

## Clinical Case of the Gaisböck Syndrome

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### Abstract

This report describes the case of a 43-year-old Caucasian man with a 6-year history of hematological abnormalities associated with arterial hypertension, primary hypercholesterolemia, and obesity. The patient presented with facial redness, low erythropoietin level, negative tests for JAK2V617F, an exon 12JAK2 mutations, and mutations associated with congenital erythrocytosis; no signs of splenomegaly were found. Only regularly performed phlebotomy, but not getting blood pressure under control made it possible to lower hematocrit and hemoglobin levels.

**Keywords:** Gaisböck syndrome; erythrocytosis; arterial hypertension; case report.

### Introduction

In 1905, Felix Gaisböck, MD (from Innsbruck University, Tyrol, Austria) first described a syndrome observed by him in hypertensive male patients having high hematocrit levels [1]. Working in the Austrian highlands for many years, he focused his scientific interests on the cardiovascular pathophysiology of living under such conditions and on sports medicine. Gaisböck syndrome is associated with high cardiovascular risk and, not rarely, with obesity, lipid profile abnormalities, as well as with high renin and uric acid blood levels; it is more common in men, especially in smokers [2]. The presented report describes a young man with a full-blooded appearance, arterial hypertension found on several medical occasions, and incidental erythrocytosis observed over several years. No factors that could be considered the causes of primary or secondary erythrocytosis were present, supporting the diagnosis of Gaisböck syndrome.

### Case Report

A 43-year-old Caucasian man was referred for consultation with a hematologist because of a 6-year history of blood test abnormalities (high RBC counts, increased hemoglobin, and hematocrit values). His past medical history was notable for arterial hypertension, primary hypercholesterolemia, and obesity. Occasionally, the blood pressure reached 170/105 mmHg. The patient was taking telmisartan 40 mg/day, amlo-

dipine 2.5 mg/day, nebivolol 2.5 mg/day, ezetimibe 10 mg/day, pitavastatin 2 mg/day, and clopidogrel 75 mg/day. A past smoker, he quit smoking 4 years ago; no episodes of sleep apnea were observed. The patient had no history of arterial or venous thrombosis.

On physical examination, the patient's temperature was 36.6°C, blood pressure 130/76 mmHg, regular pulse rate 74, respiratory rate 14 breaths/minute; his body mass index (BMI) was 31.5. In the past, BMI was as high as 35.6. The patient presented facial redness increasing in the recumbent position and palmar erythema; his tongue was dark red. No lymphadenopathy was present. The liver and spleen were of normal size.

The time changes of the red blood test parameters (hemoglobin, Hb; hematocrit, Ht; RBC count and indices) are given in (Table 1).

**Table 1:** Dynamics of red blood parameters.

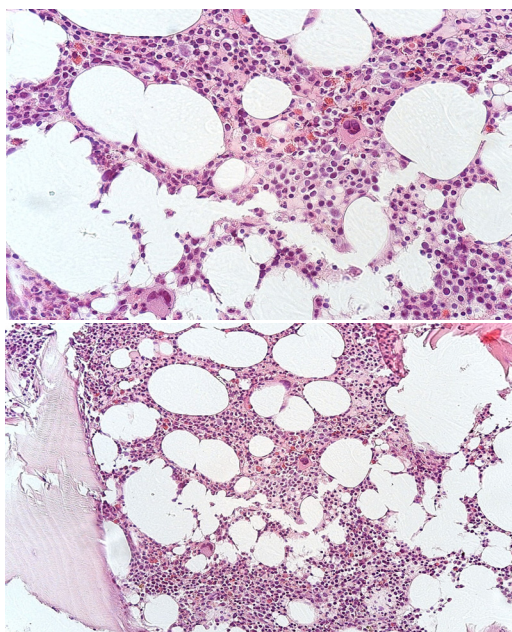
Parameters	Hb (g/dL)	RBC (x10 <sup>12</sup> /L)	MCV (fl)	MCH (pg)	Ht (%)
Normal Ranges	13.5-17.2	4.3-5.75	80.0-99.0	27.0-33.5	39.5-49.0
In debut	17.4	5.43	94.9	32.1	51.5
At present	16.1	5.11	95.9	31.5	49.0

