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

A.G.E. Davidson1, A. Martinez2, V. McMahon3, T. Gonzalez4, S. Jenkins3, Y.P. Lillquist1, C. Barker4. 1University of British Columbia/BC Children's Hospital, Cystic Fibrosis Clinic, Vancouver, BC, Canada; 2BC Children's Hospital, Department of Pediatrics, Division of Gastroenterology, Vancouver, BC, Canada; 3BC Children's Hospital, Cystic Fibrosis Clinic, Vancouver, BC, Canada; 4BC Children's Hospital, Division of Gastroenterology, Vancouver, BC, Canada

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 HLA-DQ2 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.

Outcomes of a regional paediatric CF gastroenterology clinic

P. Senthamilarasu1, L.J. Heaf2, R.M. Watling3, M.A. Dalzell1, K.W. Southern4. 1Alder Hey Children's Hospital NHS Foundation Trust, Gastroenterology, Liverpool, United Kingdom; 2Alder Hey Children's Hospital NHS Foundation Trust, Respiratory Unit, Liverpool, United Kingdom; 3Alder Hey Children’s Hospital NHS Foundation Trust, Dietetics, Liverpool, United Kingdom; 4University of Liverpool, Institute of Child Health, Liverpool, United Kingdom

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.


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.

The analysis of phenotype of CF patients HFE mutations carriers

L. Bobes1, O. Bisevich2, H. Akopyan2, N. Rohonyy1, H. Maksik2. 1Lviv Regional Specialized Children’s Hospital, Lviv, Ukraine; 2Institute of Hereditary Pathology of Academy of Medical Science of Ukraine, Lviv, Ukraine

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.