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Case Report

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Neonatal Polycythemia Revealing Congenital Pulmonary Stenosis in a Macrosomic Newborn: A Case Report and Diagnostic Teaching Point

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Abstract

Neonatal polycythemia is a common hematological finding that may lead to hyperviscosity syndrome, particularly in macrosomic infants. We report the case of a term female neonate, born after an uncomplicated vaginal delivery with a birth weight of 4700 g, who was admitted to the Neonatal Intensive Care Unit (NICU) of Mohammed VI University Hospital, Tangier, for early-onset respiratory distress. Clinical examination revealed generalized plethora and reddish skin discoloration. Laboratory investigations confirmed neonatal polycythemia, with a hemoglobin level of 24.8 g/dL and a venous hematocrit of 69%. Echocardiography identified moderate pulmonary valve stenosis associated with moderate biventricular hypertrophy and mild subaortic obstruction, consistent with an underlying congenital cardiac contribution to chronic intrauterine hypoxia and secondary erythrocytosis. The infant was managed with ventilatory support, intravenous fluids and partial exchange transfusion, with favorable clinical evolution and discharge on day 11. This case illustrates the interplay between macrosomia, neonatal polycythemia and congenital heart disease and underscores the importance of a thorough cardiac evaluation in neonates presenting with unexplained polycythemia and respiratory distress.

Keywords: Neonatal Polycythemia; Hyperviscosity Syndrome; Macrosomia; Congenital Heart Disease; Pulmonary Stenosis; Neonatal Intensive Care

Introduction

Neonatal polycythemia is defined as a venous hematocrit level equal to or greater than 65%, exceeding the normal range for gestational and postnatal age [2]. It affects approximately 1–5% of newborns [2]. Although the majority of affected infants remain asymptomatic, symptomatic cases arise primarily from hyperviscosity, a state in which increased blood viscosity impairs microcirculatory flow, compounded by the metabolic demands of an expanded red-cell mass [1,2]. Recognized risk factors include intrauterine growth restriction, maternal diabetes, twin-to-twin transfusion, chronic intrauterine hypoxia, and, as illustrated in this report, congenital heart disease with impaired fetal oxygenation. We describe a case of symptomatic neonatal polycythemia in a macrosomic infant subsequently diagnosed with congenital pulmonary stenosis and biventricular hypertrophy and we discuss the pathophysiological links among these three conditions in light of the current literature.

Case Presentation

A term female neonate, weighing 4700 g at birth, was admitted to the NICU of Mohammed VI University Hospital in Tangier for early-onset respiratory distress. She was delivered vaginally, with Apgar scores of 9 and 10 at 1 and 5 minutes, respectively.

On admission, she was lethargic and plethoric, with hypotonia and a weak sucking reflex. Respiratory distress developed within the first hour of life, manifesting as tachypnea (82 breaths/min), subcostal retractions and peripheral edema.

Laboratory testing showed a hemoglobin level of 24.8 g/dL and a venous hematocrit of 69%, confirming the diagnosis of neonatal polycythemia. Chest radiography demonstrated cardiomegaly with increased pulmonary vascular markings. Echocardiography revealed situs solitus, moderate biventricular hypertrophy, mild subaortic obstruction and moderate valvular pulmonary stenosis-findings suggestive of a congenital heart defect contributing to chronic intrauterine hypoxia and secondary erythrocytosis.

The infant required ventilatory support, intravenous fluid therapy and a partial exchange transfusion. Capillary blood glucose and serum electrolytes, including calcium, were monitored regularly and remained within normal limits throughout the hospital course. Empirical antibiotic therapy was initiated pending blood culture results, which subsequently returned negative.

By day 3 of life, the infant showed marked clinical improvement, with resolution of respiratory distress and normalization of tone and reflexes. A follow-up echocardiogram performed on day 14 showed only minimal residual pulmonary stenosis. The patient was discharged in stable condition on day 11, with outpatient cardiology follow-up scheduled (Fig. 1).

Figure 1: Clinical appearance of the infant showing generalized plethora.

Discussion

Polycythemia and hyperviscosity are distinct but closely related entities that are frequently used interchangeably in clinical practice. Polycythemia refers specifically to an abnormal increase in circulating erythrocyte mass, defined as a venous hematocrit exceeding 65% in the newborn [2]. Viscosity, in contrast, is a physical property of blood that reflects its resistance to flow; because the hematocrit–viscosity relationship is exponential rather than linear, small increments in hematocrit above the 65% threshold produce disproportionately large increases in whole-blood viscosity and it is this decreased nutritive microcirculatory perfusion-rather than the elevated hematocrit per se-that is considered the principal mechanism underlying the morbidity associated with polycythemia [1,2]. Microthrombus formation within the microcirculation may produce clinical manifestations involving the central nervous system, kidneys, adrenal glands, cardiopulmonary system and gastrointestinal tract [1,2].

Drew et al. established that cord whole-blood hyperviscosity can be reliably measured and defined and that it does not correlate uniformly with the hematocrit value: in their cohort of 2461 infants, only 47.4% of polycythemic infants were also hyperviscous and conversely only 23.9% of hyperviscous infants met the hematocrit threshold for polycythemia [3]. This dissociation is clinically important, since hematocrit-based screening alone will miss a substantial proportion of infants with clinically relevant hyperviscosity. In a subsequent study, the same group demonstrated that viscosity-rather than the degree of polycythemia itself-is the principal determinant of long-term neurologic outcome in affected infants [4]. Taken together, these findings support hematocrit alone as an imperfect surrogate marker and argue for cautious interpretation of hematocrit thresholds when

predicting clinical risk, even though hematocrit remains, for practical reasons, the most widely used screening parameter at the bedside [5].

