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CASE REPORT PEER REVIEWED | OPEN ACCESS

Spontaneous hemorrhage of peripheral neurofibroma in pregnancy: A case report

Kira A Bromwich, Margaret Rush, Matthew Janssen, Adi Hirshberg

ABSTRACT

Introduction: Neurofibromas may grow in pregnancy due to hormonal sensitivity. This report describes a case of spontaneous hemorrhage from a neurofibroma in pregnancy.

Case Report: A 27-year-old female with neurofibromatosis (NF1) presented at 30 weeks gestation with shortness of breath. Spontaneous hemorrhage was noted from her left leg neurofibroma. The patient underwent preprocedural fetal surveillance prior to proceeding with surgical debridement. Persistent fetal bradycardia was noted in setting of maternal hypotension with ongoing bleeding. An emergent cesarean delivery was performed and required debridement of the left thigh neurofibroma by the general surgical team.

Conclusion: Spontaneous hemorrhage of neurofibromas is a rare, but reported complication. Neurofibromas may increase in size in pregnancy; however, spontaneous hemorrhage has not previously been documented to occur during pregnancy.

Keywords: Hemorrhage, Neurofibroma, Neurofibromatosis, NF1, Pregnancy, Spontaneous

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INTRODUCTION

Neurofibromatosis 1 (NF1) is a rare autosomal-dominant disorder resulting in increased frequency of benign and malignant tumors throughout a patient's lifespan. The disorder is due to a mutation in the NF1 gene (chromosome 17q11.2), resulting in a loss of function of neurofibromin which can cause a wide spectrum of clinical findings and characteristic NF1-associated tumors [1]. The disease has variable expressivity, with specific manifestations of the disorder varying among affected individuals, even within the same family. Hallmarks of this disease include café-au-lait macules, axillary or inguinal freckling (Crowe sign), hamartomas of the iris (Lisch nodules), and neurofibromas [2].

Plexiform neurofibromas represent a major cause of morbidity and disfigurement in individuals with NF1, and even asymptomatic plexiform neurofibromas are associated with increased mortality [1].

In this case report, we discuss a patient presenting at 30 weeks gestation with severe hypotension and anemia secondary to spontaneously bleeding neurofibroma, ultimately requiring urgent delivery, surgical team intervention to stop the bleeding, and intensive care unit (ICU) admission.

CASE REPORT

We present a case of a 27-year-old female, with obstetric history notable for one prior preterm vaginal delivery at 33 weeks due to severe preeclampsia. She had a medical history of NF1 diagnosed in childhood. Her only ongoing symptom monitored during the pregnancy was left leg elephantiasis related to a large plexiform neurofibroma in the left thigh, which had been present since childhood. She had no other chronic symptomatic neurofibromas or sequelae from her NF1. Prenatal genetic testing of the fetus was offered to the patient, who declined.

A magnetic resonance imaging (MRI) of her spine, performed during pregnancy to evaluate candidacy for neuraxial anesthesia, visualized the large left lower extremity plexiform neurofibroma early in pregnancy and characterized it as involving the left hip and lower extremity gluteal region, with associated dilated and tortuous veins in the lumbar subcutaneous fat.

Her pregnancy was uncomplicated until 30 weeks, when she presented to the emergency department with sudden onset shortness of breath and subjective fever. She had no obstetric complaints, though notably she had presented to an outside hospital emergency department 24 hours prior for preterm contractions and was discharged home in good condition. Vital signs were notable for fever of 100.8°F, tachycardia to 130 bpm, normotensive blood pressure of 117/67 mmHg, tachypnea to 20 bpm, and normal oxygen saturation of 100% on room air. Laboratory results revealed a leukocytosis to 17.8 THO/uL, hemoglobin of 7.5 g/dL (notably changed from 9.6 g/dL two days prior), and a lactate of 4.7 mmol/L. Further workup for an infectious source revealed negative COVID-19, respiratory syncytial virus (RSV), and influenza results. A pulmonary embolism was ruled out via computed tomography-angiography (CTA). Blood cultures were performed and she was started on empiric broad-spectrum antibiotics with cefepime and vancomycin due to the described clinical picture and concern for systemic infection. At this time, she was noted to be bleeding from her left leg neurofibroma, and two hemostatic stitches were placed at the skin surface. On further examination, a 5 inch area of warmth, erythema, and induration of the left lateral mid-thigh was noted within skin folds of lymphedema and fibromatous lesions concerning for continued hemorrhage into the neurofibroma. A CT of the abdomen and pelvis revealed interval growth of patient's known neurofibroma, and a new hematoma extending to the lateral most aspect of the left thigh was noted, explaining her significant anemia (Figure 1). The constellation of these vital signs, laboratory, and exam findings were consistent with active bleeding within the neurofibroma and concerning for a superimposed infection.

