

Of these 32 patients 29 had positive tTg. This gives a sensitivity of 90%. Thirteen patients had tTg greater than 100, 12 had positive intestinal biopsy; this gives a sensitivity of 92%.

**Conclusion** Although the sensitivity of tTg in our series is 90%, it is reasonable to assess small intestinal biopsy in before subjecting children to lifelong gluten free diet.

#### 724 HLA DQ2/DQ8 TYPING IN CHILDREN DIAGNOSED WITH CELIAC DISEASE

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**Background and Aims** Genes encoding HLA DQ2/DQ8 are associated with celiac disease (CD) and testing for their presence has high negative predictive value for the diagnosis. The aim of this study was to assess the role of HLA typing in symptomatic individuals in whom the diagnosis of CD is uncertain.

**Methods** We proceeded a retrospectiv study leaded on a group of children investigated for CD in "Grigore Alexandrescu" Emergency Children's Hospital from 2007 to 2012 that underwent HLA typing. Inclusion criteria were all patient with mild enteropathy (Marsh 1, 2, 3a), moderate elevated values of tular transglutaminase (tTG) antibodies (between cut off point and 5 times normal value) and poor response to gluten free diet. The medical records of all patients investigated for CD were reviewed.

**Results** 164 patients were performed HLA typing; 26 patients satisfied the inclusion criteria; 20 (76.9%) of these had HLA DQ2/DQ8 present and 6 (23.07%) had a negative test for HLA DQ2/DQ8. The mean age of our investigated group was 23.46 months and the mean age for HLA DQ2/DQ8 negative group was 21.08 months. Sex distribution indicated 9 boys and 17 girls. Gastrointestinal symptoms dominated: 17 children had diarrhea, 9 had failure to thrive and 13 patients had both chronic diarrhea and poor weight gain.

**Conclusion** Patients with clinical suspicion of CD that have moderate levels of tTG antibodies, mild biopsy changes and poor response to gluten free diet need to have HLA typing specially at younger ages (under 3 years old).

#### 725 LYMPHOCYTE RESPIRATION IN CHILDREN WITH TRISOMY 21

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**Aims** This study aimed to measure lymphocyte mitochondrial O<sub>2</sub> consumption (cellular respiration) in children with trisomy 21.

**Methods** Peripheral blood mononuclear cells were isolated from whole blood of trisomy 21 and control children and immediately used to measure the respiratory rate. [O<sub>2</sub>] was determined as function of time from the phosphorescence decay rates (1/t) of Pd (II)-meso-tetra-(4-sulfonatophenyl)-tetrabenzoporphyrin. In sealed vials containing cells and glucose as a respiratory substrate, [O<sub>2</sub>] declined linearly with time, confirming the zero-order kinetics of O<sub>2</sub> consumption (conversion to H<sub>2</sub>O) by cytochrome oxidase.

**Results** The rate of respiration (k, in mM O<sub>2</sub> per min), thus, was the negative of the slope of [O<sub>2</sub>] vs. time. NaCN inhibited O<sub>2</sub> consumption, confirming the oxidation occurred in the mitochondrial respiratory chain. For control children (age = 8.8±5.6 yr, n=26), the mean (± SD) value of k<sub>c</sub> (in mM O<sub>2</sub> per min per 10<sup>7</sup> cells) was 1.36±0.79 (coefficient of variation = 58%; median = 1.17; range = 0.60 to 3.12; -2SD = 0.61). For children with trisomy 21 (age = 7.2±4.6 yr, n=26), the value of k<sub>c</sub> was 0.82±0.62 (coefficient of variation = 76%; median = 0.60; range = 0.20 to 2.80), p<0.001. Fourteen of 26 (54%) children with trisomy 21 had k<sub>c</sub> values of 0.20 to 0.60 (i.e., < -2SD).

**Conclusion** Thus, it appears that some children with trisomy 21 have relatively reduced lymphocyte bioenergetics. The biological implication of this finding (variation) requires further studies.

#### 726 LERI-WEILL DYSCHONDROSTEOSIS - A CASE OF COMPLETE DELETION OF ONE OF THE COPIES OF SHOX GENE

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**Introduction** Leri-Weill dyschondrosteosis (LWD), is a dominantly inherited skeletal dysplasia with disproportionate short stature owing to mesomelic shortening of the forearm and lower leg and Madelung deformity of the arm is found in 74% of children. SHOX mutations is found in 70% of cases.

**Case Report** Thirteen old month boy was admitted to genetic consultation because of short stature. The mother has disproportionate short stature. On physical examination, we found a phenotype similar with the mother, with short arms and lower legs. Height below the 5 th percentile. The skeletal x-ray confirmed mesomelic shortening of the forearm and lower legs. The x.ray did not demonstrated Madelung deformity of the arm. Molecular study using MLPA, confirmed complete deletion of one of the copies of SHOX gene - more than 440 Kb. Later on, we confirmed that he has growth hormone deficiency. The mother has also LWD.

**Discussion** LWD should be included in the diagnoses of short stature, especially if the child is disproportionate and has family history. In our case, because the mother is affected, the deletion of the SHOX gene is inherited in the pseudoautosomal region of X chromosome. The transmission is pseudodominant and so the daughters of the index case will inherited the X chromosome of the father and will be affected. The boys will inherited the Y chromosome of the father. Prenatal diagnosis and genetic counseling is available for this syndrome. Treatment options include administration of recombinant growth hormone to improve final adult height.

#### 727 WILSON'S DISEASE: A CHALLENGING DIAGNOSIS. CLINICAL MANIFESTATIONS AND DIAGNOSTIC PROCEDURES IN 32 PATIENTS

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**Introduction** Wilson disease is a neurodegenerative disease of copper metabolism. The genetic defect, localized to chromosome arm 13q, has been shown to affect the copper-transporting adenosine triphosphatase (ATPase) gene (*ATP7B*) in the liver.

**Material and Methods** Our aim was to study the clinical and laboratory characteristics of 32 children and young adults diagnosed with WD and point out the diagnostic difficulties. The study was done between 2001 and 2011. Evaluation included detailed physical examination, conventional laboratory testing, genetic analysis, and liver biopsy.

**Results** Patients with hepatic symptoms showed a considerably earlier onset of symptoms and a shorter diagnostic delay before definitive diagnosis than those with neuropsychiatric symptoms. The mean age at diagnosis was 9.12 +/- 2.59 years (range 5 years-20 years). 30 patients were symptomatic, 18 were referred because of abnormal liver function test results and/or hepatomegaly, 12 had neuropsychiatric symptoms and 2 received their diagnoses after family screening. Hepatic copper concentration was between 250 and 1200 microg/g. 12 patients had liver cirrhosis, 16 chronic hepatitis, and 2 had massive hepatic necrosis on necropsy.

**Conclusions** Any person with recurrent hepatic disease and unexplained neurologic symptoms should be investigated to have