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Short communication

Ocular involvement in atypical haemolytic uraemic syndrome☆

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ABSTRACT

Case report: The case is presented of a young man with an atypical haemolytic-uraemic syndrome (aHUS), complicated with bilateral serous retinal detachment, cotton wool spots, and a branch artery occlusion. Treatment with plasmapheresis, haemodialysis and systemic eculizumab led to the blood and urine parameters returning to normal, as well as resolution of the retinal anomalies. Genetic analysis show both mutations in complement factor H and C3.

Discussion: Haemolytic-uraemic syndrome (HUS) is a thrombotic microangiopathy characterized by microangiopathic haemolytic anemia, thrombocytopenia, and acute renal failure. Atypical HUS is caused by genetic mutation of complement system. Ocular involvement is an unusual manifestation of this rare syndrome.

Afectación ocular en un síndrome hemolítico urémico atípico

Palabras clave:
Desprendimientos serosos retinianos
Retinopatía tipo Purtscher
Síndrome hemolítico urémico atípico
Microangiopatía trombótica

RESUMEN

Caso clínico: Varón joven con síndrome hemolítico urémico atípico (SHUa) complicado con la presencia de desprendimientos serosos retinianos bilaterales (DSR), múltiples exudados algodonosos y una oclusión de rama arterial. El tratamiento con terapia plasmática, hemodiálisis y posteriormente con eculizumab, permitió una rápida normalización de los parámetros sanguíneos y urinarios y la resolución de las alteraciones retinianas. El análisis genético mostró 2 mutaciones en el factor H y C3 del sistema complemento.

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Discusión: El síndrome hemolítico urémico (SHU) es una microangiopatía trombótica con anemia hemolítica, trombocitopenia e insuficiencia renal aguda, que se denomina atípico cuando está causado por una alteración genética del sistema complemento. La afectación ocular es una rara manifestación de este infrecuente síndrome.

Introduction

The haemolytic uraemic syndrome (HUS) is a clinical entity defined by the triad of non-immune microangiopathic haemolytic anemia, thrombocytopenia and acute renal insufficiency.[1] Histological lesions are characterized by systemic thrombotic microangiopathy (TMA) that preferably compromises kidney vessels.[1] Atypical HUS (aHUS) is a very rare variant with worse prognosis and is caused by a deregulation of the alternative pathway of the complement system and determined by genetic alterations.[1] The pharmacological blockage of the complement with eculizumab, a monoclonal antibody, usually obtains a good clinic response.[1] Ocular involvement is rare in aHUS and serous retinal detachment are exceptional.[2]

Clinical case

Male, 29, who visited the emergency room due to nausea, oral intolerance, vomiting, intermittent headaches, feeling bloated and in anuria. Examination revealed skin paleness, slight hepatomegalia and arterial pressure of 210/120 mmHg. The most relevant parameters in blood analysis were Hb 6.5 g/dl, with 6.45% reticulocytes and between 3% and 4% schistocytes in smear, 129,000 platelets, urea 342 mg/dl, creatinin 31 mg/dl, Na 132 mEq/l and in urine: proteinuria 4+, nitrites+ with 1–4 leukocytes and 5–10 red blood cells through field in sediment. Chest X-ray revealed interstitial edema without cardiomegalia, while abdominal echography showed normal size kidneys with poor cortical-sinusal differentiation. The patient was diagnosed with HUS, transfusion was carried out followed by hemodialysis and plasmapheresis. Urine, blood and feces cultures were negative.

Two days after being admitted to nephrology, ophthalmological examination revealed bilateral retinal serous detachments, together with cotton wool exudates surrounding vascular arches and a small arteriolar obstruction in the LE macula. However, the patient exhibited a VA of 0.9 in both eyes (Figs. 1–3). After being administered 3 sessions of plasmapheresis, treatment with eculizumab was initiated, continuing with hemodialysis on alternating days. After 15 days, the patient started to recover diuresis and at month 2 dialysis was suspended. Kidney biopsy showed a histological condition of thrombotic microangiopathy with glomerular and vascular involvement in subacute phase, interstitial fibrosis and slight tubular atrophy.

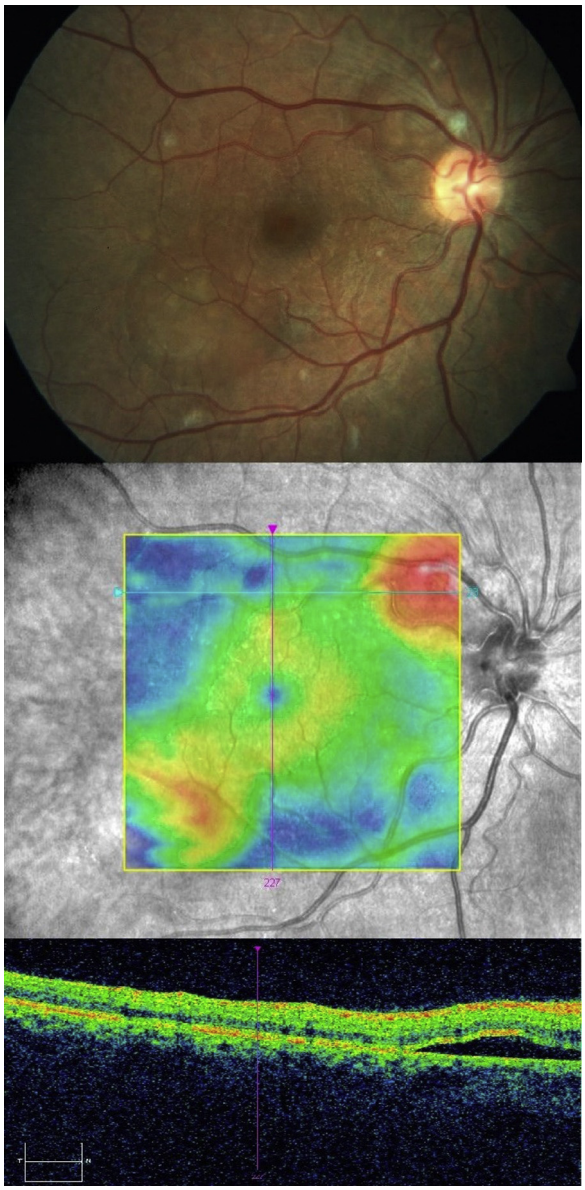

Fig. 1 – RE: retinograph with multiple cotton wool exudates and serous detachments; OCT with tomographic thickness map and section of the peripapillary serous detachment.

Complement analysis revealed a deficit of C3: 61.9 mg/dl (75–135) and factor H: 10.85 (12–56) explained by mutations in C3 and factor H, as shown by the subsequent genetic study. Both mutations were also present in the patient's sibling. Hematological parameters normalized before 2 months,