Figure 1.

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A long-term follow-up of an Imerslund-Grasbeck syndrome patient with proteinuria

Dear Editor,

Imerslund-Grasbeck syndrome (IGS) is a rare autosomal recessive disorder characterized by megaloblastic anemia due to selective vitamin B₁₂ malabsorption and asymptomatic proteinuria. IGS occurs in the first 1-2 years of life and megaloblastic anemia is responsive to parenteral vitamin B₁₂ treatment. It is thought that proteinuria is benign in IGS; however, there is no sufficient number of follow-up series in IGS.

Case report

A 22-year-old woman had been referred to our pediatric outpatient clinic with the complaints of pale skin, loss of appetite, ataxia and diarrhea-constaipation periods when she was 2-year-old. The clinical examination and laboratory studies revealed pallor of conjunctiva, megaloblastic anemia with vitamin B₁₂ deficiency (serum vitamin B₁₂ level <150pg/ml; hemoglobin: 6.5g/dl; MCV: 104fl and peripheral blood smear with hypersegmented neutrophils) and mild proteinuria (less than 0.5g/day) with absence of kidney function abnormality.

Conflict of interest

The authors declare that there is no conflict of interest associated with this manuscript.


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Two renal biopsies were performed because of persistent proteinuria; however, there was no remarkable historically changes. She was diagnosed with IGS in the light of this clinical picture. Anemia and neurological symptoms were improved with vitamin B12 therapy in the next few weeks. Mild proteinuria remains persist with normal kidney function and she is being still followed-up with periodically for proteinuria.

IGS was firstly described in 1960 by Olga Imerslund and more than 300 cases have been published to date. In IGS, vitamin B₁₂ is completely abolished and if untreated with parenteral therapy the disease is fatal. A recent study revealed a biallelic mutation either in cubulin or amnionless genes cause IGS. Both proteins act as a receptor for intrinsic factor-vitamin B₁₂ complexes as well as cubulin is an albumin binding protein important for renal tubular albumin reabsorption. Because of absence of glomerular damage in kidney biopsies progressive kidney disease is not usual. Broch et al enrolled 14 patients to a long term follow-up study and exhibited no deterioration in kidney function. Limited numbers of cases have been observed almost 50 years and renal prognosis is excellent. We aimed to announce our case with IGS who has a good renal prognosis over 20 years follow-up.

Conflict of interest
The authors declare that there is no conflict of interest associated with this manuscript.


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Reacción adversa por la administración intravenosa de hierro: ¿hiersensibilidad o efecto secundario?

Se trata de una mujer de 42 años que inicia programa de hemodiálisis por catéter yugular derecho tunelizado tras bilecromía por hiperferremia. En el posoperatorio la paciente requirió la transfusión de 2 concentrados de hematies. Diez días más tarde se comprueba: hemoglobina: 9.6 g/dl; hematocrito: 28.4%; volumen corpuscular medio: 87.1 fl; hierro: 56 µg/dl; ferritina: 233 ng/ml; índice de saturación de la transferrina: 18 %; ácido fólico: 22 ng/ml; vitamina B₁₂: 921 pg/ml; proteína C reactiva: < 5 mg/l; Kt/V: 1.7. Recibe tratamiento con omeprazol, complejo vitamínico B, ácido fólico y darbepeotina 30 microgramos semanales. Se pauta 100 mg de hierro succrosa (Venofer®) a pasar por vía intravenosa en una hora poshemodiálisis. A los 15 minutos de iniciada la infusión, la paciente refiere prurito generalizado, sensación de quemazón lingual y de hiperestesia peribucal. Exploración física: tensión arterial 100/60 mmHg, auscultación cardíaca y pulmonar normales, no lesiones cutáneas. Se interrumpe la administración de hierro, con lo que cede paulatinamente la sintomatología. En el siguiente intento se premedica a la paciente con dexclorfeniramina y paracetamol. La reacción se repite de forma idéntica y se reproduce también con hierro carboximaltosa (Ferinject®). Se consulta al Servicio de Alergología: pruebas epikutánneas negativas para ambos preparados férricos; cuadro compatible con efecto secundario. Las manifestaciones clínicas reaparecen de forma atenuada con las administraciones sucesivas de hierro, sin mayores implicaciones.

La tasa de efectos adversos relacionados con la administración de diversos preparados de hierro intravenoso (hierro dextrano de alto y bajo peso molecular, gluconato férrico, hierro succrosa) se sitúa en torno a 38 por millón⁶. El prurito asociado al hierro carboximaltosa se describe de forma aislada como poco frecuente (entre un 1/100-1000 de los pacientes)⁷;