The surgical team was consulted for management of the neurofibroma hematoma and she was admitted to the surgical ICU for treatment of sepsis. Serial complete blood counts to monitor acute anemia and the degree of hematoma expansion were performed. She received one unit of packed red blood cells (pRBC) to treat her anemia at this point. Fetal status was reassuring by a reactive non-stress test. Shortly after admission to the ICU, providers noted a new area on her thigh that had spontaneously opened and was self-expressing large volume blood clots. On bedside exploration, there was approximately 500 cc of blood pooled around the patient's thigh, with evidence of ongoing bleeding. A repeat hemoglobin at that time was 7.7 g/dL, despite previously receiving the unit of pRBC as described above. Given the ongoing bleeding, the decision was made to proceed to the operating room (OR) for a left lower extremity hematoma debridement and control of hemorrhage.

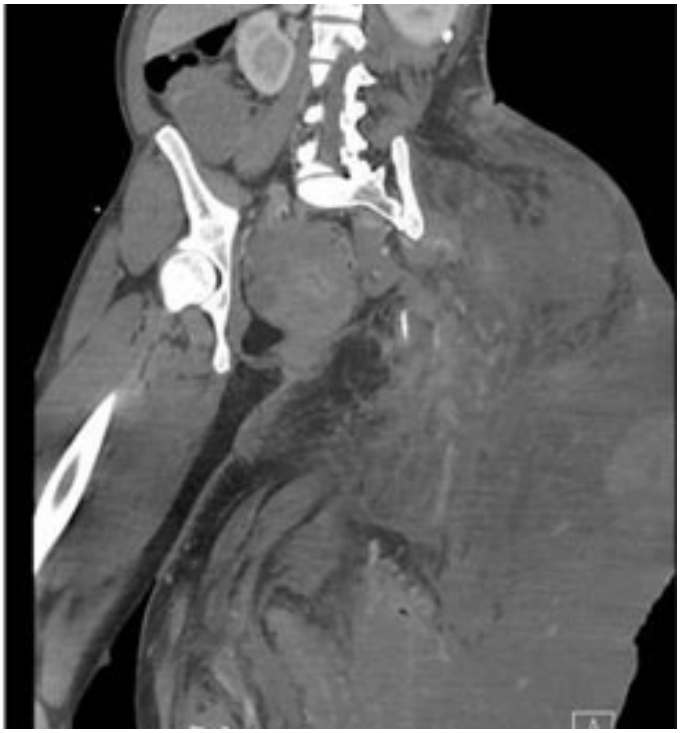

Figure 1: CT lower extremity with intravenous (IV) contrast. Imaging shows large left thigh neurofibroma, with partially visualized hematoma extending to lateral-most aspect of the left thigh concerning for active bleeding.

Prior to the OR, the patient was placed on continuous electronic fetal monitoring to assess fetal status, and a fetal bradycardia with a heart rate of 70 bpm was noted. Given this profound and persistent fetal bradycardia as well as concern for maternal hypotension with ongoing bleeding at the site of the neurofibroma, the decision was made to proceed with an emergency cesarean delivery. The patient was rapidly mobilized from the ICU to OR where she underwent an emergency uncomplicated transverse lower segment cesarean section under general anesthesia. The neonate's birthweight was 1480 grams, and arterial umbilical cord gas was notable for pH of 7.01, pCO_2 68, pO_2 of 6, and base excess of –13.0. The infant was admitted to the neonatal intensive care unit (NICU) due to prematurity and ultimately discharged home on day 45 of life in good condition.

It is of note that intraoperatively, following cesarean delivery, the surgical team was called in for debridement of the left thigh neurofibroma. The skin overlying the neurofibroma was determined to be nonviable and there was a 20 cm cavity in the left lateral thigh neurofibroma containing in excess of 750 mL of blood clot and necrotic tissue. The cavity was debrided and copiously irrigated with warm saline. At the base of the defect, a 0.5 cm diameter vein was identified and suture ligated to provide hemostasis (Figure 2). The wound cavity was then packed with surgical hemostatic agent and gauze.

