ADOLESCENT WITH CYSTIC FIBROSIS ASSOCIATED LIVER DISEASE, DIABETES MELLITUS AND POOR COMPLIANCE TO TREATMENT- CASE REPORT

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Abstract

The clinical oucome of cystic fibrosis patients seemed to be define by the pulmonary condition, but, in the recent years, the liver disease became an important feature with potential impact on clinical outcome and life expectancy. Liver disease associated with cystic fibrosis is the second cause of death among CF patients.The aim of the paper is to present the case of an 15 years old boy, followed up in our CF Center with cystic fibrosis (CF) associated liver disease (LD).

The patient diagnosed with CF at the age of 3 month, presented for the first time in our clinic at 12 years old. He was registered in our centre with the following diagnosis: Cystic fibrosis case, complete form, ΔF508 homozygous, complicated with: *Pseudomonas aeruginosa* respiratory infection, medium obstructive pulmonary syndrome, bronchiectasis, associated liver disease, clubbing and failure to thrive. After starting the treatment with ursodeoxicholic acid, hepatomegaly decreased and liver tests normalized in 6 months. Two years after, patient was admitted in our clinic a change of mood and irritability associated with refusal of therapy, presenting visible abdominal circulation, hepato- and splenomegaly. The investigations confirmed portal encephalopathy, impaired glucose tolerance, modified liver texture on ultrasound examination, and magnetic resonance investigation confirmed the multilobular cirrhosis. Hi was discharge from hospital with dietary ant therapy recomandation. In evolution, the case complicated with: portal hypertension and portal encephalopathy. Diabetes mellitus developed, with conspicuous hyperglycemia, but the adolescent refused starting the insulinotherapy. Lung function deteriorated, obstructive syndrome accentuated and frequent exacerbations of pulmonary disease occured. Psychological issue exprimed in refuse of therapy, iritability. Beside the matters concerning the medical management of a case with LD and diabetes, the psychological issues related to age or hepatic encephalopathy occur, making more difficult the manner of patient`s life and worsening the course of disease.
Key words: cystic fibrosis, liver disease, diabetes, psychological issue, children

Introduction

Improved life expectancy and prolonged follow up of patients with cystic fibrosis have allowed direct observation of an increasing number of liver-related events. A broad spectrum of hepatobiliary manifestations have been recognized that include specific alterations ascribable to the underlying cystic fibrosis transmembrane regulator (CFTR) defect as well as lesions of iatrogenic origin or that reflect the effects of a disease process occurring outside the liver.

The aim of the paper is to present the case of an 16 years old boy, followed up in our CF Center with cystic fibrosis (CF) associated liver disease (LD).

Case presentation

The patient was diagnosed with CF at the age of 3 month , presented for the first time in our clinic at 12 years old.

Clinical status at the first admittance was characterized by: moderate failure to thrive, clubbing, medium pulmonary condition and hepatomegaly.

Laboratory investigations

Biochemical findings consisted in abnormal values of liver test (AST, ALT, γ-GT). Bilirubinemia, albuminemia and coagulation parameters were normal. Also investigation for hepatitis B virus, hepatitis C virus, cytomegalvirus , Epstein Barr and αfetoprotein were negative. Concerning the pulmonary disease, tests showed medium obstructive respiratory disease, bronchiectasis and *Pseudomonas aeruginosa* present in sputum culture. The genetic test performed revealed homozygous genotype ΔF508. Ultrasound examination showed increased echogenity liver texture, without signs for vascular decompensate, like portal hypertension.

Clinical course

After starting the treatment with ursodeoxycholic acid in dose 20 mg/ kilo/day, liver function tests normalised in 6 months and a decrease of hepatomegaly was registered. He was release at home with recommendation for treatment consisting in oral antibiotics for infection associated with aerosols therapy and physiotherapy for the pulmonary condition and liposoluble vitamins supplements.

Two years after, patient was admitted in our clinic presenting visible abdominal circulation, hepatomegaly and splenomegaly (fig.1).

