Purpose

Also act as a creatine precursor.

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/mmol creatine.

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.

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).

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: neurosensorial deafness (1/16), micronuclear epilepsy (3/16), intestinal duplication (1/16), ductus arterious persistent (1/16), renal hypoplasia (2/16). We found important association with endocrinological changes (9/16), hypothyroidism in most situations, but also hypoparathyroidism, insulin 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 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 ophoxos 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 AUTONYM OF PATIENT WITH CREATE TRANSPORT DEFECT

doi:10.1136/archdischild-2012-302724.1029


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 methylnenidate for is behaviors disturbances.

Method

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