Budd-Chiari syndrome (BCS) as acute presentation of polycythemia vera (PV) in an adolescent

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Argomento: Caso clinico

Background

BCS is a rare condition characterized by obstruction of hepatic blood outflow without constrictive pericarditis or right heart failure. Adult BCS often presents during its chronic course; radiological interventions (in particular Transjugular Intrahepatic Portosystemic Shunt, TIPS) demonstrate high success rate and good outcome. The majority of BCS adult patients are diagnosed with an underlying thrombophilic disorder – especially PV, a *BCR/ABL1*$^{-}$ myeloproliferative neoplasm characterized by augmented red cell mass.

Both these conditions are extremely rare in children.

Case report

We report the case of a 16 year-old patient admitted to a Serbian hospital for acute liver failure with hepato-splenomegaly, massive ascites and erythrocytosis. The boy was referred to the PICU at Papa Giovanni XXIII Hospital in Bergamo (Italy) in October 2018. Abdominal angio-MRI showed BCS secondary to hepatic veins occlusion. Hematological investigations revealed the diagnosis of polycythemia vera: hemoglobin 18 mg/dl with hematocrit 54%, normal bone marrow biopsy, presence of JAK2V617F mutation – with no other associated thrombophilic conditions. The patient was managed with supportive medical treatment, including anticoagulation with heparin and antiaggregation with acetylsalicylic acid, until the radiological intervention was carried out. TIPS was successfully performed a week after the admission. In the post-surgical period, the patient developed chylous ascites that completely resolved in three weeks. Anticoagulation therapy continued with warfarin; cytoreductive therapy with hydroxyurea was introduced to obtain control of hematological parameters. At eight-weeks follow up, the patient was asymptomatic, with improving liver-function tests. Four months later, the TIPS is still patent and the clinical picture is resolved.

Discussion

PV is a very rare condition in childhood. Half of pediatric PV patients are asymptomatic at the time of diagnosis; thrombosis is not common in these patients. We describe the case of an adolescent patient presenting with a life-threatening complication of PV, successfully managed with TIPS.