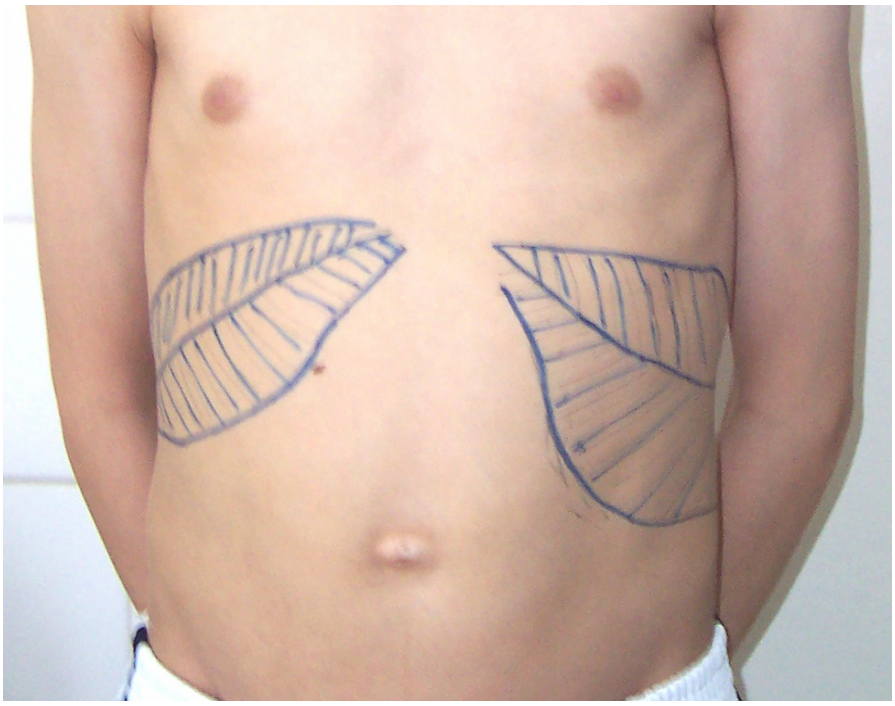

Fig.1.Spleno-hepatomegaly.

His mother observed a change of mood and irritability associated with refusal of any therapy. Investigations confirmed hepatic encephalopathy, with supression of electric rithm, impaired glucose tolerance, modified liver texture on ultrasound examination (fig.2), and magnetic resonance examination (MRI) confirmed the multilobular cirrhosis and splenomegaly with portal hypertension (fig.3). Functional changes of liver was remarqued at scintigraphy, with unhomogenous captation of reactive substance.(fig.4).

