

**184 Evaluation of the impact of SERPINA gene mutation on the occurrence of liver damage and cholestasis in patients diagnosed with cystic fibrosis**

S. Wiecek^1, H. Woł^1, B. Kordys-Darmolinska^1, U. Grzybowska-Chlebowczyk^1.

^1Medical University of Silesia, Department of Paediatrics, Katowice, Poland

SERPINA1 gene is present in about 2% of patients with cystic fibrosis, but more frequently, in approximately 5% of patients with cystic fibrosis (CF) and concomitant changes in the liver.

**Aim:** The aim of this study was to assess the impact of SERPINA gene mutation on the occurrence of liver damage and cholestasis in patients diagnosed with cystic fibrosis.

**Patients and Methods:** The analysis included 139 consecutive patients, 13 girls (43.3%) and 17 boys (56.6%), aged from 6 months to 18 years, with diagnosed CF. All patients underwent genetic testing for SERPINA gene mutation. The analysis included age, sex, clinical symptoms, type of mutation of the CFTR protein, laboratory abnormalities (levels of transaminases, GGTP, FA, protein, acid steatoctrit) and ultrasound examination of the abdomen.

**Results:** Elevated transaminases were found in 9/30 (30%), whereas elevated levels of gamma-glutamyl transferase in 6/30 (20%) children. In 13/30 patients the ultrasound examination demonstrated liver enlargement with increased echogenicity. The mutation in SERPINA gene was found in 1/30 (3.3%) patient with cystic fibrosis. Currently, this patient has normal values of transaminases, GGTP and FA, whereas, a significant worsening of respiratory symptoms is observed. There was no correlation between the occurrence of SERPINA gene mutation and clinical symptoms, type of CFTR protein mutation, results of laboratory tests of liver function and hepatocyte damage, and ultrasound examination of the abdomen.

**Conclusions:** There was no correlation between the occurrence of SERPINA gene mutation and the presence of features of liver damage and cholestasis in children diagnosed with cystic fibrosis.

---

**185 Transient elastography measurements in adults with cystic fibrosis liver disease**

G.H. Jones^1, P. Richardson^2, M. Ledson^1, M. Walsh^1, J. Greenwood^1.

^1Liverpool Heart & Chest Hospital, Adult CF Service, Liverpool, United Kingdom; ^2Royal Liverpool & Broadgreen University Hospitals, Hepatology, Liverpool, United Kingdom

**Objectives:** The optimal frequency of ultrasound scans (USS) to monitor the development of portal hypertension (PH) in CF-related liver disease (CFLD) is unknown. Transient elastography (TE), a simple non-invasive form of monitoring, is being increasingly used as an alternative to USS but there is little experience in adult CF.

**Methods:** Using a Fibroscan® 402 device, we compared the utility of TE with USS in the diagnosis of PH. TE was measured using the median value from at least 10 readings at routine outpatient review. The most recent USS was reviewed and considered abnormal if it featured heterogeneous or nodular changes or signs of portal hypertension.

**Results:** TE was compared with contemporaneous USS in 78 patients (mean age 26.8, [range 17–53], male 66.7%). The 15 with an abnormal USS had a higher TE than the remainder (median 11.7 kPa [IQR 10.0–19.6] vs 4.4 [IQR 3.7–6.3] p < 0.005). TE was highly useful in differentiating whether abnormalities were present on USS (area under receiver operating curve = 0.94), where a cut-off of 6.6 kPa excluded USS abnormality (98% negative predictive value). All patients with PH on USS had a TE of >10.8 kPa. Furthermore, USS were normal in 3 with oesophageal varices: all had raised TE.

**Conclusion:** TE can be used to identify CF patients likely to have changes of CFLD on USS, and may be a better predictor of significant disease than USS alone. Since it is quick to administer, gives immediate results and can be measured during routine outpatient review, TE may be a useful tool to guide which patients are referred on for more resource intensive investigations that require specialist staff to administer and interpret.

---

**186 Transient elastography can be used to guide monitoring of cystic fibrosis related liver disease in adult patients**

G.H. Jones^1, P. Richardson^2, M. Ledson^1, M. Walsh^1, J. Greenwood^1.