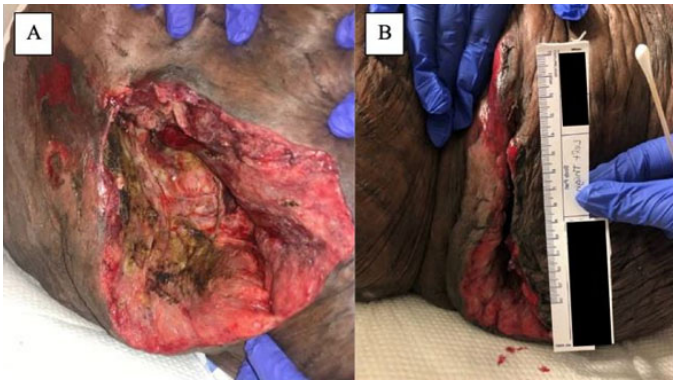

Figure 2: (A) Opened, debrided portion of patient's left leg neurofibroma status post intervention by general surgery. (B) Ruler added for size reference.

The patient was resuscitated with a total of 4 units pRBC, 4 units fresh frozen plasma, and 1 unit platelets in the operating room. The total estimated blood loss was 1800 mL, including 800 mL from the cesarean section and 1000 mL from the neurofibroma. She was subsequently transferred back to the ICU, extubated, and had an uneventful postoperative course. She required no further blood transfusion and was discharged home on day 4 postoperatively with a wound vacuum dressing over the left leg surgical site for the next 60 days. She has had subsequent follow-up with wound care outpatient, and her left leg wound continues to be healing well with no additional bleeding.

DISCUSSION

We present a case that describes a patient presenting with spontaneous hemorrhage of her known neurofibroma, ultimately requiring delivery of the fetus for fetal bradycardia likely secondary to profound maternal hypotension. Spontaneous hemorrhage of neurofibromas is a rare, but reported complication [3–5]. However, there have been no previous case reports of this occurring during pregnancy or necessitating delivery.

Though available data on this subject is mixed, NF1 has been associated with increased perinatal morbidity and mortality. For mothers with NF1, there is an increased risk of worsening hypertension or cerebrovascular complications during pregnancy. Additionally, NF1 has been associated with development of gestational hypertension, preeclampsia, growth restriction, preterm labor, and an increased rate of delivery by cesarean section [6]. Following this paradigm, our patient's first pregnancy was complicated by severe preeclampsia requiring delivery at 34 weeks gestation.

There is also data suggesting a physiological increase in size and number of neurofibromas during pregnancy. Most neurofibromas express progesterone receptors which may explain their growth during pregnancy. Indeed, there are case reports of NF lesions appearing only during pregnancy or showing signs of accelerated growth and worsening symptoms in pregnancy [7–9]. However, data also suggest that the natural history of neurofibromas is to increase in size over time, regardless of pregnancy; therefore, the meaningful impact of pregnancy on neurofibroma growth is difficult to ascertain [10]. Neurofibromas infrequently bleed spontaneously, and are rarely associated with severe hemorrhage. Few case reports have reported acute-onset hemorrhage secondary to spontaneous bleeding of neurofibromas [3–5]. While there is evidence that neurofibromas can increase in size and number during pregnancy, there has not been a documented case of hemorrhage during pregnancy, and neurofibromas are traditionally not expected to affect pregnancy outcomes [9].

CONCLUSION

We present a case where growth of a neurofibroma during pregnancy likely contributed to hemorrhage and affected obstetric management with a preterm delivery. In our case, the fetal bradycardia was likely due to maternal hypotension, which is a well-known etiology of fetal distress. We believe this is the first reported case of spontaneous hemorrhagic rupture of a neurofibroma that impacted obstetric management and required emergency cesarean delivery of a preterm fetus. In this case, prompt evaluation and management along with multidisciplinary care enabled a good maternal and fetal outcome.

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Author Contributions

Kira A Bromwich – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Margaret Rush – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Matthew Janssen – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Adi Hirshberg – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Guarantor of Submission

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Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.