significant negative correlation ($r = -0.87$). Increase of Cu, Fe levels in the patients with PA were identifiable 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/Ca ($r = -0.87$), 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 pathogenicity of diseases of breathing organs.

**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 psycho-motor delay and more than half cases hypotonia, strabism and acquired microcephaly at presentation. Other symptoms were multisystem such as: neurosensatorial deafness (1/16), mioclonic epilepsy (3/16), intestinal duplication (1/16), ductus arterious persistent (1/16), renal hypopasia (2/16). We found important association with endocrinological changes (9/16), hyperthyroidism 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, carntine 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 carntine – 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.

**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 methylyphenidate 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 μmol/m mol creatinine.

**Discussion** We believe that the clinical and analytic evolutions are associated with increase creatine peak level secondary to oral glycine. Glycine, because of is 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.

**RESPIRATORY CHAIN DISORDERS: REVIEW OF 16 CASES**

doi:10.1136/archdischild-2012-302724.1028


**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 psycho-motor delay and more than half cases hypotonia, strabism and acquired microcephaly at presentation. Other symptoms were multisystem such as: neurosensatorial deafness (1/16), mioclonic epilepsy (3/16), intestinal duplication (1/16), ductus arterious persistent (1/16), renal hypopasia (2/16). We found important association with endocrinological changes (9/16), hyperthyroidism 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, carntine 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 carntine – 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.
Background and Aims The Suspicion Index (SI) screening tool was developed to identify suspected patients with Niemann-Pick disease type C. (NP-C, Neurology, 2012). The SI provides Risk Prediction Score (RPS) based on NP-C symptoms within and across domains (visceral, neurological, and psychiatric). To further examine a) discriminatory power of the SI by age and b) symptom-associations by NP-C suspicion-level and leading symptoms.

Methods The original retrospective data were split into three age groups, where NP-C positive cases were: >16 years (n=30), 4–16 years (n=18), and < 4 years (n=23), and patients’ RPS was analysed by logistic regression. Co-occurrence of symptoms within groups of suspicion-level (low, medium, and high) and leading symptoms (presence/absence of ataxia, cognitive decline, psychosis, and splenomegaly) were analysed descriptively.

Results NP-C positive cases vs. controls showed strong discriminatory power of RPS. Area under the Receiver Operating Characteristic curve was 0.964 (>16 years) and 0.981 (4–16 years) but a weaker 0.562 for infants (< 4 years). Patients with RPS < 70 were characterised by a lack of psychiatric symptoms and low levels of neurological involvement, suggestive of a more visceral phenotype. In patients >4 years, prominent leading symptoms’ associations were: ataxia with “dystonia, dysarthria/dysphagia and cognitive decline”; psychosis with “dysarthria/dysphagia”; and psychotic symptoms with “cognitive decline and treatment-resistant psychiatric symptoms”.

Conclusions The SI tool maintains strong discriminatory power in patients >4 years but is not as useful for infants < 4 years. The SI is informative regarding the association and co-occurrence of symptoms in patients with NP-C.
1031 Aminoacidopathies: Review and Data of 12 Years Experience from a Spanish Tertiary Care Center

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