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HEPATOBLASTOMA IN EDWARD SYNDROME – A CASE REPORT WITH RADIOLOGICAL REVIEW

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ABSTRACT

Trisomy 18 (Edward syndrome) is a rare entity with incidence of 1 in 3000 to 1 in 7000 births. Less than 10 % of patients survive beyond 1 year of age. This influences the fact that malignant tumors are rarely reported in association with Edward syndrome. Hepatoblastoma is a rare tumor of infancy and childhood with an annual incidence rate of 1.8 per million in children less than 15 years of age. Both trisomy 18 and hepatoblastoma are rare conditions. Hence, a child with trisomy 18 developing hepatoblastoma is very rare. Existence of hepatoblastoma in trisomy 18 indicates a significant association. Trisomy 18 probably potentiates development of hepatoblastoma. Till now 8 cases of hepatoblastoma with trisomy 18 have been published. We report a case of a 9 month old female child of Edward syndrome with PDA and VSD referred for ultrasound scan of abdomen for failure to thrive. USG showed enlarged right hepatic lobe due to a well defined mixed echoic predominantly hypoechoic solid lesion showing mild central vascularity on color doppler. CECT abdomen showed a large well defined spherical, solid, predominantly hypodense mass in right hepatic lobe involving segments V to VIII on plain study, showing near homogeneous contrast enhancement on portal venous phase and appearing hypodense with respect to adjacent hepatic parenchyma in delayed phase with few central non-enhancing areas of necrosis.

KEYWORDS: Hepatoblastoma, hypodense, Doppler.

CASE REPORT

A 9 month old female child was referred for ultrasound scan of abdomen for failure to thrive. She gave history of recurrent fever and lower respiratory tract infection. She was a known case of Edward syndrome with Patent ductus arteriosus and Ventricular septal defect. 2D ECHO showed situs solitus levocardia, restrictive perimembranous VSD (Left → Right), 3 mm PDA (Left → Right) with gradient of 80 mm of Hg. Aortic arch was normal. Left atrium and left ventricle were dilated with no evidence of pulmonary arterial hypertension and good biventricular function. She had mild anaemia with haemoglobin – 10 grams/dl. Rest of the haemogram, renal function tests and liver function tests were normal. USG Abdomen (Figure 1) showed enlarged right hepatic lobe due to a well defined mixed echoic predominantly hypoechoic solid lesion of size approx. 4.6 x 3.7 cms, showing mild central vascularity on color Doppler with splaying of adjoining right and middle hepatic veins and portal vein radicles (Figure 2). No calcification was noted. Left hepatic lobe was normal. There was no dilatation of intrahepatic biliary radicles. Rest of the abdomen was normal. Her serum alfa-fetoprotein levels were markedly raised, i.e. > 30000 IU/ml (Normal reference range: 0.5 - 5.5 IU/ml).

Plain and contrast CT of abdomen (Figure 3) showed hepatomegaly with a large well defined spherical, solid, predominantly hypodense mass in right hepatic lobe, measuring approx. 4.8 x 4.2 x 4.7 cms in its antero-posterior , transverse and cranio-caudal dimensions respectively, involving segments V to VIII. The mass was reaching laterally upto hepatic surface and was causing splaying of adjoining hepatic veins and portal vein radicles. No calcification or fat attenuation was seen within the lesion (Figure 3A). On post contrast study, the mass showed near homogeneous contrast enhancement (CT value : 100 – 110 HU), similar to that of normal hepatic parenchyma on portal venous phase (Figure 3B and 3D) and appeared hypodense with respect to adjacent hepatic parenchyma in delayed phase (Figure 3C and 3E). Few central non-enhancing hypodense areas noted were suggestive of necrosis. Possibility of hepatoblastoma was given. In view of markedly raised levels of serum alfafetoproteins, hepatoblastoma was confirmed. Parents refused liver biopsy and further treatment. The patient died 1 month after the admission.

Chromosomal analysis (Karyotyping): of peripheral blood culture was consistent with mosaic - Edward syndrome. [mos 47, XX, +18 (25)/46, XX (5)].

