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Focal Segmental Glomerulosclerosis and Primary Polycythemia: Report of Two Cases

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Abstract:

Primary polycythemia, also known as polycythemia vera or Vaquez disease, is a myeloproliferative disorder characterized by excessive red blood cell production due to a mutation in the JAK2 protein, leading to uncontrolled proliferation of erythroid progenitor cells . Renal involvement in polycythemia vera is uncommon but can manifest as various glomerular diseases, including focal segmental glomerulosclerosis (FSGS) . We report two cases of patients with primary polycythemia and FSGS, highlighting the potential association between these two conditions and the importance of treating the underlying myeloproliferative disorder.

Case 1: A 19-year-old female patient, followed since the age of 15 years for primary FSGS diagnosed on renal biopsy performed for impure nephrotic syndrome with hematuria. Four years after complete remission, the patient presented with a relapse associated with polycythemia and thrombocytosis on complete blood count. Secondary causes of polycythemia were excluded. Bone marrow biopsy revealed findings consistent with polycythemia vera, despite negative JAK2 mutation testing. The hematology team proposed phlebotomy and myelosuppressive oral therapy. One month later, proteinuria regressed from 15 g/24h to 4.7 g/24h.

Case 2: A 45-year-old male patient, with a history of polycythemia vera diagnosed two years earlier, presented with nephrotic syndrome. Renal biopsy revealed focal segmental glomerulosclerosis. The patient was treated with phlebotomy and myelosuppressive therapy, with improvement in proteinuria.

Discussion: The association between FSGS and polycythemia vera is rare but has been reported in the literature. The pathophysiology is thought to involve hyperviscosity, increased glomerular capillary pressure, and possible direct effects of JAK2 mutations on podocytes. Treatment of the underlying myeloproliferative disorder can lead to improvement of proteinuria, as observed in our patients.

Conclusion: Primary polycythemia should be considered in patients with FSGS and unexplained polycythemia. While polycythemia vera is not curable, early detection and appropriate treatment can reduce the risk of vascular complications and may improve renal outcomes.

Key words: focal segmental glomerulosclerosis; polycythemia vera; vaquez disease; jak2 mutation; nephrotic syndrome

Introduction:

Polycythemia vera, also known as Vaquez disease, is a myeloproliferative disorder characterized by excessive production of red blood cells due to a mutation in the JAK2 protein, leading to uncontrolled proliferation of erythroid progenitor cells [1]. It belongs to a group of diseases known as myeloproliferative syndromes. The disease is caused by a mutation in the Janus kinase 2 (JAK2) gene, most commonly the V617F mutation, which leads to constitutive activation of the JAK-STAT signaling pathway and uncontrolled proliferation of hematopoietic cells [1].

Polycythemia vera is a rare disease, with an estimated incidence of 1 to 3 per 100,000 person-years [1]. It typically presents in the sixth decade of life, with a slight male predominance [1]. Clinical manifestations include headache, dizziness, pruritus, splenomegaly, and an increased risk of thrombotic and hemorrhagic complications [1].

Renal involvement in polycythemia vera is uncommon but can manifest as various glomerular diseases, including focal segmental glomerulosclerosis (FSGS), membranous nephropathy, and minimal change disease [2, 3]. The pathophysiology is thought to involve hyperviscosity, increased glomerular capillary pressure, and possible direct effects of JAK2 mutations on podocytes [3, 4].

FSGS is a glomerular disease characterized by segmental sclerosis of the glomerular tuft and is a common cause of nephrotic syndrome in adults [5, 6]. It can be primary (idiopathic) or secondary to various causes, including viral infections, drugs, obesity, and hematological disorders [5, 6]. The association between FSGS and polycythemia vera is rare but has been reported in the literature [2, 3, 7, 8].

We report two cases of patients with primary polycythemia and FSGS, highlighting the potential association between these two conditions and the importance of treating the underlying myeloproliferative disorder.

