

See discussions, stats, and author profiles for this publication at: <https://www.researchgate.net/publication/26320953>

Ataxia Telangiectasia with Hepatocellular Carcinoma

Article in Indian pediatrics · July 2009

Source: PubMed

CITATIONS

2

READS

23

2 authors:



M. M. Patil

BLDE University

13 PUBLICATIONS 57 CITATIONS

[SEE PROFILE](#)



Shankar Patil

BLDE University

18 PUBLICATIONS 146 CITATIONS

[SEE PROFILE](#)

Ataxia Telangiectasia with Hepatocellular Carcinoma

Ataxia telangiectasia(AT) is heralded by ataxia beginning around 2 years and progresses to loss of ambulation by adolescence. The telangiectasia becomes evident by mid childhood. Hematological and soft tissue malignancies are known to be associated with AT.

We came across a 10 year boy, a known case of AT, presenting with mass per abdomen since 1 month. A large painless firm to hard, smooth surfaced mass was felt in the epigastrium with hepatomegaly. USG revealed 9×9cm well defined mass in the left lobe of liver and two hyper echoic lesions in the right lobe of the liver. FNAC revealed poorly differentiated hepatocellular carcinoma (HCC). At the time of diagnosis of AT two years back, his serum alfa fetoprotein(AFP) was 56.9 ng/mL. Now his serum AFP is raised to 100000ng/mL, a substantial increase. He is hepatitis B and hepatitis C negative, liver function tests were normal.

After thorough search of literature we found only 3 reported cases of AT with HCC(1,2). But no case has ever been reported from India. Serum AFP is normally elevated in AT but generally not beyond the range of 300-400ng/mL. In HCC it increases to over 400-500ng/mL. Extremely elevated serum AFP could be a pointer towards underlying malignancy like hepatocellular carcinoma.

MM Patil and SV Patil
*Department of Pediatrics,
 BLDEA's Shree BM Patil Medical College and
 Research Center, Bijapur, Karnataka,
 India.
 E-mail: mmp076@yahoo.com.*

REFERENCES

1. Weinstein S, Scottolini AG, Loo SY. Ataxia telangiectasia with hepatocellular carcinoma in a 15-year-old girl and studies of her kindred. Arch Pathol Lab Med 1985; 109: 1000-1004.
2. Krishna Kumar G, Al Saadi A, Yang SS, McCaughey RS. Ataxia-telangiectasia and hepatocellular carcinoma. Am J Med Sci 1979; 278: 157-160.