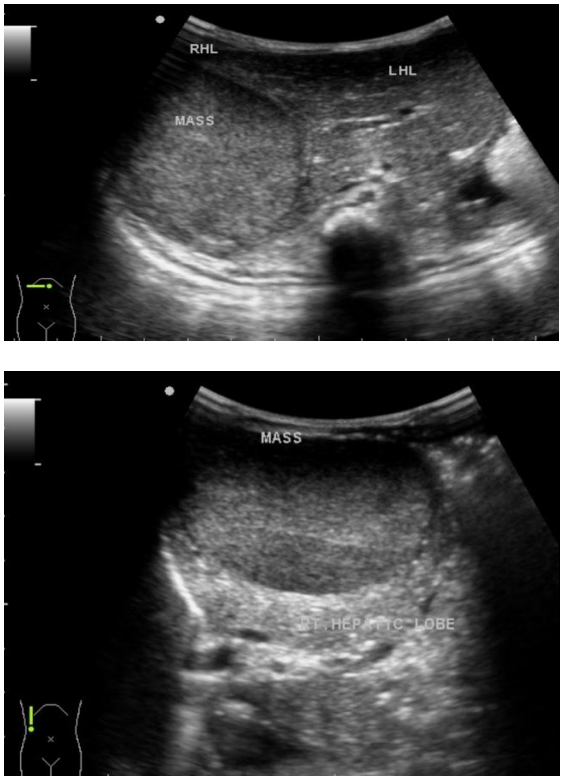


Figure 1: USG liver – Axial (A), Sagittal (B) planes

USG liver (axial & sagittal plane) showing well defined hypoechoic solid mass in right hepatic lobe with no calcification.

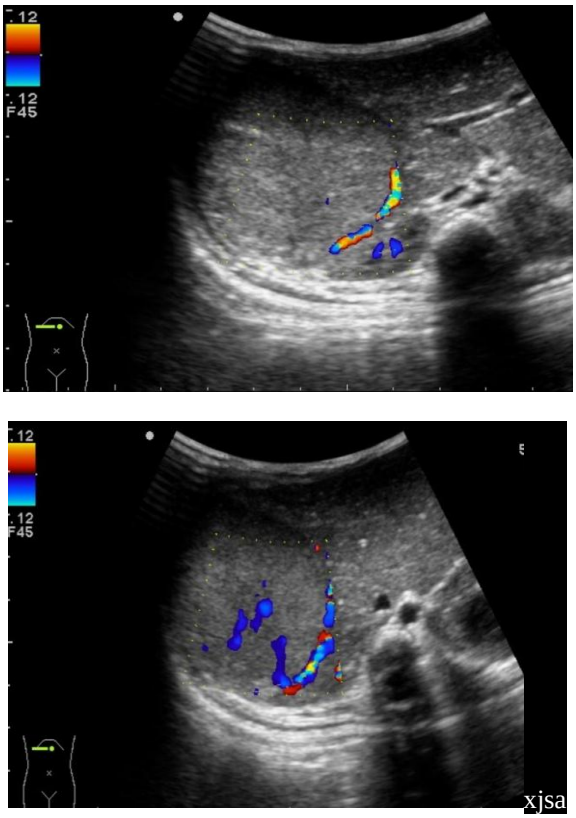
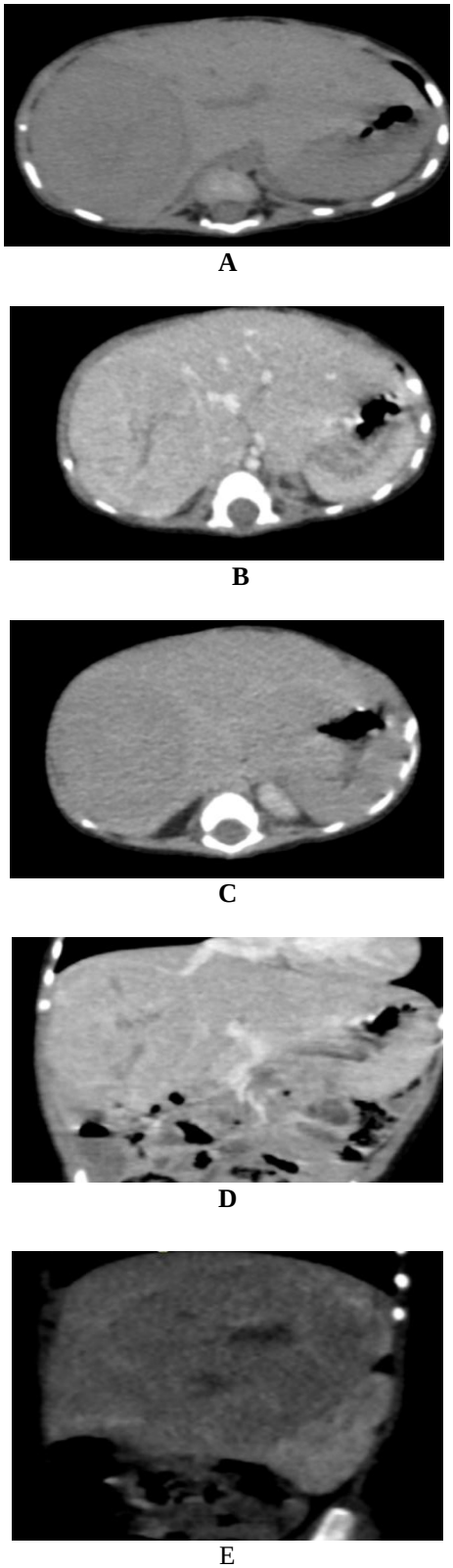