Case Reports:

Case 1

A 19-year-old female patient, followed since the age of 15 years for primary FSGS diagnosed on renal biopsy performed for impure nephrotic syndrome with hematuria, presented with a relapse four years after complete remission. The relapse was associated with polycythemia and thrombocytosis on complete blood count. Secondary causes of polycythemia were excluded. Bone marrow biopsy revealed findings consistent with polycythemia vera, despite negative JAK2 mutation testing. The hematology team proposed phlebotomy and myelosuppressive oral therapy. One month later, proteinuria regressed from 15 g/24h to 4.7 g/24h.

Case 2

A 45-year-old male patient, with a history of polycythemia vera diagnosed two years earlier, presented with nephrotic syndrome. Renal biopsy revealed focal segmental glomerulosclerosis. The patient was treated with phlebotomy and myelosuppressive therapy, with improvement in proteinuria.

Discussion

Epidemiology of Polycythemia Vera and FSGS:

Polycythemia vera is a rare myeloproliferative disorder with an estimated incidence of 1 to 3 per 100,000 person-years [1]. The prevalence of FSGS in the general population is estimated at 2 to 7 per 100,000 [5]. The association between these two conditions is rare, with only a few cases reported in the literature [2, 3, 7, 8]

A literature review identified only a few cases of FSGS associated with polycythemia vera. The majority of these cases were reported in adults, with a slight male predominance [2, 3, 7] . Our cases add to the growing body of evidence supporting an association between these two conditions.

Pathophysiological Mechanisms

The pathophysiology of renal involvement in polycythemia vera is not fully understood but is thought to involve several mechanisms [2-4]:

- **Hyperviscosity:** The increased red blood cell mass leads to hyperviscosity, which can cause renal microvascular injury and glomerular hypertension [4].
- **Increased glomerular capillary pressure:** The hypervolemia and increased cardiac output associated with polycythemia vera can lead to increased glomerular capillary pressure, contributing to glomerular injury [2, 3].
- **Direct effects of JAK2 mutations:** JAK2 mutations have been detected in podocytes and other renal cells in some patients with myeloproliferative disorders, suggesting a potential direct role of the mutant JAK2 protein in glomerular injury [3, 4].
- **Secondary FSGS:** FSGS in the setting of polycythemia vera may be secondary to hyperfiltration and glomerular hypertension, rather than a primary podocytopathy [2, 3].
- In our cases, the regression of proteinuria following treatment of the underlying polycythemia suggests that the FSGS was secondary to the myeloproliferative disorder.

Clinical Presentation

The clinical presentation of FSGS in patients with polycythemia vera is similar to that of primary FSGS, with nephrotic syndrome, proteinuria, edema, and, in some cases, hematuria [2, 3]. However, patients with polycythemia vera may also have symptoms related to the underlying myeloproliferative disorder, such as headache, dizziness, pruritus, and splenomegaly [1].

In our cases, the patients presented with nephrotic syndrome and proteinuria, consistent with the literature. The association with polycythemia was detected either at the time of FSGS relapse or during the initial presentation.

Diagnosis

The diagnosis of polycythemia vera is based on the World Health Organization criteria, which include elevated hemoglobin or hematocrit, the presence of JAK2 mutations, and bone marrow biopsy findings [1]. In our cases, one patient had negative JAK2 mutation testing but bone marrow biopsy findings consistent with polycythemia vera.

The diagnosis of FSGS requires renal biopsy, which shows segmental sclerosis of the glomerular tuft [5, 6]. The presence of FSGS in a patient with polycythemia vera should prompt consideration of the secondary nature of the glomerular disease [2, 3].

Treatment

The treatment of FSGS in patients with polycythemia vera should address both the underlying myeloproliferative disorder and the glomerular disease [2, 3, 7].

- **Treatment of polycythemia vera:** Phlebotomy is the mainstay of treatment for polycythemia vera, aiming to maintain hematocrit below 45% [1]. Myelosuppressive therapy, such as hydroxyurea, is used in patients with high-risk disease or those who require frequent phlebotomy [1]. The JAK1/JAK2 inhibitor ruxolitinib is used in patients who are intolerant or resistant to hydroxyurea [1].
- **Treatment of FSGS:** The treatment of secondary FSGS should focus on the underlying cause [6,7]. In patients with polycythemia vera, controlling the myeloproliferative disorder can lead to improvement of proteinuria [2,3]. Corticosteroids and other immunosuppressive agents are generally not recommended for secondary FSGS [6,7].