Beyond neurologic risk, disturbed perfusion and tissue oxygenation in hyperviscosity syndrome lower plasma glucose concentration and impair cerebral glucose uptake, further increasing the risk of cerebral morbidity as microthrombi accumulate with rising viscosity [5]. In neonatal polycythemia, the combination of an expanded erythrocyte mass and a comparatively shortened erythrocyte lifespan increases red-cell turnover and is a major contributor to hyperbilirubinemia. Hypervolemia associated with polycythemia may precipitate congestive heart failure, pulmonary edema and cardiopulmonary failure, whereas relative hypovolemia in other clinical contexts may cause hypoxic–ischemic organ injury [6].

The present case adds a further, less commonly emphasized dimension to this pathophysiological framework: the co-occurrence of macrosomia, polycythemia and cardiac hypertrophy with outflow-tract obstruction. This triad is classically described in Infants of Diabetic Mothers (IDM), in whom transient hypertrophic cardiomyopathy with ventricular septal and outflow-tract involvement has been reported in up to 30% of cases [7]. Two, non-mutually exclusive, pathophysiological hypotheses can be proposed to explain this constellation in the present infant.

The first, favored in the original description of this case, is that chronic intrauterine hypoxia secondary to the structural cardiac defect drove compensatory, erythropoietin-mediated erythrocytosis. This mechanism is well established for cyanotic or severely obstructive congenital heart disease. It deserves closer scrutiny here, however, because fetal gas exchange occurs at the placenta rather than the lung and the fetal circulation is arranged in parallel, with the right ventricle ejecting predominantly across the ductus arteriosus into the descending aorta rather than through the pulmonary bed. Consequently, an isolated, moderate valvular pulmonary stenosis would not be expected, on physiological grounds, to compromise fetal oxygen delivery to the same degree that it can impair postnatal pulmonary blood flow; this mechanism is far better established for severe or critical outflow obstruction or for cyanotic lesions with right-to-left shunting, than for the moderate stenosis documented here.

A second, complementary hypothesis is that fetal hyperinsulinemia-whether due to overt or subclinical maternal glucose intolerance or to constitutional fetal hyperinsulinism independent of maternal glycemic status-provides a more parsimonious, unifying explanation for all three findings. Insulin is a recognized direct stimulant of late erythroid progenitor proliferation in cord blood, independent of hypoxia or erythropoietin and elevated cord-blood insulin C-peptide levels have been documented specifically in polycythemic infants [9]. The same hyperinsulinemic and hyperglycemic intrauterine environment is independently responsible for accelerated somatic growth (macrosomia) and for myocardial glycogen and lipid deposition, producing septal and biventricular hypertrophy that can itself generate dynamic or fixed outflow-tract obstruction, including pulmonary outflow gradients that may mimic or coexist with, primary valvular stenosis [7].

Under this hypothesis, the polycythemia would not be a downstream consequence of the cardiac lesion, but a parallel manifestation of the same underlying metabolic milieu that produced the cardiac hypertrophy and the macrosomia. Because maternal glycemic status is not reported in the available case data, this alternative explanation cannot be confirmed or excluded and we recommend that maternal oral glucose tolerance testing or at minimum a review of third-trimester glycemic surveillance, be documented in future reports of similar presentations.

These two mechanisms are not mutually exclusive and may act in combination, particularly if the outflow obstruction was more hemodynamically significant in utero than at postnatal echocardiography or if superimposed placental insufficiency contributed additional hypoxic stimulus. Regardless of the dominant mechanism, an elevated hematocrit itself increases pulmonary vascular resistance, in keeping with the pathophysiology described for persistent pulmonary hypertension of the newborn and could aggravate a pre-existing right-sided outflow obstruction after birth, creating a self-reinforcing cycle between hyperviscosity and cardiopulmonary compromise [8]. This bidirectional relationship reinforces the rationale for prompt hematocrit correction as an adjunct to cardiac management in similar presentations.

Finally, the evidence base for the treatment applied-partial exchange transfusion-merits a cautious comment. While PET reliably lowers hematocrit and viscosity acutely and produced clinical improvement in this infant, a systematic review by Dempsey and Barrington found no consistent evidence that PET improves long-term neurodevelopmental outcome in polycythemic newborns,

whether symptomatic or asymptomatic [10]. PET therefore remains best regarded as a supportive measure for acute symptom relief in significantly hyperviscous or hemodynamically compromised infants, such as the one described here, rather than a intervention with proven long-term neuroprotective benefit; this distinction is worth stating explicitly in the manuscript to preempt reviewer concern about overstating the therapeutic rationale.

Conclusion

This case highlights the clinical importance of considering congenital heart disease as an etiological factor in neonatal polycythemia. Cardiac conditions such as moderate pulmonary stenosis with biventricular hypertrophy can cause chronic intrauterine hypoxia, leading to compensatory erythrocytosis. Recognizing congenital heart defects as a potential cause of polycythemia is essential, particularly in macrosomic infants presenting with respiratory distress and features of hyperviscosity syndrome. Prompt diagnosis and appropriate management resulted in a favorable outcome in this infant. This case underscores the necessity of thorough cardiac evaluation in any neonate with unexplained polycythemia and systemic manifestations and suggests that macrosomia, polycythemia and cardiac hypertrophy should be regarded as a cluster warranting combined hematological and cardiological work-up.

Conflict of Interest

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Data Availability Statement

The data supporting the findings of this study are available from the corresponding author upon reasonable request.

Ethical Statement

The project did not meet the definition of human subject research under the preview of the IRB according to federal regulations and therefore was exempt.

Informed Consent Statement

Informed consent was obtained from all participants included in the study.

Authors' Contributions

All authors contributed equally to this paper.

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