Figure 2: USG liver with Doppler – Axial (A), Sagittal (B) planes

USG liver with Doppler (axial & sagittal plane) showing well defined hypoechoic solid mass in right hepatic lobe causing splaying of hepatic veins and portal vein radicles on doppler with mild central vascularity.



A

B

C

D

E

Figure 3: CT abdomen – Axial (A, B, C), coronal venous phase (D), sagittal delayed (E)

CT abdomen (plain, venous and delayed phases) showing well defined solid mass in right hepatic lobe appearing hypodense on plain CT (Figure 1A), nearly isodense with adjacent hepatic parenchyma in portal-venous phase (Figure 1B, 1D) and appearing hypodense in delayed phase (Figure 1C, 1E) with central non-enhancing hypodense areas of necrosis.

INTRODUCTION

Trisomy 18 (Edward syndrome) is a rare entity with incidence of 1 in 3000 to 1 in 7000 births. Less than 10% of patients survive beyond 1 year of age. This influences the fact that malignant tumors are rarely reported in association with Edward syndrome. Till now 8 cases of hepatoblastoma with trisomy 18 have been published.[1] This is 9th case of hepatoblastoma in a patient with trisomy 18. All the published cases till now were women, possibly due to high preponderance of females among the children with Edward syndrome and longer survival of females with trisomy 18, compared to males. Both trisomy 18 and hepatoblastoma are rare conditions. Hence, a child with trisomy 18 developing hepatoblastoma is very rare. Existence of hepatoblastoma in trisomy 18 indicates a significant association. Trisomy 18 probably potentiates development of hepatoblastoma. To recognize real frequency of hepatoblastoma in Edward's syndrome, careful clinical and post-mortem studies are needed as some patients may die from different causes with unrecognizable hepatoblastoma.[1]

DISCUSSION

Trisomy 18 also called Edward syndrome is a chromosomal disorder due to presence of an extra chromosome 18, either full, mosaic trisomy or partial trisomy 18q. It is 2nd common autosomal trisomy syndrome after trisomy 21. Its live born prevalence is estimated as 1 in 6000 to 1 in 8000, but its overall prevalence increases with increasing maternal age. The first reported infant of Edward syndrome was described in 1960 by Edwards et al. and Smith et al.[2,3] It has recognizable pattern of major and minor anomalies with increased risk of neonatal and infant mortality and significant cognitive and psychomotor disability. Trisomy 18 pregnancies have a higher risk of foetal loss and still birth.[4,5] Currently most diagnoses are made in prenatal period based on screening by maternal age, maternal serum marker screening and amniocentesis, followed by pregnancy termination in a significant percentage of cases.[6] The prevalence of Edward's at birth is higher in females as compared to males (F : M % = 60.4). Frequency of foetal loss is higher for males as compared to females[4,5] Live born females showed better survival compared to males.[7] Trisomy 18 results from full, mosaic, or partial trisomy 18q. 94% of cases are complete/full trisomy 18. The extra chromosome is present due to non-dysjunction. The frequency of non-dysjunctional errors increases with advancing maternal age. Children with trisomy 18 have IUGR, microcephaly, short stature, mental retardation, cranio-facial abnormalities (camptodactyly, overlapping fingers, nail hypoplasia), short sternum, congenital heart diseases, horse shoe kidney, omphalocoele.

