Case Report :

DIAGNOSIS OF ALPHA -1- ANTITRYPSIN DEFICIENCY BY SERUM PROTEIN ELECTROPHORESIS

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ABSTRACT

Alpha-1-antitrypsin deficiency is a hereditary disease leading to hepatitis and cirrhosis. It is the most common genetic cause of liver disease in children which must inherit the tendency from both parents to develop. It acquires the highest priority in the differential diagnosis in a child with chronic liver disease. In this case report we substantiate the role of serum protein electrophoresis, in diagnosing alpha-1-antitrypsin deficiency.

KEY WORDS: Alpha-1-antitrypsin deficiency, Serum protein electrophoresis.

INTRODUCTION

Alpha-1-antitrypsin deficiency is an autosomal recessive disorder with an incidence of 1:3000-7000 of the PIZ genetic type (1). There are more than seventy genetic variants. This disorder is characterized by reduced serum levels of alpha 1 antitrypsin, an antiprotease that provides the major defense for the lower respiratory tract against the ravages of neutrophil elastase, a powerful destructive protease. This results in emphysema. Either deficient or absent serum levels of alpha-1-antitrypsin are found in patients with early onset of emphysema. Decreased levels of this protein leads to damage, scarring and abnormal liver function. Fortunately, for reasons that are not understood, only 10-20% of the babies born with this deficiency have liver disease. Currently there is no treatment for this disease. However, certain complications can be avoided. Thus early detection of this dreadful and common disorder by serum protein electro-

phoresis in childhood by a simple technique will be of much use. Serum protein electrophoresis falls into this category of a simple and reliable test in diagnosing this disorder (2). In our study an effort was made to use this technique in a suspected case of alpha -1 -antitrypsin deficiency.

CASE REPORT

A year old male child was referred from a local hospital with complaints of abdominal distention for 20 days, fever for 15 days and swelling of both the legs for 3 days. He was also observed to develop bluish patches all over the body since eight days. The symptoms progressed gradually.

Five months earlier the child was admitted in a local hospital with episode of loose stools. As a routine investigation, chest x-ray was taken and later started on anti- tubercular therapy on a prophylactic basis. On examination bilateral pedal edema was present. Per abdomen examination revealed distended abdomen with positive fluid thrill. The investigations carried out are listed in table 1:

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Table 1. Preliminary investigations report of the patient

Investigation	Result	Normal Range
Haemoglobin	13.9g/dl	13-18g/dl
Prothrombin time	30sec	10-14sec
E.S.R.	02mm/hr	0-9mm/hr
Mantoux	Negative	

Pheripheral smear showed a Normocytic Normochromic blood picture with a relative neutrophilic showing a mild shift to the left.

Since E.S.R was low and mantoux being negative, antitubercular therapy was withdrawn and taking into consideration the increased prothrombin time, further investigations (in table 2 and table 3) to evaluate the cause of ascitis were carried out.

Table 2. Other Investigation report of the patient, Liver Function Tests.

Investigation	Result	Normal Range
Total Serum Protein	5.0gm/dl	6.0-8.0g/dl
Serum Albumin	3.4gm/dl.	3.2-5.5g/dl
Bilirubin (Total)	5.0mg/dl	0.1-1.0mg/dl
„ (Conj.)	2.0mg/dl	0.0-0.4mg/dl
Aspartate transaminase	249U/L	upto 37 U/L
Alanine transaminase	215U/L	upto 40 U/L
Alkaline Phosphatase	836U/L	240-770 U/L
γ - Glutamyl transferase	118U/L	11-50 U/L

Table 3. Ascitic Fluid Examination

Total Count	70
Differential Count	Neutrophils 1 , Lymphocytes 49/50 cells.
Gram's Stain	No pus cells.
Culture/Sensitivity	No Growth.

The above investigations point towards a chronic liver pathology (3). Considering the possibilities of various differential diagnoses of all causes of chronic liver pathology the following investigations were done to rule out each one of them.

Urine for Reducing Substance - Negative (To r/o Galactosemia)

Urine Protein - Trace. (To r/o nephrotic syndrome)

Ultra sound abdomen- Features suggestive of chronic parenchymal liver disease with splenomegaly , portal hypertension and ascites

Fundus examination - Normal (To r/o Wilson's disease)

Hepatitis B Antigen - Negative (To r/o infective hepatitis)

Hepatitis C Antigen- Negative (To r/o infective hepatitis)

Elisa for CMV- IgA and IgG- Negative (To r/o intra-uterine infection)

Serum protein electrophoresis - Pattern clearly indicates decreased alpha -1 Globulin suggesting alpha -1- antitrypsin deficiency. (fig. 1).

Thus a diagnosis of alpha -1- antitrypsin deficiency was established since alpha-1-antitrypsin constitutes the major part of the alpha 1 region in serum protein electrophoresis (4).

Fig1. Densitometric scanning of serum protein electrophoresis showing traces of protein in the alpha 1 region.

Conclusion

Availability of gel electrophoretic methods has revealed finer details in the sub fractions of serum proteins. This enabled us to identify the disorder associated with alpha -1- antitrypsin deficiency. At this stage techniques such as immunofixation, immunoprecipitation and typing of genetic variants by electrofocussing would be more appropriate (5).

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