P1554 INCREASED PREVALENCE OF THROMBOEMBOLIC EVENTS IN PATIENTS WITH CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE I

Topic: 29. Iron metabolism, deficiency and overload

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Background: Congenital Dyserythropoietic Anemia Type I (CDA-I) is a rare disorder of erythropoiesis. All CDA-I patients are expected to suffer from iron overload and chronic hemolysis. Some patients with severe anemia may undergo splenectomy. Hemochromatosis, chronic hemolysis and splenectomy are all found to increase the risk for thromboembolism in thalassemic patients. Studies on thalassemic patients report an increased risk of thromboembolism, with an incidence ranging between 1.7 and 9.2% in thalassemic patients, compared to an annual incidence of 0.75-2.69 per 1000 in the general population. As a disease of ineffective erythropoiesis, CDA-I shares few pathophysiological characteristics with thalassemia. Yet, there are no reports on the prevalence of thromboembolism in CDA-I patients, with only one case of pulmonary embolism reported in an adult patient.

Aims: This study sought to evaluate prevalence of thromboembolic events in a group of CDA-I patients, and try to ascertain if this group of patients is at an increased risk of thromboembolism requiring a special follow-up.

Methods: a retrospective case-control study including 110 CDA-I patients (study group) and 326 age and sex-matched patients of the same ethnicity diagnosed with iron deficiency anemia (control group). Medical charts of all patients in both groups were retrieved from the hospital’s main archive and reviewed for the presence of venous and/or arterial thrombosis. Charts were screened to determine risk factors for thromboembolic events. Patients were risk-stratified using the 2010 Caprini Risk Assessment Model for thromboembolism.

Continuous variables were expressed as means and standard deviations (SD). The Student’s t-test was used to compare the distribution of continuous variables. Correlation between continuous variables were assessed using Pearson correlations, where r>0.5 and p value < 0.05 indicated significant correlation. To analyze the association between exposure variables and thromboembolic events, logistic regression models were used to estimate unadjusted and adjusted odds ratios (ORs) with their 95% confidence intervals (CI 95%). The chi-square test of independence was used to determine if there is a significant relationship between categorical variables.

Results: We identified three cases (2.7%) with thromboembolic events in the CDA group and one case (0.3%) in the control group. All of them were female patients. Caprini risk assessment for venous thromboembolism was low to moderate for the three CDA patients and high for the IDA patient. When compared to the control group, patients with CDA-I were nine times more likely to develop a thromboembolic event (OR 9.11, 95% CI=1.15-185.27, p=0.057). All three CDA patients had a history of remarkable hemolysis and iron overload. Two of them underwent splenectomy. Gender was strongly associated with increased risk of thromboembolism, as all cases with thromboembolism in this study were female patients. Caprini scores ranged from mild to moderate risk (score =2-4) for CDA patients and was high (score =7) for the IDA patient. Mean values of ferritin, iron, LDH and bilirubin were significantly higher (p<0.001), and hemoglobin level was significantly lower (p<0.001) in CDA group compared to control group. As expected, iron was significantly correlated with ferritin (r=0.617, p<0.001), and bilirubin was significantly correlated with LDH (r=0.631, p<0.001).

Summary/Conclusion: These findings show that CDA patients appear to be at increased risk for thromboembolic
events.

A high index of suspicion may help in early diagnosis and intervention in these patients.