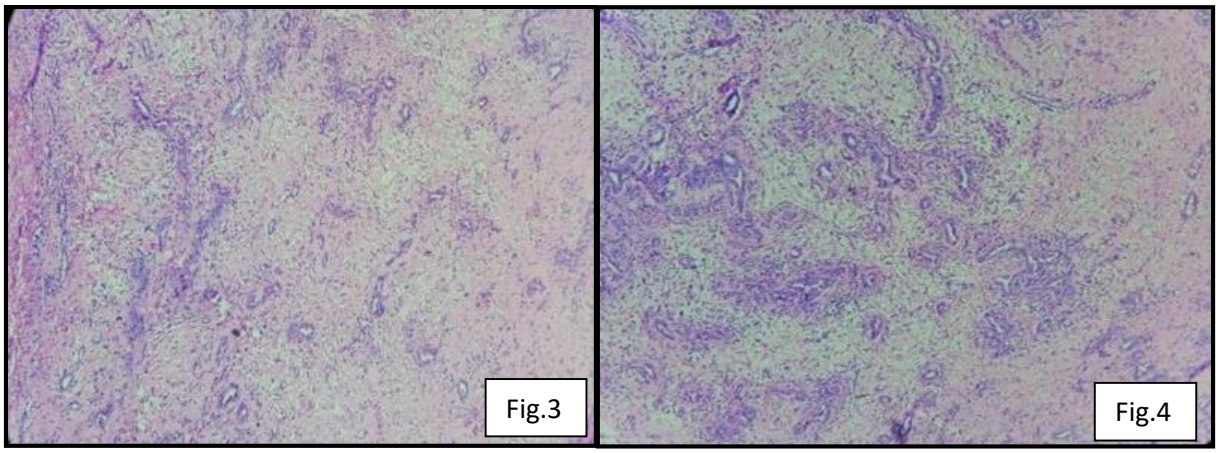


Fig.3(10x) & Fig.4.(10x): Histologically, neoplasm composed of elongated and tortuous ductular structures surrounded by loose fibromyxoid stroma

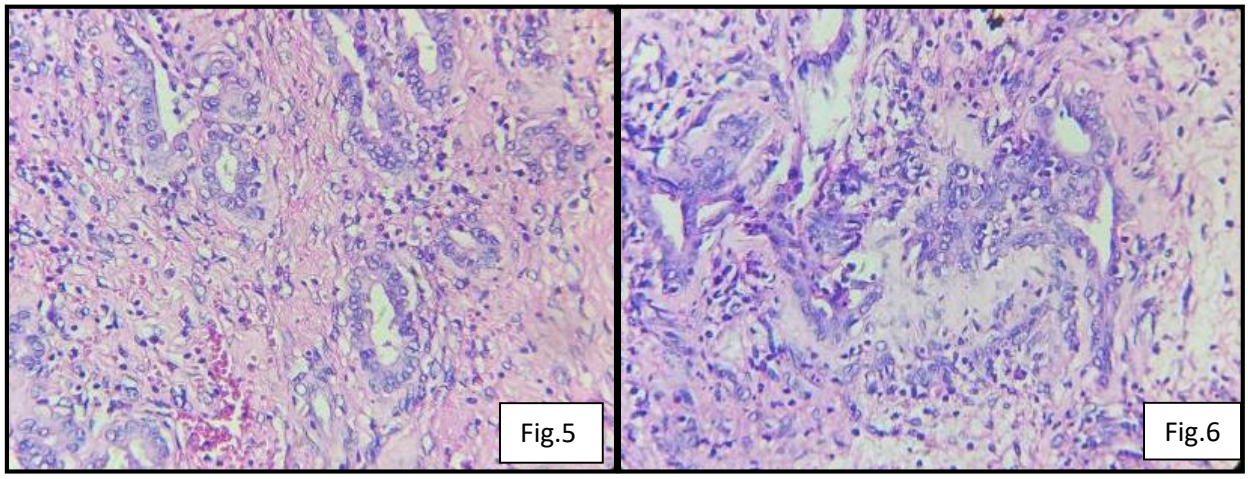


Fig.5(40x) & Fig.6(40x) : Elongated and tortuous ductular structures surrounded by loose fibromyxoid stroma

