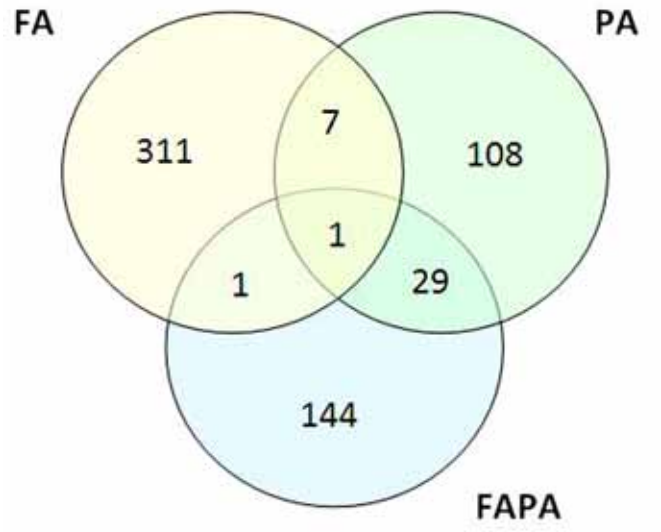

Abstract 730 Figure 2 Number of differentially expressed transcripts compared to control ($p<0.01$), 96h after birth

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