304 Celiac disease in patients with cystic fibrosis: a common association?

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Celiac Disease (CD, gluten induced enteropathy) is a relatively common severe chronic gastrointestinal disease. believed to be present in up to 1% of Caucasians in our population. The clinical features of CD include steatorrhoea, malnutrition, failure to thrive which are also common features of Cystic Fibrosis (CF). The coincidence of CF and CD have been reported previously, and one study based on clinical and biochemical but not serological evaluation, followed by small bowel biopsy (SBB) found CD confirmed in 5 of 1100 CF patients. The advent of serological screening tests, particularly tissue trans-glutaminase (tTG) has led to increased ascertainment of CD, though the diagnosis should still be confirmed SBB. We report results from the first survey of CF patients using modern serological testing (tTG) and SBB. We screened 114 CF patients aged 1–18 yrs. attending the CF clinic at BC Children's Hospital in Vancouver. 7 patients had elevated tTG. Of these, 5 were also +ve for HLADQ2 or DQ8. SBB in 4 was positive for CD, while 3 patients await SBB. A serological prevalence of 7% and confirmed SBB diagnosis of CD in 4% is greater than expected for the general population. Our results should be confirmed by other studies, but suggest that routine serological tTG screening for CD is indicated in all CF patients.

305 Outcomes of a regional paediatric CF gastroenterology clinic

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Introduction: In CF, liver disease is the third highest cause of death and gastrointestinal problems impact on quality of life. A designated CF Gastroenterology clinic (CFGC) was established in 2001 to facilitate diagnosis and management. Families are reviewed by a Gastroenterologist, a CF Dietitian and a CF Physician. We have assessed the impact of this clinic on patient care.
Method: Retrospective case note review of patients attending the CFGC 2001–2009.
Results: Forty three patients attended the clinic, 38/43 (20 male, 18 female) were reviewed, 5 patient's case notes were unavailable. The mean age at presentation was 9.5 years (range 1–17). Reasons for referral: poor weight gain (13), abnormal liver function tests (LFT's) (10), abdominal pain (9), loose stools (5), vomiting (5). Coeliac disease was excluded by serology in five patients and by biopsy in nine. All patients with abnormal LFT's were treated with ursodeoxycholic acid, 6 had portal hypertension and 3 were referred to UK liver centres. Eight patients had clinical gastro-oesophageal reflux, 7/8 had endoscopy and/or contrast swallow, all normal. Nine children had a percutaneous endoscopic gastrostomy tube for nutritional supplementation.
Outcome: 31 patients were discharged to their usual CF care, 22/31 symptoms improved, 9/31 diagnosed and given a plan of care. Two patients transferred to adult services, 4 continue to attend the CFGC and 1 declined follow-up.
Conclusion: Attendance at the CFGC has resulted in a positive intervention in the majority of patients. The clinic has been particularly helpful in supporting families with the move to interventional feeding.

306 The analysis of phenotype of CF patients HFE mutations carriers

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The variable clinical manifestations of CF suggest the influence of modifier genes. Genetic and environmental factors that determine whether an individual will develop associated complications are still being determined. Mutation analysis is widely recommended for presymptomatic diagnosis of CF and HH (hereditary hemochromatosis) the most frequent autosomal recessive diseases. Five C282Y heterozygous carriers, fourteen H63D heterozygous carriers and one compound heterozygous C282Y/H63D were identified out of 62 CF patients. We have analyzed the phenotype of 20 (9 female, 11 male) CF patients HFE mutations carriers aged from 2 to 25 years old. As the results showed the high frequency of HFE mutations among patients with CF we have analyzed the frequency of C282Y and H63D mutations in CF patients with different severity of CF manifestation. The obtained results revealed no correlation between HFE gene mutations and severity of CF manifestation, meconium ileus occurrence, gender and development of hepatobiliary disturbances. HFE C282Y/H63D compound heterozygous patient, seven years old boy, F508del homozygous had meconium ileus at the birth. The further studies of a larger group of patients and monitoring them for long time will clarify the HFE mutations influence on CF phenotype.

307 Clinical and genetic characteristics of cystic fibrosis patients with liver cirrhosis

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The aim of study was to analyze the clinical and genetic characteristics of patients with liver cirrhosis associated with cystic fibrosis (CF). Liver cirrhosis was defined by ultrasonographic findings of distinct heterogeneity of liver parenchyma and nodular liver surface and/or by liver biopsy findings. Portal hypertension was suggested by enlarged spleen and abnormal portal venous flow.
Ten patients with CF (nine males and one female) were found to have liver cirrhosis, three of them with portal hypertension. Hepatic synthetic function was sufficiently maintained in all of cirrhotic patients. Average age of patients with cirrhosis was 15.5 years (range 8–23). All patients had pancreatic insufficiency. Nutritional status expressed as standard deviation score (Z) for weight (zW), for height (zH), for weight for height (zW/H), and body mass index (BMI) showed these values: $zW = -0.73 \pm 0.98$, $zH = -0.65 \pm 0.45$, $zW/H = -0.05 \pm 0.59$, and $BMI = 18.86 \pm 2.36$. CF patients with liver cirrhosis tended to have milder pulmonary disease, with mean FVC and FEV_1 values of 96.5 ± 9.6 and 92 ± 14, respectively. Five of them were with chronic *Pseudomonas aeruginosa* infection. Genetic analysis showed higher frequency of F508del mutation in the group with cirrhosis (80%). The other mutations found among these patients were: G542X, Y1092X and R1662.
In conclusion, liver cirrhosis associated with CF usually develops during the first decade. Patients with mail gender, pancreatic insufficient and with severe CFTR mutations are exposed to higher risk for developing liver cirrhosis. Liver cirrhosis does not significantly impact the pulmonary function and the nutritional status, until the end-stage liver disease.