In the Name of God

case presentation

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Case presentation

An 8 yr old boy presented with Hx. of abnormal gait , dystonia , difficulty in walking , and dysarthria for a few mo,s.

PEx.:

He was plethoric, had significant neurological findings and also had hepatomegaly with stigmata of CLD(cirrhotic).

FHx.:

Was positive for index case (his brother) with the same problems.

Case presentation

Workups (in the original center) :

CBC: Hb:19 ,18 , WBC , Plt: WNL , ABG: Nl

AST:238 , ALT:218 , TP:6.7 , Alb:4 , Cer:22mg/dl

KF ring was negative (not seen , 2 times)

24 hr urine CU:85mg

Case presentation

US: Hepatomegaly with course echogenicity and signs of PHT.

Brain MRI: Basal ganglia involvement

What is the Dx?

With impression of Wilson disease , was treated with trientine for about 5 mo., but had no improvement.

Case presentation

He was referred to us for further evaluation.

The unusual points which were incompatible with Wilson disease in this case were:

1- Polycythemia

2- Absence of KF ring

Case presentation

Considering unusual points, other possibilities with the same presentations were considered.

A case with:

1- Significant neurologic involvement

2- Polycythemia

3- Chronic liver disease(Cirrhosis)

4- Positive FHx

5- Negative KF ring

Case presentation

DDx:

The only disease with the same presentations (neurologic problems , polycythemia and cirrhosis) was Hypermanganesemia.

Serum manganese (Mn) was requested, which was very high.

Final Dx.:

Familial (Genetic) Hypermanganesemia

Case presentation

He was treated with:

1- IV, Disodium Calcium Edetate,
20mg/kg/dose (5 doses /month).

2-Fe supplementation

FU: He was followed 3 ,6 and 9 mo , after treatment :

1- Had significant neurologic improvement

2- Nl Hb

3- Nl LFT

Manganese(Mn)

Introduction

Manganese (Mn) is an essential element for metabolic pathways

but it can be toxic when present in excessive amounts in the body.

Normal manganese blood level is 1.7-2.4mg/dl

Hypermanganesemia:

1-Mild <7mg/dl

2- moderate:7-12mg/dl

3- Severe >12mg/dl

Introduction

There are two types of hypermanganesemia with dystonia:

1- Hypermanganesemia with dystonia, polycythemia and cirrhosis (HMDPC)

2- Hypermanganesemia with dystonia

They are distinguished by their genetic causes and certain specific features.

It is an autosomal recessive disorder.

Introduction

This is a rare disorder and is one of the potentially treatable

inherited metal storage disorders.

Mn accumulates abnormally in the brain, blood and liver.

Together with Wilson's disease, is the only potentially treatable

inherited metal storage disorder to date.

It can be fatal if left untreated.

Disodium Calcium Edetate is the treatment of choice.

Pathogenesis

Hypermanganesemia with Dystonia, Polycythemia and Cirrhosis

(HMDPC) occurs due to mutation in the *SLC30A10* gene.

Hypermanganesemia along with dystonia, polycythemia,

characteristic MRI brain findings in the basal ganglia, and

chronic liver disease are the hallmarks of an inherited Mn

transporter defect due to mutations in the *SLC30A10* gene

Management

Regular chelation therapy with intravenous disodium calcium edetate improves blood manganese levels and Neurologic findings and halts liver disease.

Chelation therapy with calcium disodium edetate increases urinary manganese excretion and lowers its blood level.

It also slows disease progression.

Management

Chelation therapy with calcium disodium edetate has been shown to produce a good response, especially if initiated at an early age.

The younger the patient, the better is the response to chelation therapy.

Chelation therapy with calcium disodium edetate 1500 mg/m^2/day (20mg/kg/dose) with five daily doses every 4-6 weeks.

With thanks