Results Participants were 10% affluent, 65% from middle-class social groups and 25% of relatively deprived minority status. Two – thirds of parents regard themselves as overprotective and highly anxious, 32% present responsible and caring and 1% tend to be indifferent. 19% of parents initially refused admission. The major pre-admission emotions are self- blame, anger and fear, positively associated to previous hospital stays. Stress and discomfort dominate through the in-hospital phase, during which 23% of parents experienced psychosomatic symptoms. Negative emotions become reversed when the need for information by doctors and nurses is satisfied. Fathers expressed less effectively both positive and negative emotions.
Limitations Parents of children with chronic diseases were excluded from the study.
Conclusions Children's sickness and hospitalisation disorganizes the family, resulting in parental emotional disturbance, confusion and uncertainty. Stress-related emotions are better coped with, when parents feel they are respectfully treated and adequately fed-back by the hospital personnel.

PO-0381 KARTAGENER SYNDROME ASSOCIATED TO NEPHROPATHY: A BROADER SPECTRUM OF CILIOPATHY?

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Introduction Kartagener Syndrome (KS), an autosomal recessive disorder, subgroup of primary ciliary dyskinesia, is characterised by situs inversus, bronchiectasis and chronic sinusitis.
Objective To report a case of KS and associated nephropathy, diagnosed at 6 years in Hospital Alcides Carneiro (HEAC).
Method Descriptive and observational evolution of MESF, hospitalised in the paediatric ward of the HEAC, with abdominal pain and nephritic proteinuria. Clinical diagnosis of SK was established only at 6 years, despite dextrocardia diagnosed at 3 months and several subsequent hospitalizations due to respiratory infections. Nephritis was identified at 4 years, slowly progressive and without specific etiological cause determinated. Tomography showed bronchiectasis and *situs inversus totallis*. No identifiable chromosomal abnormalities seen in conventional karyotyping and no molecular study could be performed. Clinical diagnosis, however, made possible hearing loss and lung infections prophylaxis and genetic counselling. Although there are some reports of genitourinary abnormalities associated with SK, it's important to determine if KS is the specific cause of nephropathy, due the intimate relationship of other ciliopathys with renal disease, as observed in Bardet-Biedl syndrome and Meckel syndrome. In this patient the aetiology of nephropathy is unclear so far, and most common causes have been discarded.
Conclusion Factors such as low incidence estimated at 1:17000 births and pathophysiological complexity are considered contributory to late diagnosis, worsening of pulmonary disease and consequently worse prognosis. We couldn't perform molecular diagnosis in the several genes related to SK, which could provide preimplantation diagnosis and establish the best genotype-phenotype, especially in correlation with nephropathy.

PO-0382 DEVELOPMENTAL DELAY IN TRISOMY 21 CHILDREN FROM ROMANIA

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Aims Trisomy 21 is the most frequent autosomal chromosomopathy. The developmental delay is a constant finding. This study aimed to evaluate the auxological parameters of Romanian trisomy 21 patients and to correlate them with the cytogenetic findings.
Material and method We conducted an observational study on 136 patients with cytogenetically confirmed trisomy 21 that were evaluated recording their auxological parameters. The nutritional status of the patients was evaluated using growth charts from WHO until 5 years and from CDC over 5 years. For infants, the ponderal index was used. For patients ≥1 year, we used growth charts for weight and BMI and standard deviation score for height. The growth parameters were statistically evaluated considering three age groups (newborn, infant, child ≥1 year) and the cytogenetic anomalies.
Results 91.2% of patients had regular trisomy 21. The mean birth weight was 2904grams and the mean birth length 49,6cm. 80% of the infants associated protein-caloric malnutrition; the length was affected in 54.9% of 1 month - 1 year patients. 49.4% of patients ≥1 year were underweight, 37.03% were overweight and 75.3% associated short-stature. Only 14.5% of patients <1 year and 8.6% of patients ≥1 year had a normal nutritional status. No significant differences were found in relation to the cytogenetic diagnosis.
Conclusions The somatic delay varied in relation to age. In infants underweight dominated. In children ≥1 year short-stature was the main finding and one third of patients were overweight. Only a reduced number of trisomy 21 children had normal auxological parameters.

PO-0383 MONITORING IGG ON ORAL FLUID FOR THE MANAGEMENT OF CHILDREN AT RISK FOR CONGENITAL TOXOPLASMOSIS

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Background and aims Despite progress made for the diagnosis of congenital *Toxoplasma* infection *in utero* and at birth, serological follow-up in first year of life remains required to exclude or confirm congenital infection. To reduce the constraints of this follow-up, we developed a test to detect anti-*Toxoplasma* IgG in oral fluid.
Methods In 362 patients referred for *Toxoplasma* serology oral fluid was collected on two micro-sponges in parallel to blood sampling. A pilot study on 212 patients aged >15 months (274 samples) was performed to validate sampling procedures and develop an in-house indirect ELISA for the detection of

anti-*T. gondii* IgG in oral fluid. It was then applied to 150 children aged 0–15 months (341 samples) born from 133 women who seroconverted during pregnancy and 17 who remained seronegative. IgG on oral fluid were compared to serum IgG detected with MEIA AxSYM® Toxo IgG (Abbott Laboratories).
Results The pilot study validated the acceptability and the safety of the test and the adequate duration of sampling. IgG detected in serum and in oral fluid had a parallel kinetics among newborns (correlation coefficient: 0.59, $p < 0.0001$), with a concordant decline in the non-infected ones ($n = 110$), and matching raising or stable IgG in those who were congenitally-infected ($n = 23$).
Conclusions Collection of oral fluid is painless and inexpensive. Our new test provides a simple and rapid method to detect anti-*Toxoplasma gondii* IgG and to manage newborn at risk for congenital infection. It could have many other applications in pregnant women and other groups of patients.