^1Liverpool Heart & Chest Hospital, Adult CF Service, Liverpool, United Kingdom; ^2Royal Liverpool & Broadgreen University Hospitals, Hepatology, Liverpool, United Kingdom

**Objectives:** Evaluation of cystic fibrosis liver disease and the relation with risk factors in a Romanian centre

L. Cincu^1, L. Pop^1, Z. Popa^2, L. Tamas^1, B. Almajan Guta^2, P. Matusz^2.

^1University of Medicine and Pharmacy Victor Babes, Pediatric II Department, Timisoara, Romania; ^2University of Medicine and Pharmacy Victor Babes, Department of Anatomy and Embriology, Timisoara, Romania

**Introduction:** Liver disease is the second non-pulmonary cause of death in cystic fibrosis, with increasing life expectancy, became an important management problem. Predisposing factors like male sex, pancreatic insufficiency, meconium ileus and severe mutation are incriminated to influence the occurrence of cystic fibrosis associated liver disease (CFLD). Transient elastography is being increasingly used as an alternative to USS but there is little experience in adult CF.

**Methods:** Using a Fibroscan® 402 device, we compared the utility of TE with USS in the diagnosis of PH. TE was measured using the median value from at least 10 readings at routine outpatient review. The most recent USS was reviewed and considered abnormal if it featured heterogeneous or nodular changes or signs of portal hypertension.

**Results:** TE was compared with contemporaneous USS in 78 patients (mean age 26.8, [range 17–53], male 66.7%). The 15 with an abnormal USS had a higher TE than the remainder (median 11.7 kPa [IQR 10.0–19.6] vs 4.4 [IQR 3.7–6.3] p < 0.005). TE was highly useful in differentiating whether abnormalities were present on USS (area under receiver operating curve = 0.94), where a cut-off of 6.6 kPa excluded USS abnormality (98% negative predictive value). All patients with PH on USS had a TE of >10.8 kPa. Furthermore, USS were normal in 3 with oesophageal varices: all had raised TE.

**Conclusion:** TE can be used to identify CF patients likely to have changes of CFLD on USS, and may be a better predictor of significant disease than USS alone. Since it is quick to administer, gives immediate results and can be measured during routine outpatient review, TE may be a useful tool to guide which patients are referred on for more resource intensive investigations that require specialist staff to administer and interpret.

---

**187 Evaluation of cystic fibrosis liver disease and the relation with risk factors in a Romanian centre**

L. Cincu^1, L. Pop^1, Z. Popa^2, L. Tamas^1, B. Almajan Guta^2, P. Matusz^2.

^1University of Medicine and Pharmacy Victor Babes, Pediatric II Department, Timisoara, Romania; ^2University of Medicine and Pharmacy Victor Babes, Department of Anatomy and Embriology, Timisoara, Romania

**Introduction:** Liver disease is the second non-pulmonary cause of death in cystic fibrosis, with increasing life expectancy, became an important management problem. Predisposing factors like male sex, pancreatic insufficiency, meconium ileus and severe mutation are incriminated to influence the occurrence of cystic fibrosis associated liver disease (CFLD). Transient elastography is being increasingly used as an alternative to USS but there is little experience in adult CF.

**Methods:** Using a Fibroscan® 402 device, we compared the utility of TE with USS in the diagnosis of PH. TE was measured using the median value from at least 10 readings at routine outpatient review. The most recent USS was reviewed and considered abnormal if it featured heterogeneous or nodular changes or signs of portal hypertension.

**Results:** TE was compared with contemporaneous USS in 78 patients (mean age 26.8, [range 17–53], male 66.7%). The 15 with an abnormal USS had a higher TE than the remainder (median 11.7 kPa [IQR 10.0–19.6] vs 4.4 [IQR 3.7–6.3] p < 0.005). TE was highly useful in differentiating whether abnormalities were present on USS (area under receiver operating curve = 0.94), where a cut-off of 6.6 kPa excluded USS abnormality (98% negative predictive value). All patients with PH on USS had a TE of >10.8 kPa. Furthermore, USS were normal in 3 with oesophageal varices: all had raised TE.

**Conclusion:** TE can be used to identify CF patients likely to have changes of CFLD on USS, and may be a better predictor of significant disease than USS alone. Since it is quick to administer, gives immediate results and can be measured during routine outpatient review, TE may be a useful tool to guide which patients are referred on for more resource intensive investigations that require specialist staff to administer and interpret.