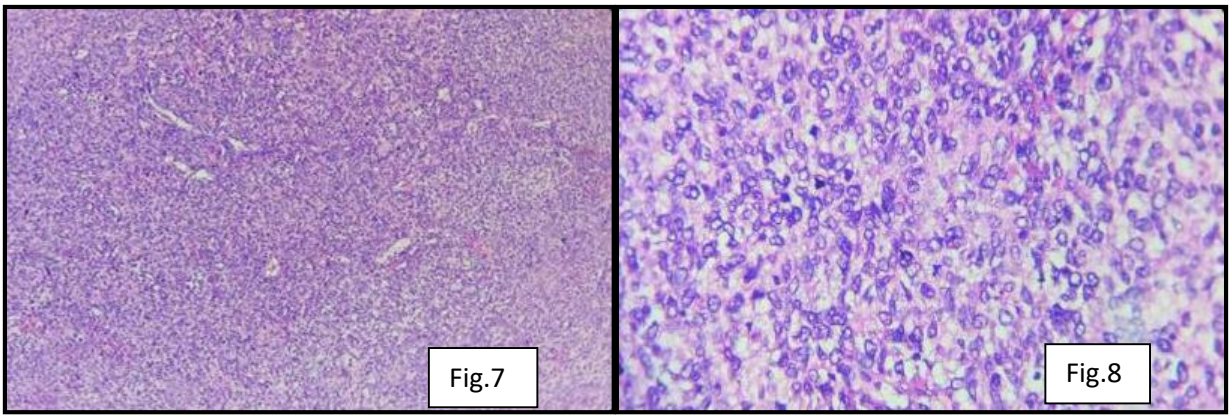
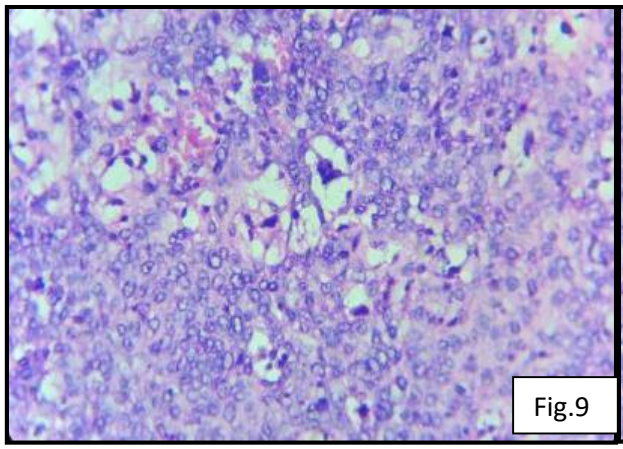


Fig.7(10x) & Fig.8(40x) : Cellular area of neoplasm composed of cells arranged in sheets



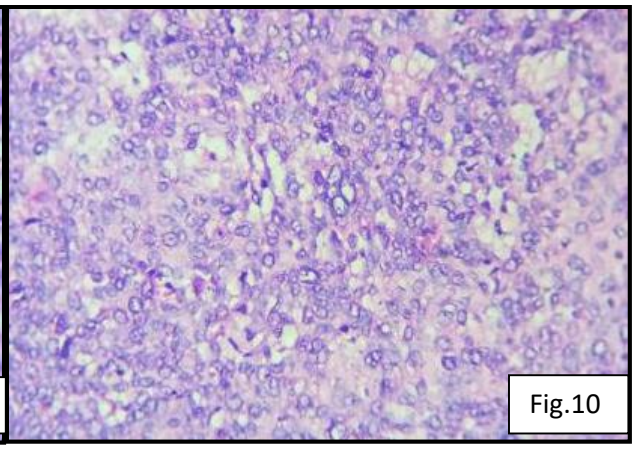


Fig.9(40x) & Fig.10(40x) : Cellular area composed of plump spindly to epithelioid cells with pale eosinophilic and clear cytoplasm, oval plump vesicular nucleus and coarse chromatin. Mitotic figures noted