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Leukocyte and platelet counts were within normal limits. Endogenous erythropoietin (EPO) level was low (1.62 mU/mL, normal range 4.3-21.0); pO<sub>2</sub> was normal. Blood uric acid elevated to 77.5 mg/L (normal range 35-60.5). Other biochemical blood parameters were within normal limits, including holotranscobalamin level – 129 pmol/L (15-147), folic acid 9.3 ng/mL (3.1-20.5), and ferritin 115 ng/mL (20-250). Abdominal and chest CT scans were normal with no evidence of splenomegaly and lung pathology.

*JAK2V617F* and an exon 12*JAK2* mutations, which are the diagnostic criteria for polycythemia vera, were not found. No mutations in the erythropoietin receptor (*EPOR*) gene (coding exons 1-8), the *VHL* gene (exons 1-3), and the *PHD* gene (exons 2-3), as well as *PDH2/EGLN1* mutation, were found, thereby excluding other possible causes of congenital erythrocytosis.

Microscopic evaluation of the bone marrow biopsy showed slightly increased erythropoietic proliferation without morphological evidence of any myeloproliferative neoplastic disorder, like polycythemia vera (**Figure 1**).



**Figure 1:** (A, B). Normocellular for age bone marrow (approximately 50%) with relative erythroid predominance. Maturing and mature cells of the granulocytic germ are present in sufficient numbers. Megakaryocytes mainly presented by cells with nuclear hyposegmentation are found in low numbers.

Medical correction of the blood pressure and BMI reduction from 36.5 to 31.5 did not result in stable normalization of hematocrit. Therapeutic phlebotomy was regularly performed during the last year to normalize hematocrit, hemoglobin level, and RBC count.

## Discussion

Erythrocytosis, a condition characterized by increased red blood cell mass, can be absolute or relative [3]. Absolute erythrocytosis is either primary or secondary. Erythrocytosis can develop for multiple reasons. Primary erythrocytosis results from a genetically determined bone marrow defect leading to increased production of red blood cells. In such cases,

the EPO levels are below the normal range. Primary erythrocytosis, like polycythemia vera, can be attributed to a *JAK2* mutation resulting in dysregulated production of cells of all three hematopoietic germs.

Secondary erythrocytosis arises when RBC production is driven by EPO [4]. In patients with secondary erythrocytosis, EPO levels are raised or inappropriately normal for the raised hemoglobin. Erythrocytosis can be congenital or acquired, depending on whether it is present from birth or develops later in life. In congenital cases, erythrocytosis may not often be tested for or detected at birth, but symptoms are usually present at a young age [5]. Relative erythrocytosis results from any condition that reduces plasma volume, such as vomiting, diarrhea, fever, or diuretic use. But still there remains a group of patients with an unidentifiable cause of erythrocytosis; in such cases, erythrocytosis is categorized as idiopathic [4].

Lacking the technology with radioactive chromium, we were unable to measure the red cell mass, but the overproduction of erythroid cells in the bone marrow confirms that it is an absolute, not relative erythrocytosis. The endogenous EPO level was below normal values, indicating its primary nature. Bone marrow showed erythroid hyperplasia with generally normal morphology of other elements in contrast to trilineage hyperplasia and pleomorphic megakaryocytes typical for polycythemia vera; in addition, no splenomegaly was found on the CT scans plus the *JAK2* mutation test was negative, all these data taken together contradicting the diagnosis of a myeloproliferative disease. Based on the absence of mutations in the *EPOR*, *PHD*, or *VHL* genes, we can also exclude the diagnosis of congenital erythrocytosis including the so-called Chuvash polycythemia, which may be in the population of Russia. Thus, based on the presence of absolute erythrocytosis in combination with elevated hematocrit, arterial hypertension, and such factors as primary hypercholesterolemia and obesity, we could diagnose the Gaisböck syndrome in our patient. The phenomenon described by F. Gaisböck back in 1905 is still relevant, but a link between RBC overproduction and high blood pressure remains poorly understood.

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