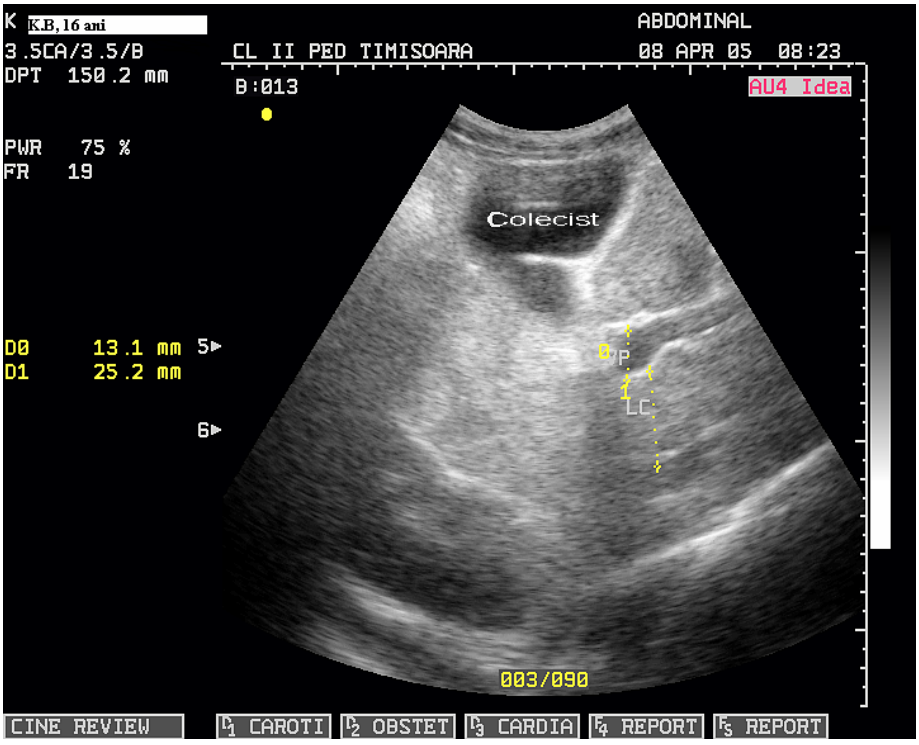

Fig.2-Ultrasound examination.

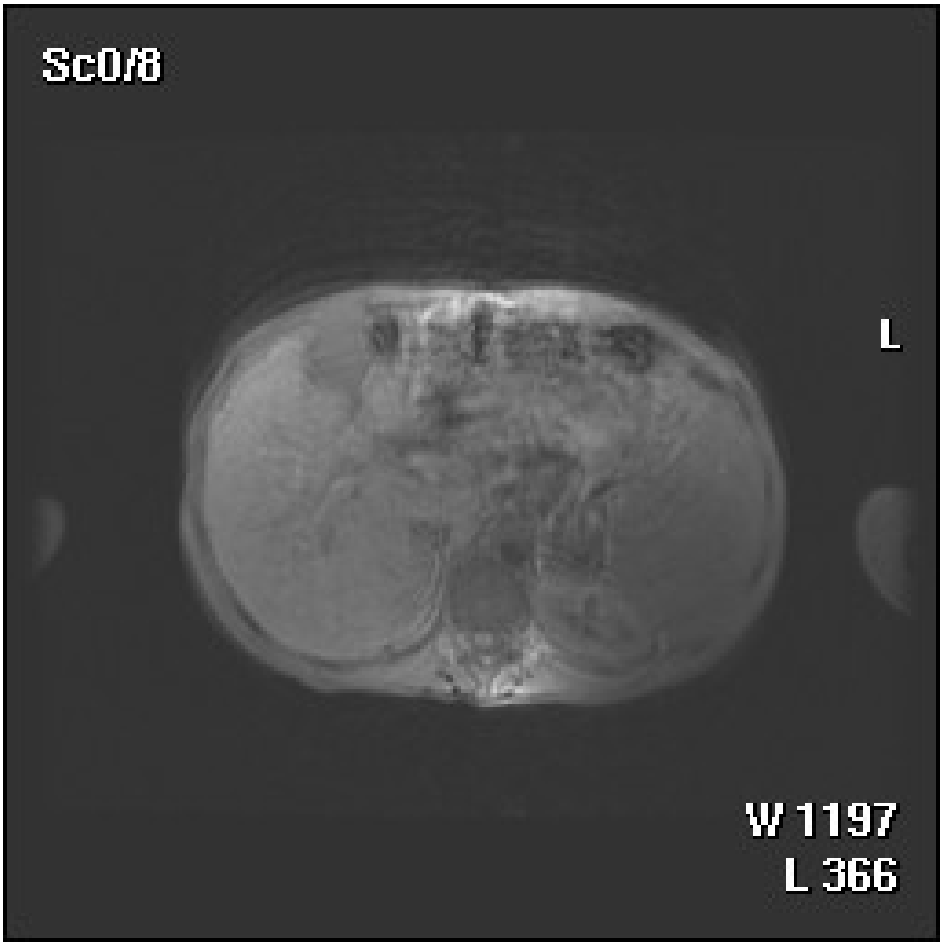

Fig.3 MRI scan.

