

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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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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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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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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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