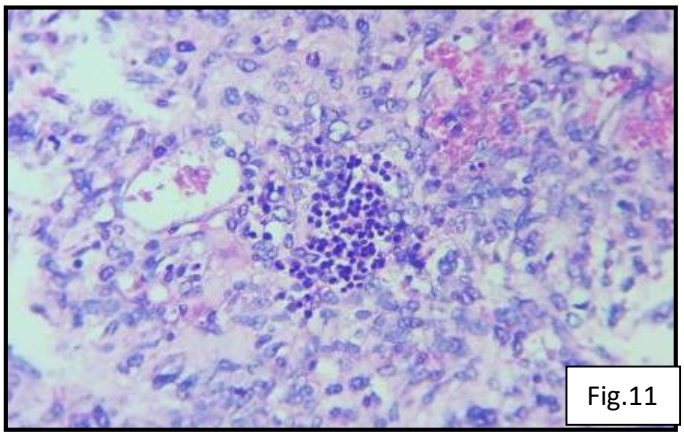
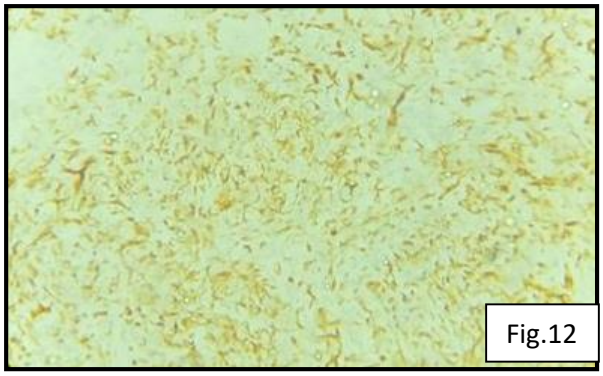


Fig.11: Foci of extramedullary hematopoiesis(400x)

Immunohistochemical examination : Neoplastic cells are positive by desmin(Fig.12), Bcl-2(Fig.13) and Ki-67 >30%(Fig.14). CK7 highlighted the disorganized bile duct proliferation. Neoplastic cells are negative for CK7(Fig.15), Hepar-1(Fig.16) and CK AE1/AE3(Fig.17). Neoplastic cells are negative for S100 (Fig.18), neoplastic cells are negative for Myogenin(Fig.19) and Myo-D1(Fig.20)

A diagnosis of mesenchymal hamartoma liver showing malignant transformation to undifferentiated embryonal sarcoma was given.



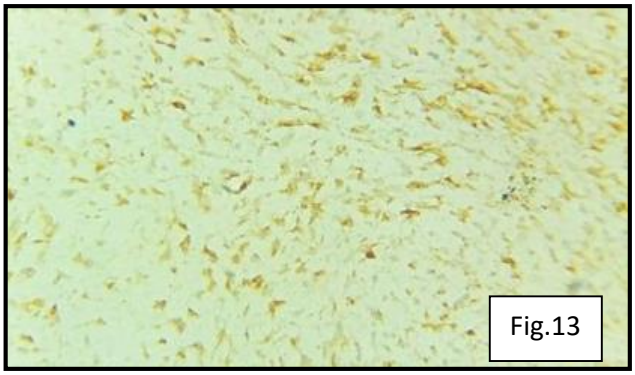


Fig.12: Neoplastic cells are positive immunostaining for desmine & Fig.13: Positive immunostaining for Bcl2

