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Case report

Clinical Presentation of a Patient with Congenital Polycythemia

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A B S T R A C T

The authors describe a 19 year old male with an isolated but absolute erythrocytosis with iron deficiency without evidence for polycythemia vera as well as another causes of erythrocytosis. The polycythemia was due to a recently described von Hippel-Lindau (VHL) mutation. By stopping iron therapy there was no more requirements for phlebotomy in this patient.

Key words: *congenital polycythemia, phlebotomy, iron deficiency, iron supplementation*

Introduction

Absolute polycythemia or erythrocytosis is a condition with increased red blood cell mass, and it can be either acquired or congenital. These hematological disorders are divided into primary where the erythropoietic compartment is intrinsically abnormal, and secondary where the erythropoietic compartment is normal but is responding to pathologic events outside the bone marrow, leading to an increased erythropoietin drive. Both primary and secondary polycythemias may be either congenital or acquired[1]. Polycythemia is rarely seen in children[2,3].

Case Report

A 12-year-old white boy was admitted in June 1997 for erythrocytosis, which was detected incidentally during an emergency room visit for a febrile illness and headache. Prior medical history was unremarkable. The patient had not traveled to high altitudes. There was no family history of hematologic disease. On examination the boy was in a very good general condition with facial and conjunctival plethora, all other clinical findings were within normal limits, blood pressure 110/80 mmHg. He was without pruritus, tinnitus, or vertigo. Hemogram: erythrocytes (E) $9.02x10^{12}$/L, hemoglobin (Hgb) 196 g/L, hematocrit (Hct) 0.65; MCV (mean corpuscular volume) 78 fL, MCHC (mean corpuscular hemoglobin concentration) 269 g/L. White blood cell and platelet counts were normal. Determination of the volume of erythrocytes by Cr-51 sodium chromate: 53.3 ml/kg (N ≤35 ml/kg). All causes of secondary polycythemia were eliminated by appropriate investigations. Vitamin B_{12} and folic acid in serum were within normal limits. Serum iron (Fe) level was low, 8.5 μmol/L (normal range: 9.5–29.8 μmol/L), UIBC 78.1 μmol/L, TIBC 86.6 μmol/L, ferritin 11 μg/L (N 15–332 μg/L). SaO_2 96% (pulse oxymeter). 2,3-diphosphoglycerate 5.7 mmol/l (N <5.5 mmol/L). Serum erythropoietin levels were from 23.2 to max. 72 mU/mL. Erythropoietin level after phlebotomy was 55 mU/mL. Tests of coagulation and platelet function were between referent values. Bone marrow cytology and histology were normal. Serum level of 2,3-diphosphoglycerate was normal, and so was hemoglobin electrophoresis. Abdominal ultrasound, CT scan of the abdomen and brain as well as isotopic renogram were within normal limits. Family members (parents and sister) had normal red blood counts.

As already recently reported, DNA analysis showed homozygosity for 571C→G (H191D) von Hippel-Lindau tumor-suppressor gene (VHL) mutation, the first example of homozygous VHL germline mutation causing polycythemia other than the VHL 598C→T mutation[4].

During clinical observation the patient had no significant medical problem. He reported headaches and concentration difficulties when his hematocrit was high (Hct≥ 0.60, Hgb≥200 g/L, E≥$9.0x10^{12}$/L), and these symptoms were immediately relieved by isovolemic phlebotomy.

Because of the slightly decreased values of serum iron and ferritin the patient was administered iron supple-

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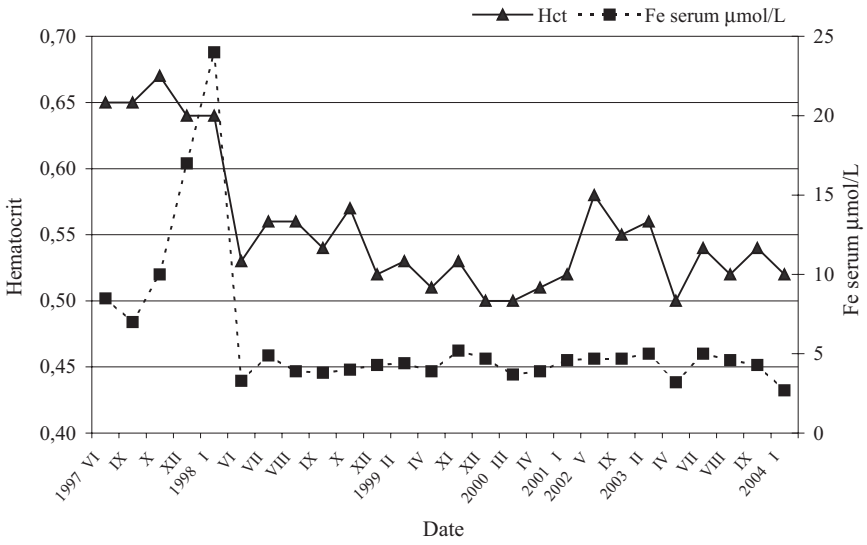

Fig. 1. Correlation between hematocrit and serum iron in our patient. Phlebotomy is pointed with ↑.

mentation therapy and aspirin. He had normal gastrointestinal iron absorption as well as intestinal permeability testing.

During the next 12 months phlebotomy was required on seven occasions. On iron supplementation, his serum iron concentration increased, ranging between 6.1 and 17 μmol/L.

At one year, his hematology findings were found to have considerably improved. Hct levels were 0.51–0.56 and Hgb concentration 144–159 g/L, while serum iron concentration decreased to 2.3–4.9 μmol/L and ferritin to 1.8–5 ng/L (Figure 1). The reticulocytes count in that time was up to 0,020. The patient admitted he had discontinued iron supplementation and made decision to reduce intake of iron by food, so he started diet without animal protein and iron rich vegetables. We realized soon that hematocrit would not reach high values to require phlebotomy anymore. Over the next four years, the patient did not take iron supplementation. After withdrawal of iron supplementation, his Hct value was 0.58 on a single occasion. The high values of Hct (0.60– 0.68) that previously were regularly recorded have not been observed since; the boy is free from any medical problem and has not required new phlebotomy.

When last seen in May 2004, his Hct was 0.52 and he had no complaints, and still without impact on neurological or cognitive function. He is now 19 years old and a successful university student and active tennis player. His height is 180 cm, weight 69 kg, blood pressure 95/70 mmHg, c/p 76/min.

Conclusion

We present the patient with congenital polycythemia in whom the frequency of phlebotomy has been kept lower by obviating iron supplementation than when iron was administered. We find that Swedish authors had a similar outcome with patients with polycythemia vera[5]. Our outcome suggests that if iron supplementation is avoided, the frequency of phlebotomy can be kept lower than when iron is administered in patients with congenital polycythemia.

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KLINIČKI PRIKAZ PACIJENTA S KONGENITALNOM POLICITEMIJOM

S A Ž E T A K

Autori prikazuju devetnaestogodišnjeg bolesnika, praćenog unazad sedam godina, sa izoliranom apsolutnom eritrocitozom i sideropenijom u kojeg je isključena policitemija vera kao i ostali uzroci eritrocitoze. Policitemija je posljedica nedavno opisane von Hippel-Lindau (VHL) mutacije. Prestankom terapije željezom prestala je potreba za flebotomijom kod ovog bolesnika.