A case of mitochondrial neurogastrointestinal encephalopathy or MNGIE syndrome, why not?

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Introduction

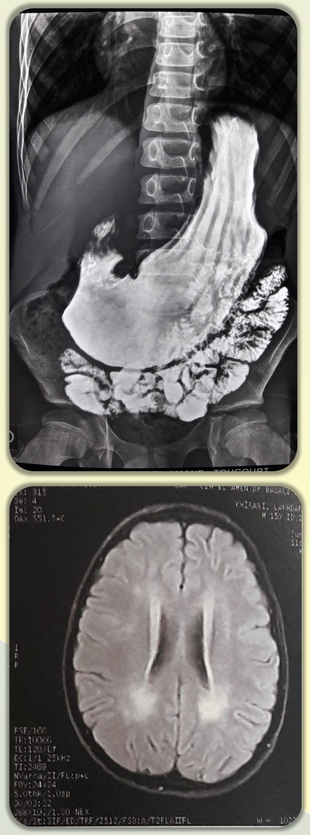
Mitochondrial neuro-gastro-intestinal encephalopathy or MNGIE syndrome is characterized by progressive gastrointestinal dysmotility, cachexia, peripheral neuropathy, ophthalmoplegia and leuko-encephalopathy

Objective

To describe a case of MNGIE syndrome and show the diagnosis difficulty

Material & methodes

15-year-old boy from a consanguineous marriage, with no past medical history,who presents a chronic clinical manifestation evolving since the age of 12 years with chronic vomiting,anorexia,normal transit, 2 afebrile seizures episodes,an academic delay.The clinical examination shows cachexia, a failure to thrive, skin heat,asthenia,a tender abdomen without abdominal mass or hepato-slpenomegaly, absence of oral lesion or phanere disorders, proctological examination is normal, no neurological deficiency, blood pressure 100/80 correct for age, negative urine chemistry.The complete biological check-up (complete hepatic function, renal, haemostasis, thyroid, phosphocalcic,coeliac serology) shows hyponatremia at 118mmol/ l the upper digestive endoscopy reveals esophagitis and erythematous gastritis ,the oesophagus and duodenal biopsies are normal. the oesophago-gastro-duodenal transit shows gastroparesis without obstruction.The brain MRI shows an incipient leukodystrophy.the radio-clinic picture is suggestive of a myoneuro-gastrointestinal encephalopathy.the study of the TYMP gene (NM_001953) reveals a homozygous mutation.



Results

Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE) is a clinically recognizable autosomal recessive disorder .The onset of MNGIE syndrome ranges from 10 to 40 years, the symptomatology is progressive and dominated by severe gastrointestinal disorders (vomiting, gastroparesis) leading to cachexia as in the case of our patient, the neurological involvement dominated by ophthalmoplegia and peripheral neuropathy which are absent in our patient at this stage. The brain imaging shows very often a leukodystrophy which remains subclinical

Conclusions

The MNGIE syndrome is a pathology of unknown prevalence, and of difficult diagnosis often delayed because of the ignorance of the disease.

References

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