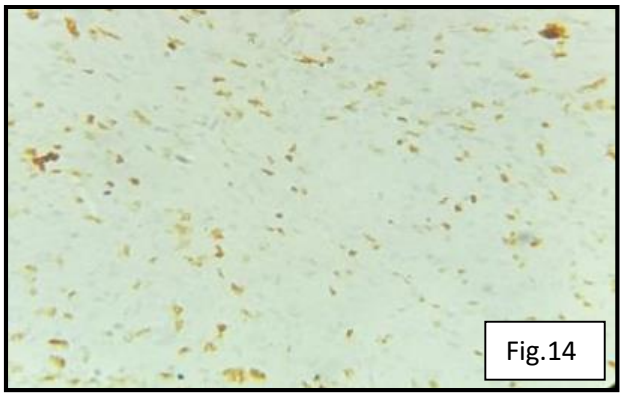


Fig.14: Ki-67 is >30% in the cellular areas

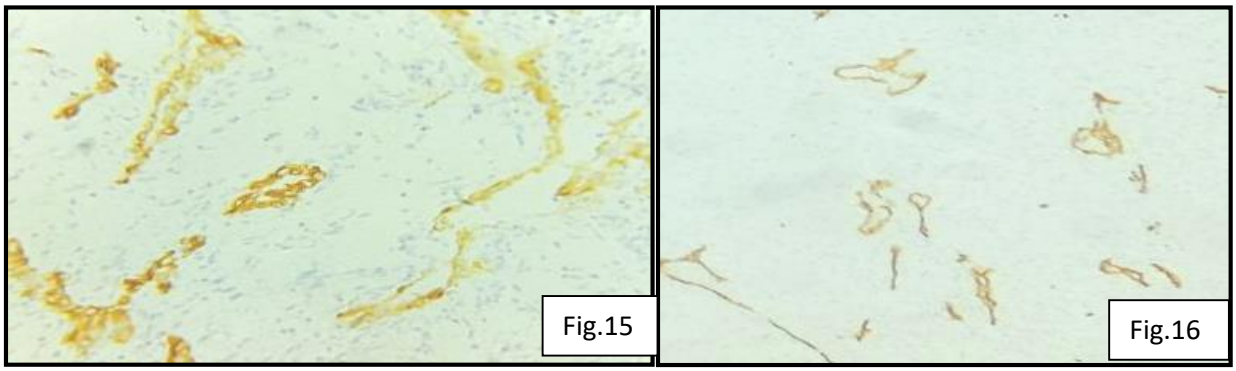


Fig.15: Neoplastic cells are negative for CK 7 & Fig.16: Negative for Hepar-1

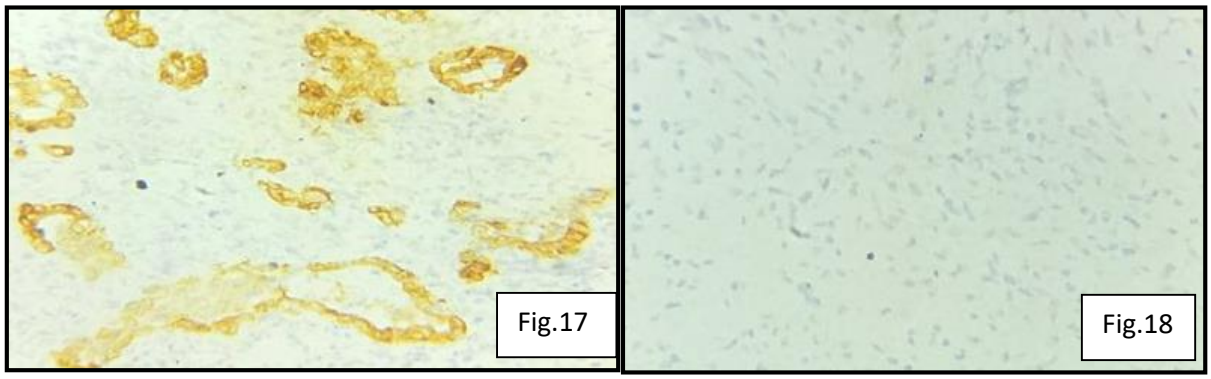


Fig.17: CK AE1/AE3 taken by ductal cells & Fig.18: S100 is negative by neoplastic cells

