

## Abstracts

**Background** Disregulation of blood pressure (BP) and lipid metabolism is the basis of metabolic syndrome (MS), but interrelation of these MS components is not well studied today.

**Aim** To investigate influence of BP upon regulation of lipid metabolism in obese children.

**Patients and Method** 40 obese patients aged 10–16 years (13.9±0.27 y.o.) with high BP (HBP; 143.6±1.1/82.4±2.9 mm) – 1<sup>st</sup> group. 40 obese patients with normal BP (118.25±1.0/69±0.8 mm, p<0.01) – comparison group (2<sup>nd</sup> group). Examination included BP measurement and analysis of lipid profile.

**Results** Dyslipidemia in 1<sup>st</sup> group was significantly more frequent than in 2<sup>nd</sup> group: 85% versus 67.5% (p<0.05). Comparison of lipid spectrum showed that different disorders of lipid metabolism were more frequent and more significant in 1<sup>st</sup> group in comparison with 2<sup>nd</sup>: hypertriglyceridemia – 57.5% versus 22.5% (p<0.01) and 2.36±0.15 mmol/l (mean value) versus 1.64±0.04 mmol/l (p<0.01); decreased HDL-C – 37.5% versus 10% (p<0.01) and 0.91±0.01 mmol/l versus 1.03±0.02 mmol/l (p<0.05); increased LDL-C – 45.0% versus 45.7% and 3.72±0.11 mmol/l versus 3.32±0.14 mmol/l (p=0.06); increased index of atherogenicity – 75% versus 60% (p<0.05) and 3.93±0.1 versus 3.12±0.1 (p<0.01).

**Conclusion** Arterial hypertension in obese children was associated with more frequent and significant disorders of lipid metabolism. So, arterial hypertension in obese children should be estimated as an additional risk factor of atherogenicity.

#### 1025 THE ASSESSMENT OF THE VITAMIN D SUPPLY IN POLISH CHILDREN AT THE AGE OF 9–12 YEARS - MULTICENTRE RESEARCH

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<sup>1</sup>D Chlebna-Sokol, <sup>2</sup>J Golec, <sup>3</sup>J Karalus, <sup>3</sup>ZP Halaba, <sup>4</sup>E Karczmarewicz, <sup>5</sup>J Konstantynowicz, <sup>6</sup>B Kulik-Rechberger, <sup>7</sup>M Niedziela, <sup>4</sup>A Dobrzańska. <sup>1</sup>*Propedeutic Paediatrics and Bone Metabolic Diseases; 2*Medical University of Lodz, Lodz; <sup>3</sup>Medical University of Silesia, Katowice; <sup>4</sup>The Children's Memorial Health Institute in Warsaw, Warsaw; <sup>5</sup>University of Białystok, Białystok; <sup>6</sup>University of Lublin, Lublin; <sup>7</sup>University of Poznan, Poznan, Poland

**Introduction** It is universally known that the systemic deficiency of vitamin D may hamper the correct peak bone mass acquisition.

**Aim** The aim of the study was to determine the vitamin D supply in schoolchildren in Poland.

**Patients and methods** The study comprised 6 research centers from Poland. The healthy schoolchildren at the age of 9–11.99 were examined. In every child the liver metabolite of vitamin D was detected twice: after the winter and summer. The serum was analysed with the immunochemiluminescence method. The sufficient 25 OHD serum concentration was recognized at range of 20–100 ng/ml.

**Results** The 715 of children were examined. The greatest vitamin D shortages were observed in Szczecin and Białystok – in 95% and in 90% children. In Katowice and Lublin the lower concentration was detected in 89% and 88% of children and in Łódź and Poznań in 77% and 74%. The results of the 25OHD improved considerably after the summer. The greatest shortages were obtained in Poznań and Szczecin – in 52.9% and 42.1%. In Łódź the decreased concentration was observed in 41.5% of children. The lowest shortages were revealed in Lublin, Białystok and Katowice- in 28%, 26.3% and 26.3%.