PO-0384 THE PRACTICAL METHOD TO DIAGNOSIS OF FOURTEEN CASES OF GLYCOGEN STORAGE DISEASES IN OUR LABORATORY

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Glycogen storage diseases (GSD) are a group of inherited disorders of metabolism that result in storage of excess glycogen. Several well defined defects in one of the enzymes involved in the synthesis or degradation of glycogen have been described. There are over 15 types and they are classified based on the enzyme deficiency and the affected tissue (liver, muscle or both).

In this study, we wish to report the biochemical investigations adopted in main infantile GSD diagnosed in our laboratory.

Four steps diagnostic procedure have been assumed, taking into account several frequent clinical observations leading to further targeted biochemical parameters:

1. Assessment of the metabolic disorders with standard tests (fast blood glucose, uric acid, triglycerides, total cholesterol, ASAT, ALAT, CK, lactic acid).

2. Quantitative determination of glycogen in leucocytes (or erythrocytes) after extraction, precipitation and treatment with an throne reagent.

3. Oral galactose test with blood lactate and glucose estimations, in combination with a glucagon tolerance test to screen the main types of liver glycogenosis.

4. Lysosomal acid a-glucosidase activity when GSD type II (Pompe disease) is suspected.

Since 1995 and on the basis of this screening procedure and clinical features, 14 cases of GSD have been categorised:

· 6 forbes's disease (GSD III, debranching-enzyme deficiency)
· 3 von Gierke's disease (GSD I, glucose-6-phosphatase deficiency)
· 2 pompe's disease (GSD II, maltase acid deficiency)
· 1 Andersen's disease (GSD IV, branching-enzyme deficiency)
· 1 Hers's disease (GSD VI, hepatic phosphorylase deficiency)
· 1 GSD IX (phosphorylase kinase deficiency)

Our laboratory diagnostic approach include simple screening tests easy to implement in clinical chemistry laboratories. Thus, Pompe disease diagnosis is easily done in our laboratory. The measurement of tissue enzyme activities (liver and muscle) of the other enzymes is limited to some specialised laboratories. The molecular diagnosis offers a good alternative for GSD type 0, I and III but requires better financial means.

PO-385 WITHDRAWN

Miscellaneous

PO-0385a ADVERSE CHILDHOOD EXPERIENCES IN ALBERTA, CANADA: A POPULATION BASED STUDY

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Objective Adverse childhood experiences (ACEs) are associated with poor health outcomes in adulthood. We developed ACE risk profiles using a domain-specific approach and examined ACEs as risk factors for diagnosed physical and mental health conditions.
Method A computer-assisted telephone survey was conducted with a random sample of adults in Alberta, Canada. Eight questions were asked on adversity during childhood, based on the original ACE survey and modified to reflect the Canadian context and research methodology. Descriptive and multivariable analyses were conducted.
Results Among the 1207 respondents, the majority were married or living common-law (65.8%), had completed post-secondary education (78.2%), and were Caucasian (86.2%) with a mean age of 52.4 years (SD=16.3). Approximately one-third (27.3%) experienced at least one type of abuse, and almost half (49.5%) experienced at least one form of household dysfunction. ACEs were highly interrelated. Sixty-three percent fell into the low risk profile, with the remaining 37% divided among the three higher risk profiles. Overall, ACE risk profile was significantly associated with diagnosed mental health condition/addiction and chronic pain, controlling for sociodemographic characteristics.
Conclusion The ACE risk profile of ACEs in both the abuse domain and the household dysfunction domain conferred the greatest risk for poor health outcomes in adulthood. Given the interrelated of ACEs, a more comprehensive approach to conceptualization of ACEs is warranted. Results have implications for prevention of ACEs and recovery from ACEs to decrease disease burden. Strategies may include effective programs to prevent exposure to toxic stress and support nurturing and stable relationships for children and families.

Neonatal Brain and Development

PO-0386 NEONATAL SEIZURES ON AEEG MONITORING AFTER IN UTERO EXPOSURE TO VENLAFAXINE

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We report an unusual presentation of withdrawal from venlafaxine in a preterm baby of 29 weeks gestation, who presented with myoclonic seizures on second day of life. The seizures were confirmed with amplitude integrated EEG (aEEG). Other causes of neonatal seizures were excluded. She responded to treatment with phenobarbitone and phenytoin. Her MRI scan of the brain was normal and remains well on follow up. We believe this case to be the first report of seizure in a preterm baby resulting from maternal venlafaxine use and details the contribution of aEEG in the care of one such infant.