Use of ammonium tetrathiomolybdate in Wilson disease


Background and importance

Wilson disease is a rare autosomal recessive disorder. It is characterized by an excessive accumulation of copper in the body, mainly in the liver, brain and cornea, leading to different manifestations, in which neuropsychiatric and hepatic ones predominate.

Therapeutic management:
- copper chelating agents (D-penicillamine, trientine)
- drugs that hinder the absorption of copper (zinc salts)
- Ammonium tetrathiomolybdate, an experimental treatment, has also been used for periods of 8 weeks in patients with neurological presentation under compassionate use.

Aim and objectives

To evaluate the effectiveness and toxicity of ammonium tetrathiomolybdate in a patient with Wilson disease.

Material and methods

A 33-year-old man with Wilson disease

Neurological manifestations + increased transaminase levels + Presence of Kaysher-Fleisher ring + Mutation c3359T>A (p.Leu1120*) on exon 15 in the ATP7B gene

1. TRIENTINE
2. ZINC SULFATE + AMMONIUM TETRATHIOMOLYBDATE
3. TRIDENTE
4. D-PENICILLAMINE
5. AMMONIUM TETRATHIOMOLYBDATE
6. ZINC SULFATE + AMMONIUM TETRATHIOMOLYBDATE

4 months later
Clinical worsening

7 weeks later
Worsening of liver function

1. TRIENTINE
2. ZINC SULFATE + AMMONIUM TETRATHIOMOLYBDATE
3. TRIDENTE
4. D-PENICILLAMINE
5. AMMONIUM TETRATHIOMOLYBDATE
6. ZINC SULFATE + AMMONIUM TETRATHIOMOLYBDATE

6 months later
Deterioration of neurological and functional situation. In the following months, the neurological clinic progressively improved

Currently, 15 months later

Results

Currently, after 15 months of treatment with ammonium tetrathiomolybdate combined with zinc sulfate, patient has experienced motor and cognitive-behavioral improvement, and maintains normal hematologic and hepatic function.

Conclusion and relevance

In our patient, ammonium tetrathiomolybdate has been effective and well tolerated for a prolonged period. It could be an alternative to patients with neurological manifestations.