

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 feto protein(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.

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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.