#### Conclusions

1. The lower concentration of vitamin D in as many children indicates on adverse diet and climatic conditions.
2. The results of this study confirm the necessity of the prophylaxis of vitamin D deficiency in schoolchildren in Poland.
3. The considerable improvement of the 25OHD serum concentration after the summer may provide favourable influence of the sunlight.

#### 1026 EXCESSIVE FLUORIDE INTAKE IS ASSOCIATED WITH HYPERPARATHYROIDISM AND HYPOTHYROIDISM IN CHILDREN AND ADOLESCENT, JEDDAH- SAUDI ARABIA

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<sup>1</sup>RM Al-Raddadi, <sup>2</sup>SM Bahijri, <sup>3</sup>T Al-Khateeb. <sup>1</sup>*Postgraduate Center for Family and Community Medicine; 2*King Abdulaziz University, Jeddah, Saudi Arabia

**Background** Exposure to Fluoride (F) has increased significantly, so that individuals may be consuming more than recommended. Reported effects of excessive intake include reduced serum free thyroxine (FT4), triiodothyronine (FT3), calcium and increased parathyroid hormone (PTH) concentration.

**Objective** To investigate the prevalence of excessive Fluoride intake in apparently healthy children and adolescents, and explore its association changes in thyroid and parathyroid function in Jeddah-Saudi Arabia.

**Methods** 145 apparently healthy children and adolescents were recruited. 60 individuals satisfied selection criteria, and agreed to be enrolled. Subjects were examined dentally and clinically. Weights and heights were measured to calculate body mass index. Dental hygiene practices and fluoride intakes were recorded using recall method and food frequency questionnaires. Blood samples were obtained for the estimation of free thyroxine, triiodothyronine, thyroid stimulating hormone, Parathyroid hormone, calcium and phosphate. Fluoride was estimated in a samples of drinking water, beverages, and fasting urine of subjects. Total Fluoride intakes were calculated and used to subdivide groups into high and low or optimal intake subgroups.

**Results** Excessive Fluoride intake was identified among 36.7% of the individuals.

Calculated intake correlated with urinary excretion ( $r = 0.54$ ,  $p=0.0003$ ).

Significantly higher mean thyroid stimulating hormone and Parathyroid hormone and lower mean of free thyroxine, triiodothyronine, calcium and phosphate were found in various high intake subgroups, with some subjects having abnormal values.

**Conclusion** Excessive F intake is common, and is associated with hyperparathyroidism and hypothyroidism in studied population.

#### 1027 MINERAL PROFILE OF PNEUMOCOCCAL DISEASES IN THE CHILDREN

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<sup>1</sup>OM Horlenko, <sup>2</sup>VI Rusyn, <sup>3</sup>OO Boldizhar, <sup>2</sup>FV Horlenko, <sup>2</sup>NY Kishko, <sup>1</sup>AO Yankovska, <sup>3</sup>OM Moskal, <sup>1</sup>OA Pushkarenko, <sup>1</sup>AI Tomey, <sup>1</sup>GB Kosssey. <sup>1</sup>*Pediatrics and Infectious Diseases; 2*Surgical Diseases; <sup>3</sup>Internal Medicine, Uzhgorod National University, Medical Faculty, Uzhgorod, Ukraine

**Background and Aims** A mineral homeostases is association of processes of sorbtion, distributing, and elimination of mineral composition. Mineral matters play an important role in maintenance of acid-basic balance, osmolality, participate in the function of many enzymic systems, assists development of inflammatory process.

**Methods** Focus group included 21 children, aged 11.1±0.95 with community-acquired pneumonia, Pneumonia lower lobes acuta (PLA) in which was identificate S.Pneumonia.