Hepatoblastoma is a rare tumor of infancy and childhood with an annual incidence rate of 1.8 per million in children less than 15 years of age. Majority are diagnosed before age of 2 years in an otherwise normal children with 1.4 : 1 to 2: 1 predominance in males.[1] It sometimes occurs in association with other congenital malformations especially Beckwith-Wiedemann syndrome, Gardener syndrome, Familial adenomatous polyposis, type Ia glycogen storage diseases (Von Gierke's disease and Edward syndrome).[1,8]

8 cases of hepatoblastoma in children with Edward's have been published since 1987, when Dasouki and Barr reported the first case. Maruyama et al. carried out review of published cases. All the cases were girls. Half of these cases were older than 1 year at the time of detection of hepatoblastoma. 5 of them has karyotype 47,XX,+18 in another one, chromosomal analysis of peripheral blood culture showed mosaic trisomy 47, XX, +18/46, XX (5 : 1).[1] Bove et al. mentioned a case of 1 year old boy, mosaic for trisomy 18.[9] Epithelial type hepatoblastoma with different histological patterns was diagnosed in all patients.[8]

CONCLUSION

Hepatoblastoma is a common hepatic tumour in paediatric age group. Existence of hepatoblastoma in trisomy 18 indicates a significant association. Trisomy 18 probably potentiates development of hepatoblastoma. This is 9th case of hepatoblastoma in Edward syndrome reported till date.

REFERENCES

1. Kitanovski L, Ovcak Z, Jazbec J. Multifocal hepatoblastoma in a 6-month-old girl with trisomy 18: a case report. J Med Case Rep., 2009 Jun 23; 3: 8319 PMID: 19830224.
2. Edwards JH, Harnden DG, Cameron AH, Crosse VM, Wolff OH. A new trisomic syndrome. Lancet., 1960 Apr 9; 1(7128): 787-90. PMID: 13819419.
3. SMITH DW, PATAU K, THERMAN E, INHORN SL. A new autosomal trisomy syndrome: multiple congenital anomalies caused by an extra chromosome. J Pediatr., 1960 Sep; 57: 338-45. PMID: 13831938.
4. Morris JK, Savva GM. The risk of fetal loss following a prenatal diagnosis of trisomy 13 or trisomy 18. Am J Med Genet A., 2008 Apr 1; 146A(7): 827-32. PMID: 18361449.
5. Won RH, Currier RJ, Lorey F, Towner DR. The timing of demise in fetuses with trisomy 21 and trisomy 18 Prenat Diagn., 2005 Jul; 25(7): 608-11. PMID: 16032775.
6. Irving C, Richmond S, Wren C, Longster C, Embleton ND. Changes in fetal prevalence and outcome for trisomies 13 and 18: a population-based

study over 23 years. J Matern Fetal Neonatal Med., 2011 Jan; 24(1): 137-41. PMID: 20384468.
7. Cereda A[1], Carey JC. The trisomy 18 syndrome. Orphanet J Rare Dis., 2012 Oct 23; 7: 81. PMID: 23088440.
8. Chung EM, Lattin GE Jr, Cube R, Lewis RB, Marichal-Hernández C, Shawhan R, Conran RM. From the archives of the AFIP: Pediatric liver masses: radiologic-pathologic correlation. Part 2. Malignant tumors. Radiographics., 2011 Mar-Apr; 31(2): 483-507. PMID: 21415193.
9. Bove KE, Soukup S, Ballard ET, Ryckman F. Hepatoblastoma in a child with trisomy 18: cytogenetics, liver anomalies, and literature review. Pediatr Pathol Lab Med. 1996 Mar-Apr; 16(2): 253-62. PMID: 9025831.