

Case Report

New cases of Neuroferritinopathy

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Abstract

Male patient, aged 27 years, has ESRD and on regular hemodialysis for 6 years. Over the last 3 years, His serum ferritin has been dropped to very low levels without corresponding anemia or iron deficiency. Possible causes were looked for and Neuroferritinopathy was considered. Brain MRI was done and revealed related changes. Finally, neuroferritinopathy was suggested.

Another female patient, aged 33 years, was admitted for IV iron therapy as her serum Ferritin was very low, 5 ng/ml. However, her remaining lab. Tests revealed normal both Hb.%, 12.5 g/dl, and Tsat, 38%. Her serum ferritin was repeated but it was again so low, 3 ng/ml. Finally, the patient was diagnosed as Neuroferritinopathy and iron therapy was cancelled.

Learning points

New cases of severe hypoferritinemia, without neither anemia nor iron deficiency, are appearing in early adulthood patients. Neuroferritinopathy should be highly suggested as the underlying diagnosis of such cases, requiring a very cautious use of iron supplements that should be only guided by iron saturation, not other lab. Parameters.

Background

Neuroferritinopathy is an extremely rare disease with less than 100 reported cases worldwide. It is a form of progressive neurodegeneration with brain iron accumulation, causing parkinsonism, dystonia, cerebellar signs, cognitive decline and other neurologic deficits [1]. It is due to a mutation in the FTL gene encoding the ferritin light chain leading to decreased iron binding capacity and excessive iron release with a consequent accumulation in tissues, especially the brain [1-3]. Lab. Tests shows low serum Ferritin and brain MRI invariably

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reveals abnormal deposition of iron in the basal ganglia as well as cystic changes and necrosis in basal ganglia [4].

Case Presentation

A 27 years old male Saudi patient has ESRD due to neurogenic bladder, on maintenance hemodialysis for 6 years. He is doing well with his sessions without intra-dialytic complications. His maintenance home therapy include daily oral iron (Ferglobin cap. od) for all these 6 years as he has a past history of severe reactions (anaphylaxis) to IV iron (Ferosac). On revising his past lab. Tests, it was surprising that his serum Ferritin was always low (51 then 31 then 14) despite normal Tsat and being even not anemic. Oral iron was stopped pending more evaluation.

Another female Saudi patient, aged 33 years, and having no history of any CKD. She is a VIP patient and having no symptoms. However, she was admitted for IV iron therapy as her serum Ferritin was very low, 5 ng/ml, as per her treating hematology consultant. However, her remaining lab. Tests revealed normal both Hb.%, 12.5 g/dl, and Tsat, 38%. Her serum ferritin was repeated but it was again so low, 3 ng/ml. Finally, the patient was diagnosed as Neuroferritinopathy and iron therapy was cancelled. The situation was communicated with both the patient and her treating consultant. All were satisfied with holding iron therapy as there was neither anemia nor definite iron deficiency.

Investigations

Lab. Tests in the last year for the 1st patient were as following

1. Hb.% : 11-12 g/dl.
2. Tsat is : 30-35%.
3. Ferritin : 8-22 ng/ml. (Ref. 200-500 ng/ml for HD patients)
4. Iron : 5.5 – 11.2 umol/L. (Ref. 60-170 ug/dl OR 11-30 umol/L)
5. TIBC : 22 – 38 umol/L (Ref. 240-450 ug/dl OR 43-81 umol/L)

MRI Brain

It revealed microcystic changes in the basal ganglia.

Treatment

The patient is off both ESA and Iron therapy for the 2 years.

Outcome and follow-up

For the last 2 years, both his Hb.% and iron saturation are maintained with targets.

Funding statement

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Declaration of interest

There is no conflict of interest by all authors.

Patient consent

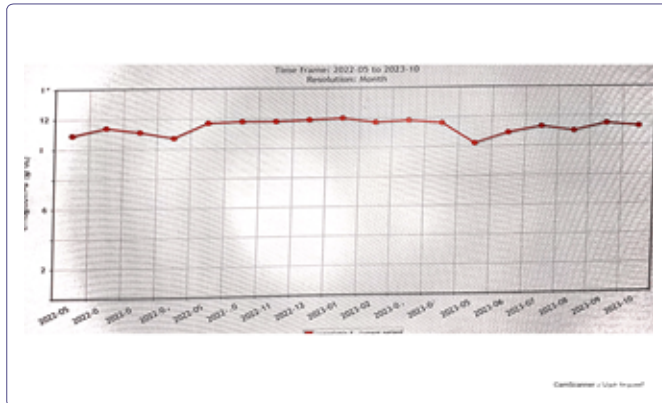
Both patients agreed for publication and consents were done.

