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.

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

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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.

729 INTERLEUKIN-13 RECEPTOR A_1 GENE POLYMORPHISM AND IL-13 BLOOD LEVEL IN ATOPIC AND NON ATOPIC ASTHMATIC AND ALLERGIC RHINITIS CHILDREN

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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.

730 BRAIN GENOMIC RESPONSE AFTER PERINATAL ASPHYXIA AND FETAL ASPHYCTIC PRECONDITIONING

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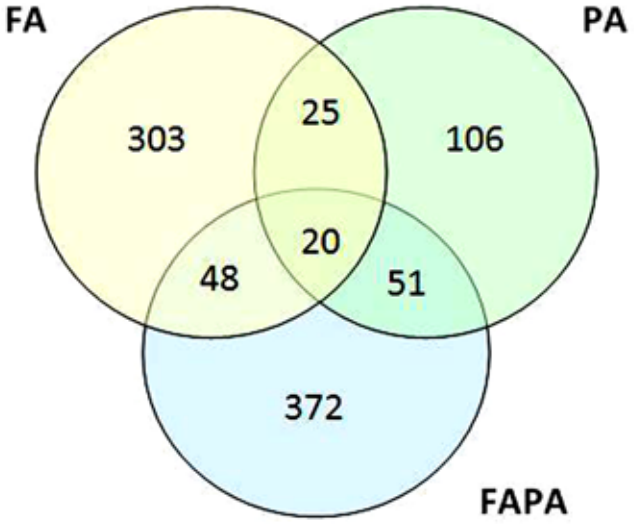
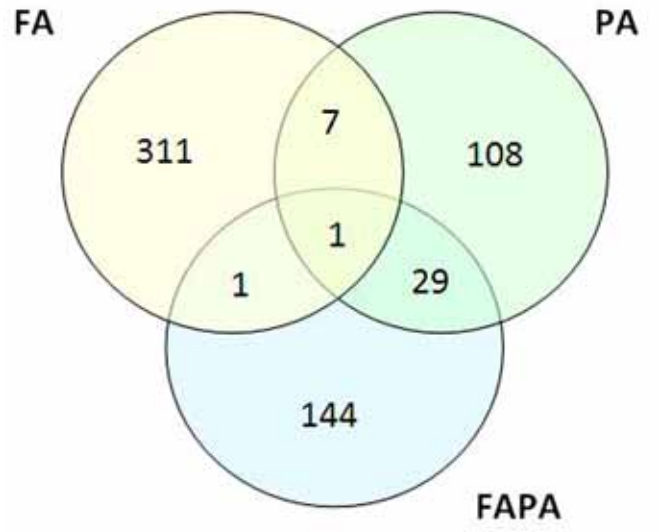
Background and Aims Fetal asphyctic (FA) preconditioning has been shown to be effective in attenuating brain damage incurred by a subsequent perinatal asphyctic event. Unraveling mechanisms of this endogenous neuroprotection, is an important step towards new clinical strategies for asphyctic neonates. 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



Abstract 730 Figure 2 Number of differentially expressed transcripts compared to control ($p<0.01$), 96h after birth

Many of these transcripts are involved in synaptic transmission and metabolism. Interestingly, we also found changes in several histone clusters and histone deacetylases.

Conclusions This is the first study to investigate whole genome expression in a preconditioning and ashyxia model that includes the fetal-to-neonatal transition and therefore truly resembles perinatal asphyxia. Our results warrant further research into epigenetic mechanisms of neuroprotection.

731 PRADER WILLI SYNDROME (PWS) - PARTICULAR MOLECULAR PROFILE AND DIAGNOSTIC PROTOCOL IN ROMANIA

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Background and Aims PWS-a rare genetic disease with a 1/12000–1/15000 newborns frequency, caused by deletion of some genes on paternal origin or maternal disomy of 15'th chromosome. Main clinical manifestations are: neonatal hypotonia, excessive childhood feeding, obesity, cognitive deficiencies. Our aims are to implement molecular genetic/epigenetic tests and to develop interdisciplinary clinical research protocol, which allow early diagnostic and intervention in Romania.

