and intervention in Romania.

Disciplinary clinical research protocol, which allow early diagnostic implementation molecular genetic/epigenetic tests and to develop inter-

cognitive deficiencies. Our aims are to childhood feeding, obesity, cognitive deficiencies. Our aims are to research project (CNMP/P 1IE Jurca- Simina, 1M Gafencu, 2D Dan, 3M Puiu.

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 his-
tone clusters and histone deacetylases.

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

TURNER SYNDROME: A CLINICO-CYTOGENETIC STUDY OF 37 CHILDREN

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M Kammoun, ‘S Mougu, ‘R Brahem, ‘N Ghali, ‘I Bel Haj Hmida, ‘S Dimassi, ‘N Soyah, ‘I E Gzel, ‘A Saad. 1Departments of Cytogenetics and Reproductive Biology, Farhat Hached University Teaching Hospital; 2Departments of Endocrinology, Farhat Hached University Teaching Hospital; 3Department of Nephrology, Sahou University Teaching Hospital, 4Departments of Pediatrics, Farhat Hached University Teaching Hospital, Sousse, Tunisia

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, skele-
tal malformations, renal and cardiac anomalies and endocrine disorders.

We analyzed the clinical and cytogenetic profiles of 37 TS chil-
dren 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 mosaicism and 5.4% have 45, X/46, XY mosaicism Interestingly, FISH revealed the presence of SRY gene. The remaineds had structural abnormalities: 55.1% had isochro-

me 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 fea-
tures. 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 hor-
mone 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 mosaicism which may be cryptic requiring SRY probe FISH screening a condition that exposes to gonadoblastome and special chirurgical preventive treatment.

BACTERIAL 16S rRNA GENETIC MARKERS FOR FECAL SAMPLES TO DIFFERENTIATE CHOLEDOCHAL CYST FROM BILIARY ATRESIA

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T Okada, S Honda, H Miyagi, A Taketomi. Department of Gastroenterological Surgery I, Hokkaido University Graduate School of Medicine, Sapporo, Japan

Background and Aims Microbiota in fecal content from choledochal cyst (CC) and biliary atresia (BA) individuals at the opera-
tion 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 (BstI), 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

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 pro-
tocol. Genetic tests results show a particular molecular profile in Romania with only 47% positive methylation results unlike literature (99%).