

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

doi:10.1136/archdischild-2012-302724.0724

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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

doi:10.1136/archdischild-2012-302724.0725

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Aims This study aimed to measure lymphocyte mitochondrial O₂ 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. [O₂] 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, [O₂] declined linearly with time, confirming the zero-order kinetics of O₂ consumption (conversion to H₂O) by cytochrome oxidase.

Results The rate of respiration (k, in mM O₂ per min), thus, was the negative of the slope of [O₂] vs. time. NaCN inhibited O₂ 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_c (in mM O₂ per min per 10⁷ 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_c 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_c 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

doi:10.1136/archdischild-2012-302724.0726

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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 SCHOX 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

doi:10.1136/archdischild-2012-302724.0727

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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

Wilson's disease. Detection of WD in children and young adults remains very difficult. The most important investigation is liver biopsy with the assessment of liver copper. Genetic analysis may help in doubtful cases.

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DNA DAMAGE AND APOPTOSIS EVALUATION IN LEUKOCYTES OF ICTERIC NEWBORNS AFTER PHOTOTHERAPY TREATMENT BY USING OF THE NEUTRAL COMET ASSAY

doi:10.1136/archdischild-2012-302724.0728

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Background and Aims Phototherapy is the most common form of treatment for jaundice. one of the various endpoints that can be used to investigate the potential of blood as a predictor of radiosensitivity is DNA damage and apoptosis. The aim of this study was to investigate the possible relation between phototherapy and DNA damage and apoptosis, in neonates with hyperbilirubinemia.

Methods The study included 15 healthy full-term newborns as control. The phototherapy group consisted of 30 non-physiologic jaundice that after 16 hours phototherapy, 1 ml peripheral venous blood were obtained of them.

Results DNA damage immediately after phototherapy termination (IPT) was higher in jaundice infants than control one ($p < 0.01$). After 24 hours cells repaired their damages, as there wasn't any difference between these groups about this value. DNA damage in the phototherapy group was higher at IPT than 24 hours after that, but in the control one there weren't any differences between them (respectively $p < 0.001$, $p > 0.05$). Apoptosis value at 24 hours after phototherapy termination was higher than the IPT in the phototherapy group and was statistically significant ($P < 0.001$).

Conclusions Since repair proofreading and fidelity properties isn't absolutely, then always this is probable that errors maybe occur during extensive DNA damages repair and finally these errors can cause mutation in DNA. If this event be in important and sensitive region of genome, harmful effects would menace phototherapy-treated infants' later-life. So this is necessary to investigation of long-term genotoxic effects of phototherapy in phototherapy-treated neonates.

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INTERLEUKIN-13 RECEPTOR α_1 GENE POLYMORPHISM AND IL-13 BLOOD LEVEL IN ATOPIC AND NON ATOPIC ASTHMATIC AND ALLERGIC RHINITIS CHILDREN

doi:10.1136/archdischild-2012-302724.0729

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Objectives To assess the value of serum interleukin (IL) 13 levels as an immunological marker in atopic upper respiratory diseases, to clarify its differences in atopic and non atopic bronchial asthma and to determine the role of an IL-13 R α_1 gene single nucleotide polymorphism (SNP) (A1398G) in the pathogenesis of these diseases.

Methods Seventy-five patients were compared with 25 age-matched healthy volunteers. Serum total immunoglobulin (Ig) E and IL-13 levels were measured by enzyme-linked immunosorbent assay and the IL-13 R α_1 gene (A1398 G) was screened by specific polymerase chain reaction.

Results There was a non significant association between G allele frequencies of the IL-13 R α_1 (1398) gene polymorphism as compared to in controls. There were a significant increase in the serum level of total IgE & IL-13 towards heterozygous AG and

homozygous GG than homozygous AA in atopic asthma, non atopic asthma, and allergic rhinitis patients. There was a significant increase in the serum level of total IgE & IL-13 towards homozygous GG than heterozygous AG in atopic asthma non atopic asthma, and allergic rhinitis patients for IgE and in all groups for IL-13.

Conclusion interleukin-13 receptor α_1 subunit gene A1398G polymorphism does not contribute to asthma or allergic rhinitis susceptibility, although the interleukin-13 receptor α_1 subunit gene locus might be involved in the control of immunoglobulin E production, IL13 can used as an immunological marker in atopic upper respiratory diseases and to differentiate between atopic and non atopic bronchial asthma.

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BRAIN GENOMIC RESPONSE AFTER PERINATAL ASPHYXIA AND FETAL ASPHYCTIC PRECONDITIONING

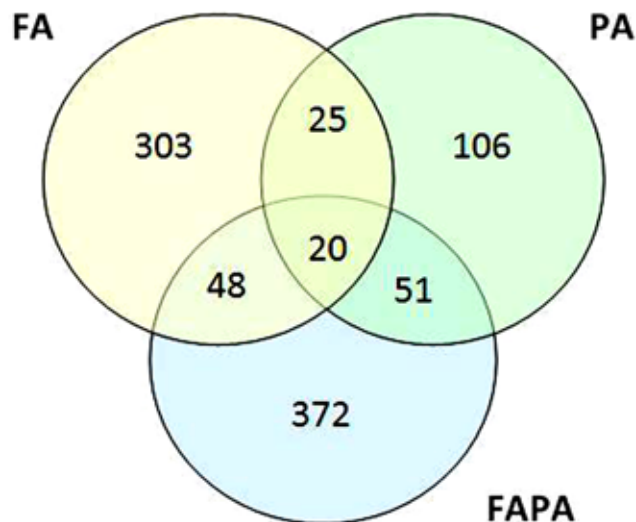
doi:10.1136/archdischild-2012-302724.0730

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Background and Aims Fetal asphyctic (FA) preconditioning has been shown to be effective in attenuating brain damage incurred by a subsequent perinatal asphyctic event. Unraveling mechanisms of this endogenous neuroprotection, is an important step towards new clinical strategies for asphyctic neonates. Genomic reprogramming is, at least in part, responsible for the protective effect of preconditioning in the brain. Therefore we investigated differential gene expression with whole genome micro-array.

Methods FA preconditioning was induced on E17 by reversibly clamping the uterine circulation for 30 min. Perinatal asphyxia (PA) was induced at E21/22 by submersing the uterine horns including pups in a water bath for 19 minutes. Four experimental groups were sacrificed 6 and 96h after birth: Control, FA, PA, and preconditioned animals that underwent perinatal asphyxia (FAPA). Whole genome transcription was investigated with the Affymetrix Gene1.0ST chip and analyzed with the Bioconductor Limma package.

Results Figure 1 and 2 depict the number of differentially regulated transcripts respectively 6 and 96 hours after birth.



Abstract 730 Figure 1 Number of differentially expressed transcripts compared to control ($p < 0.01$), 6h after birth