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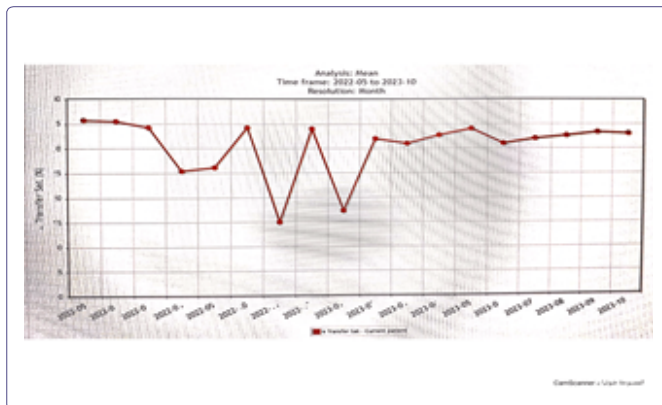
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Lab Curves over 2 years

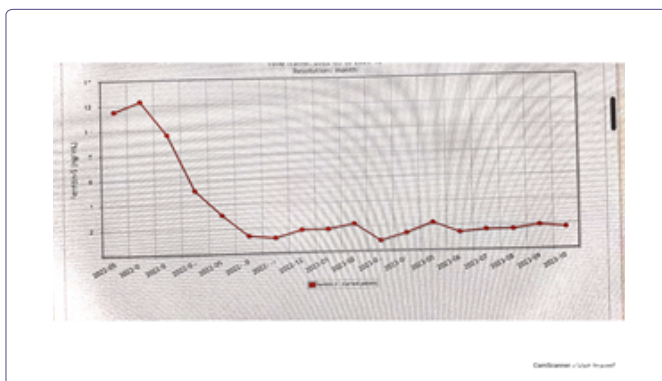
1- Hb.% : (Maintained within target)



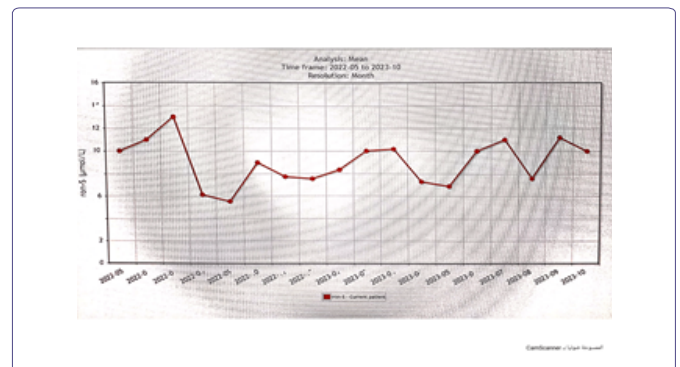
2- Tsat : (Maintained within target)



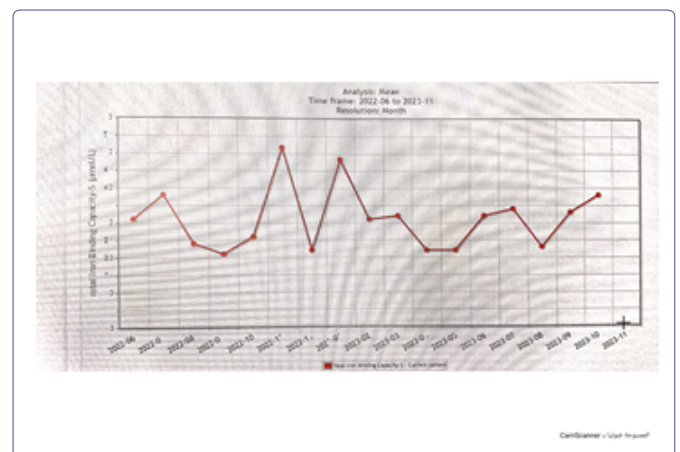
3- Serum Ferritin : (persistent drop)



4- Serum iron : (always low)



5- TIBC : (always low)





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