**Result** In the majority of cases the results of physical examination were satisfactory. The level of zinc in blood plasma of PA patients were 0.68±0.17 mg/l, 0,670.03 mg/l in urine, copper – 0.42±0.03 mg/l in blood plasma, 0.36±0.02 mg/l in urine, iron- 0.75±4.2 mg/l in blood plasma, 23.48±1.75 mg/l in urine, phosphorus – 473.10±11.25 mg/l in blood plasma, 312.50±11.84 mg/l in urine, iodine – 70.23±5.81 mg/l in blood plasma, 60.19±1.21 mg/l in urine. There were a positive correlation with the levels of Fe/Cu in blood serum ( $r=0.64$ ). With the level of calcium ratio of Fe/Cu had a

significant negative correlation ( $r = -0.87$ ). Increase of Cu, Fe levels in the patients with PA were identificate in the prevalence cases.

**Conclusions** Microelement status of patients with pneumonia is characterized by synergistic correlation between Fe and Cu ( $r = 0.64$ ), and reverse dependence between Ca and Fe/Cu ( $r = -0.87$ ), increase of Cu, Fe levels. Our dates show the dynamics of development of inflammatory process in Lung and possible role of violations of microelement status of child in pathogeny of diseases of breathing organs.

#### 1028 RESPIRATORY CHAIN DISORDERS: REVIEW OF 16 CASES

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J Sales Marques, Al Pinto Pais, D Rocha, A Vieira, H Santos. *Centro Hospitalar Vila Nova de Gaia/Espinho EPE, Vila Nova de Gaia, Portugal*

**Introduction** Respiratory chain disorders (RCD) are a heterogeneous group of diseases associated with multisystemic disorders. The diagnosis should be considered if there are 2 major criteria or 1 major and 2 minor criteria (Modified Walker Criteria).

**Purpose** Medical records of 16 cases of RCD diagnosed in Metabolic Unit of our hospital, between 2005 and 2010 were analyzed.

**Results** The results showed that all the patients have psychomotor delay and more than half cases hypotonia, strabism and acquired microcephaly at presentation. Other symptoms were multisystem such as: neurosensory deafness (1/16), mioclonic epilepsy (3/16), intestinal duplication (1/16), ductus arteriosus persistent (1/16), renal hypoplasia (2/16). We found important association with endocrinological changes (9/16), hypothyroidism in most situations, but also hypoparathyroidism, insipidus diabetes, growth hormone defect and hyperinsulinemia. Complex 2 deficiency was the most common cause of RCD (8/16). In one case we found depletion in mitochondrial DNA. No histopathology abnormalities were found in the muscle biopsy. Only (8/16) exhibited elevated plasma lactate. The treatment with Coenzyme Q10, carnitine and ketogenic diet seemed to improve their clinical course (less epileptic crisis after ketogenic diet – 6/6, better concentration after coenzyme Q10 – 6/16, less hypotonia after carnitine – 5/11).

**Discussion and Conclusion** Mitochondrial cytopathies should be considered in patients with an unexplained combination of neuromuscular and/or nonneuromuscular symptoms, with a progressive course, even if oxphos studies are normal.

Endocrinological changes are an important association of respiratory chain disorder and hormone screening must be included in all the patients with this metabolic disease.

#### 1029 GLYCINE SUPPLEMENTATION IMPROVES THE CONCENTRATION, SOCIALIZATION, PERCEPTION AND AUTONOMY OF PATIENT WITH CREATINE TRANSPORT DEFECT

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J Sales Marques, D Rocha, Al Pinto Pais. *Centro Hospitalar Vila Nova de Gaia/Espinho EPE, Vila Nova de Gaia, Portugal*

**Introduction** Creatine transport defect (CTD) is a x-linked disorder with neurological symptoms. Glycine can help inhibit the neurotransmitters and supply the body with glucose needed for energy. Also act as a creatine precursor.

**Purpose** Treat with oral glycine in the period of 12 months, a patient with CTD and compare the clinical, analytic and brain magnetic resonance spectroscopy results, before and after therapy.

**Material** A fourteen year-old boy with CTD diagnosed at the age of five years, treated unsuccessfully with daily 20 mg of oral methylphenidate for is behaviors disturbances.