In our cases, treatment of polycythemia vera with phlebotomy and myelosuppressive therapy led to regression of proteinuria, supporting the secondary nature of the FSGS.

Prognosis:

The prognosis of FSGS in patients with polycythemia vera is generally good if the underlying myeloproliferative disorder is adequately controlled [2,3]. However, patients with polycythemia vera are at increased risk of thrombotic and hemorrhagic complications, which can affect overall survival [1].

While polycythemia vera is not curable, early detection and appropriate treatment can reduce the risk of vascular complications and may improve renal outcomes. In our cases, the patients responded well to treatment of polycythemia vera, with improvement in proteinuria.

Conclusion:

Focal segmental glomerulosclerosis can be associated with primary polycythemia (Vaquez disease). The pathophysiology is thought to involve hyperviscosity, increased glomerular capillary pressure, and possible direct effects of JAK2 mutations on podocytes. Treatment of the underlying myeloproliferative disorder can lead to improvement of proteinuria, as observed in our two cases. While polycythemia vera is not curable, early detection and appropriate treatment can reduce the risk of vascular complications and may improve renal outcomes. Clinicians should consider polycythemia vera in patients with FSGS and unexplained polycythemia.

References:

1. Tefferi, A. M. Vannucchi, and T. Barbui, (2024) "Essential thrombocythemia: 2024 update on diagnosis, risk stratification, and management," (in eng), Am J Hematol, vol. 99, no. 4, pp. 697-718. *10.1002/ajh.27216*
   View at Google Scholar | ***View at Publisher***
2. S. Okuyama et al.,(2007) "Focal segmental glomerulosclerosis associated with polycythemia vera: report of a case and review of the literature," (in eng), Clin Nephrol, vol. 68, no. 6, pp. 412-415, *https://doi.org/10.5414/cnp68412*
   View at Google Scholar | ***View at Publisher***
3. M. Iyoda et al., (2005) "[Focal segmental glomerulosclerosis in a patient with polycythemia vera]," (in jpn), Nihon Jinzo Gakkai Shi, vol. 47, no. 7, pp. 828-833,
   View at Google Scholar | ***View at Publisher***
4. James, (2008) "The JAK2V617F mutation in polycythemia vera and other myeloproliferative disorders: one mutation for three diseases?," (in eng), Hematology Am Soc Hematol Educ Program, pp. 69-75, 2008, *https://doi.org/10.1182/asheducation-2008.1.69*
   View at Google Scholar | ***View at Publisher***
5. S. De Vriese, S. Sethi, K. A. Nath, R. J. Glassock, and F. C. Fervenza, (2018) "Differentiating Primary, Genetic, and Secondary FSGS in Adults: A Clinicopathologic Approach," (in eng), J Am Soc Nephrol, vol. 29, no. 3, pp. 759-774, https://doi.org/10.1681/ASN.2017090958

View at Google Scholar | ***View at Publisher***

6. V. D. D'Agati, F. J. Kaskel, and R. J. Falk, (2011) "Focal segmental glomerulosclerosis," (in eng), N Engl J Med, vol. 365, no. 25, pp. 2398-2411, *http://doi.org/10.1056/NEJMra1106556*
View at Google Scholar | ***View at Publisher***
7. M. C. Abdulla, S. Sathyan, Z. Shemin, and N. Mampilly, (2018) "Essential Thrombocythemia Presenting as Paraneoplastic Glomerulonephritis," (in eng), Indian J Nephrol, vol. 28, no. 3, p. 249, *https://doi.org/10.4103/ijn.IJN_335_17*
View at Google Scholar | ***View at Publisher***
8. Y. Song, Y. Zhen, H. Ma, B. Feng, and Y. Xie, (2025) "Case Report of Hepatic Sinusoidal Obstruction Syndrome Complicated with Myeloproliferative Neoplasm and Focal Segmental Glomerulosclerosis," (in eng), Case Rep Gastroenterol, vol. 19, no. 1, pp. 502-508, https://doi.org/10.1159/000546801
View at Google Scholar | ***View at Publisher***