Materials and Methods This study is part of a multicenter research project (CNMP/Partnerships, 2008–2011), on 19 Romanian PWS patients, 12 females, 7 males, between 6 months and 29 years. For diagnostic, were used major and minor criteria (Gunay-Aygun) as clinical methods and 5 genetic tests.

Results All patients have a clinical diagnostic score above 5, 63% of them having a maximal major criteria number with 100% neonatal hypotonia, 95% feeding difficulties at infants and hyperphagia after and a BMI till 60.2kg/cm^2. 15% of patients have all minor criteria positive, with lethargy at infants, viscous saliva and small extremities predominance. 5% of patients have a positive 15q11–q13 microdeletion, 79% a FISH positive and for 47% patients MS-PCR is positive. Techniques like MS-MLPA were late introduced in Romania, 20% of our patients having them.

Conclusions The study indicates a relative correlation between clinical score and cytogenetic/molecular PWS confirmation and emphasizes the importance of early diagnostic. Interdisciplinary clinical criteria, karyotype, FISH and methylation analysis (MS-PCR, MS-MLPA) are the main steps for a successful diagnostic protocol. Genetic tests results show a particular molecular profile in Romania with only 47% positive methylation results unlike literature (99%).

732 TURNER SYNDROME: A CLINICO-CYTOGENETIC STUDY OF 37 CHILDREN

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Turner syndrome (TS) is defined by total or partial loss of the sex chromosomes X. Features vary widely including short stature and ovarian failure inconstantly associated with characteristic face, skeletal malformations, renal and cardiac anomalies and endocrine disorders.

We analyzed the clinical and cytogenetic profiles of 37 TS children diagnosed with TS from January 2007 to December 2011 in the aim to establish genotype- phenotype correlations.

Growth delay and hypothyroidism were noted respectively in 89.2% and 19.4%of patients. Diabetes and celiac disease was observed in 5.6% of cases. 35% of our cohort had a 45, X karyotype, 8.1% had 45, X/47, XXX mosaïcism and 5.4% have 45, X/46, XY mosaïcism Interestingly, FISH revealed the presence of *SRY* gene. The remainders had structural abnormalities: 35.1% had isochromosome Xq which was homogenous in roughly half of cases. 10.8% were diagnosed with a terminal deletion Xp and 5.4% with a ring of chromosome X.

There was no correlation between genotypes and clinical features. The short stature in girls with TS is thought to be related to the haploinsufficiency of the *SHOX* gene on Xp22.3. As a result, treatment with GH is now routinely adopted even if the GH hormone is normally secreted. The higher risk of autoimmune diseases in women with TS could result from haploinsufficiency of the *FOXP3* gene on Xp 11.23.

Otherwise, we highlight the importance of detection of 45, X/46, Xy mosaïcism which may be cryptic requiring SRY probe FISH screening a condition that exposes to gonadoblastome and special chirurgical preventive treatment.

733 BACTERIAL 16S $_R$ RNA GENETIC MARKERS FOR FECAL SAMPLES TO DIFFERENTIATE CHOLEDOCHAL CYST FROM BILIARY ATRESIA

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Background and Aims Microbiota in fecal content from choledochal cyst (CC) and biliary atresia (BA) individuals at the operation were compared using 16S rRNA gene libraries and terminal restriction fragment length polymorphism (T-RFLP).

Methods From 2002 to 2011, 1 infant with CC and 7 infant with BA (infants ≤ 2 months of age) were treated at our institute. Fecal samples were obtained at the radical operation for CC and BA. Total fecal DNA was isolated and PCR was performed. The amplification of the fecal 16S rDNA, restriction enzyme (BslI), size-fractionation of T-RFs and T-RFLP data analysis were performed. To compare the T-RFLP patterns among samples between CC and BA patients, the

730 Brain Genomic Response after Perinatal Asphyxia and Fetal Asphyctic Preconditioning

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Notes