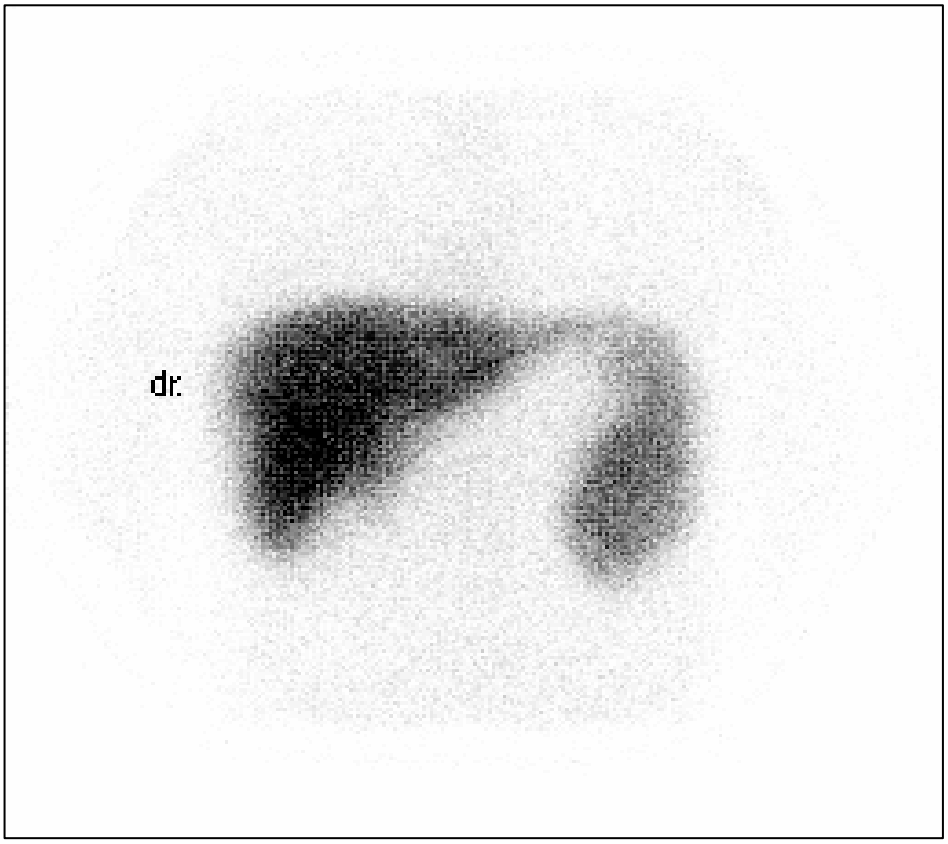

Fig.4-Hepato-biliary scintigraphy.

In evolution, the case complicated: concerning the liver disease, multilobular cirhosis ocured with portal hypertension and neuropsychiatric symptoms became more evident. Diabetes mellitus developed, with conspicuous hyperglycemia, but the adolescent refused starting the insulinotherapy. The lung function deteriorated, the obstructive syndrome accentuated and frequent exacerbations of pulmonary disease occured.

Psychological issues were accentuated by the occurence of encephalopathy, beside the insulin injections necessary for control of diabetes in to a very difficult period of his life, the adolescence.

Finally he was convinced to accept the treatment with insulin, ursodeoxocholic acid, inconstantly diet and aerosol therapy and, in pulmonary exacerbation - intravenous antibiotherapy.

In conclusion, liver disease is a relatively frequent and early complication of cystic fibrosis. The pathogenesis is apparently multifactorial, with contributions of environmental and genetic determinants.

Its impact on quality of life and survival will increase in future years, the early detection and treatment will become an important issues. Ursodeoxycholic acid is the only treatment currently available, but novel therapeutic options are being evaluated.

Beside the matters concerning the medical management of a CF case with liver disease and diabetes, the psychological issues related to age or hepatic encephalopathy raise, making more difficult the manner of patient`s life and worsening the course of disease.

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