**Method** Oral glycine in a dose of 250 mg/kg/day, divided in two doses.

**Results** After 12 months, we found some improvement in concentration, socialization, perception and autonomy. No evolution of speech. The creatine peak was slightly better than before (5 to 8 mm). Urine creatine level reduces from 24427 to 10994  $\mu\text{mol}/\text{mmol}$  creatinine.

**Discussion** We believe that the clinical and analytic evolutions are associated with increase creatine peak level secondary to oral glycine. Glycine, because of its inhibitory action over the neurotransmitters, in research studies has shown that helps improve memory retrieval loss in those patients that suffer from a wide variety of sleep-depriving conditions, including schizophrenia, Parkinson and Huntington diseases. He also has a sedative effect and is used in attention-deficit, by reducing the excitability of nerves cells. Glycine is useful in patients with CTD and should be encouraged to use because has no side effect and can improve some behaviours disturbances that is common in this disease.

#### 1030 A CASE OF RHIZOMELIC CHONDRODYSPLASIA PUNCTATA IN NEWBORN

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N Karabayir, G Keskindemeci, E Adal, O Korkmaz. *Kanuni Sultan Suleyman Training and Research Hospital, Istanbul, Turkey*

Rhizomelic chondrodysplasia punctata is a rare autosomal recessive peroxisomal disease. The main features of the disease are shortening of the proximal long bones, punctate calcifications in the metaphysis and epiphysis of long bones and the thoracic and lumbar vertebrae, dysmorphic face, and severe growth retardation, whereas cervical spinal stenosis may also rarely be present. Imaging of the brain and spinal cord in patients with this disorder may aid prognosis and guide management decisions. We report the newborn diagnosed as rhizomelic chondrodysplasia punctata with cervical stenosis. As far as we know, our case is the first case of autosomal recessive form with a cervical spinal stenosis detected in the neonatal period.

#### 1031 AMINOACIDOPATHIES: REVIEW AND DATA OF 12 YEARS EXPERIENCE FROM A SPANISH TERTIARY CARE CENTER

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L Ferreras Antolín, E Moreno Medinilla, J Blasco Alonso, V Navas López, J Serrano Nieto, A Urda Cardona, A Jurado Ortiz, C Sierra Salinas. *Hospital Marterno Infantil Carlos Haya, Málaga, Spain*

**Background/Aims** Range and severity of symptoms hugely variable in aminoacidopathies, mainly diagnosed during acute episodes. Tandem Mass spectrometry (MS/MS) used in our unit since 2010 for diagnosing asymptomatic infants (very important for prognosis).

**Methods** Retrospective, descriptive study in which field data were collected from clinical histories of patients diagnosed of aminoacidopathies (excluding phenylketonuria) since 2000 till 2012.

**Results** 30 children detected. 22/30 male; 24/30 caucasians.

10/30 diagnosed by newborn screening, all of them asymptomatic: 1 methylmalonic acidemia (MMA), 3 glutaric aciduria type 1, 2 homocystinuria, 2 methylcrotonylglycinuria, 1 hipermetioninemia and 1 maple syrup urine disease.

20/30 diagnosed because of clinical symptoms, 2/20 since MS/MS newborn screening was performed. Mean age of clinical debut in intoxication type aminoacidopathies was 64.5 days (median 8 days). Most frequent symptoms were clouding of consciousness (9/20), convulsions (2/20) and apnoea (2/20). Laboratory results showed metabolic acidosis (6/20), hyperammonemia (8/20), coagulation defects (4/20) and hipoglycemia. Main complications were: shock (9/20), multiple organ failure (5/20), coagulopathy (4/20), brain injury (1/20), liver failure (1/20) and seizures (2/20). The final diagnosis was: 5 OTC-deficiency, 2 citrullinemia, 3 methylmalonic



## 1027 Mineral Profile of Pneumococcal Diseases in the Children

OM Horlenko, VI Rusyn, OO Boldizhar, et al.

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