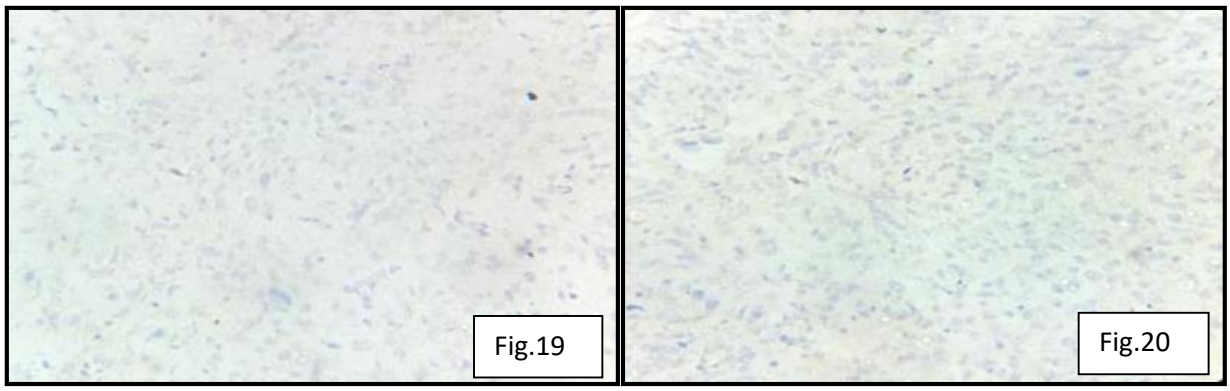


Fig.19: Neoplastic cells are negative for myogenin & Fig.20: Negative for myo-D1

Child was refered to an oncology center with the report of mesenchymal hamartoma with malignant transformation to undifferentiated embryonal sarcoma and planned for liver transplantation. Took 1 cycle of chemotherapy. After few days, child developed worsening of symptoms with marked abdominal distension and pain, marked ascitis and associated

respiratory distress. She admitted and underwent resection of tumor along with removal of 90% of liver. Intra-operative and post-operative days are uneventful. Now, child is improving and active. Remaining liver tissue is providing adequate liver functions.

DISCUSSION

Liver mesenchymal hamartoma (LMH) is a rare benign tumor, occurring primarily in the pediatric population, with occasional risk of malignancy.[3] It is the third most common tumor of the liver in children after hepatoblastoma and infantile hemangioma. It was first described by Maresh in 1903 and Edmond was the first to use the term "mesenchymal hamartoma" of the liver on postulating that the lesion might result from a failure in the normal development of the embryonic fetal liver or might represent a degenerative change of an accessory lobe.[4] It was previously considered as a developmental anomaly of ductal plates, while newer insights into genesis of LMH are in progress like ischemia, toxic metabolites and a true neoplastic process. Previous understanding of a purely benign nature of the tumor is being abrogated by a real malignant transformation.[5] Grossly, tumor may be solid or cystic and accumulation of fluid within the cyst leads to increase in size. Histologically, LMH composed of disorderly arranged bile ducts, hepatic parenchyma and mesenchymal tissue.

The typical presentation is either asymptomatic or rapid abdominal distention with a palpable mass on physical examination. The rapid expansion of the tumor is believed to be due to degeneration of the mesenchyme and accumulation of fluid within the tumor.[6] Symptoms like vomiting, fever, constipation, diarrhea, and weight loss are uncommon. The radiological appearance is one of a large, uni- or multicystic, avascular mass occupying part of the liver. It is having an indolent behavior and carries excellent prognosis.[7] Surgical resection has been the standard treatment for this tumor. Marsupialization is also proposed in cases not amenable to resection. Management depends on the location of the lesion. A link between mesenchymal hamartoma and undifferentiated embryonal sarcoma has been postulated on the basis of overlapping clinicopathological features.[8] Undifferentiated embryonal sarcoma is a rare tumor typically occurs in children 6 to 10 years old. It is the most common hepatic malignant mesenchymal tumor in children. Also arise as a malignant transformation from mesenchymal hamartoma of liver which is exceptional. They share common genetical abnormality of tranlocation involving 19q13.[9] Histologically characterized by anaplastic cells with eosinophilic cytoplasm and bizarre nuclei and frequent atypical mitosis and spindling in a myxoid stroma. By immunohistochemistry, neoplastic cells are positive for desmin and BCl2, while negative for Hepar 1, S-100 and myogenin.[10,11]

Prognosis of LMH is good, while that of undifferentiated embryonal sarcoma is poor. Complete surgical excision is generally done and offers the best outcome. Improved outcomes have reported with the use of adjuvant preoperative and postoperative therapy.

CONCLUSION

Mesenchymal hamartoma is one of the causes of cystic mass in the liver, which is benign in fashion. But rarely can arise a malignant transformation from this benign lesion, most commonly to undifferentiated embryonal sarcoma. This transformation may occur several years after an incomplete resection of the lesion and is having a poor prognosis. So a prompt surgical management is absolutely indicated.

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