JCEM Case Reports, 2024, **2**, 1–2
https://doi.org/10.1210/jcemcr/luae032
Advance access publication 1 April 2024
Image in Endocrinology

Pheochromocytoma Discovery During Pregnancy Leads to Neurofibromatosis Diagnosis

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Key Words: pheochromocytoma, pregnancy, neurofibromatosis type 1, café-au-lait macules, axillary freckling

Image Legend

A 27-year-old woman at 23 weeks gestation presented with abdominal pain. Her history was notable only for a placental abruption in a prior pregnancy. Her blood pressure was 160/83 mm Hg. Examination revealed iris hamartomas, café-au-lait macules (Fig. 1A), and axillary freckling (Fig. 1B). Ultrasound showed a 9-cm cystic right suprarenal mass (Fig. 1C) with intermediate T2 hyperintense signal on coronal T2-weighted magnetic resonance imaging (Fig. 1D). On 24-hour urine collection, metanephrine and normetanephrine levels were 605 mcg/day (3.067 µmol/day) (reference range, 36-299 mcg/day [0.183-1.516 µmol/day]) and 8547 mcg/day (46.652 µmol/day) (reference range, 95-650 mcg/day [0.519-3.548 µmol/day]), respectively. A pheochromocytoma was diagnosed. Her adrenergic symptoms and blood pressure were ultimately well controlled with doxazosin and metoprolol. She delivered a healthy son via uncomplicated cesarean delivery at 34 weeks gestation. Six weeks later an uncomplicated laparoscopic right adrenalectomy was performed. Pathology

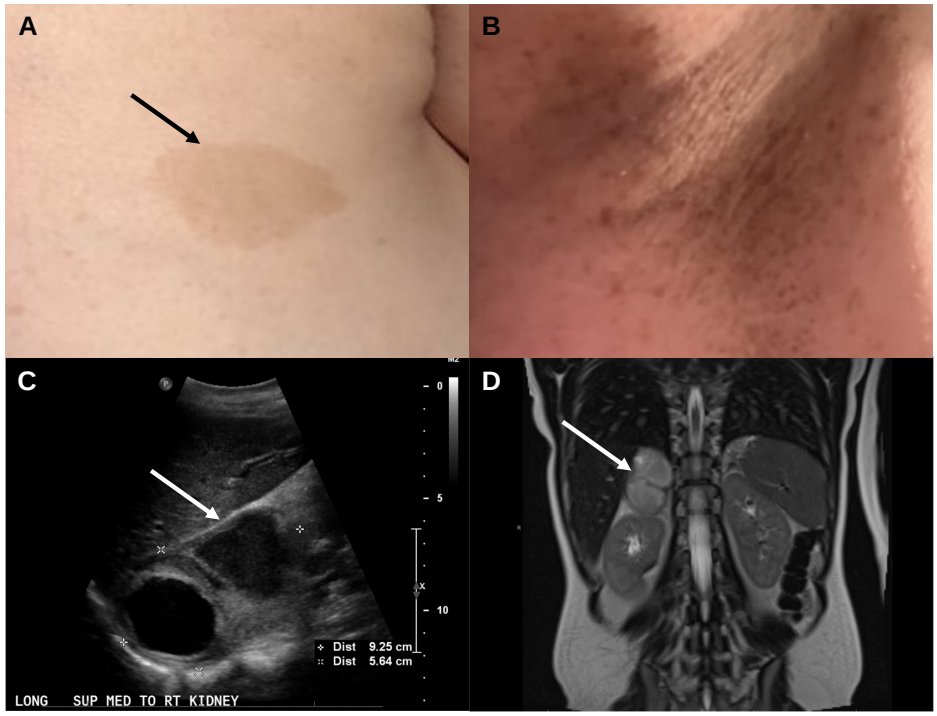

Figure 1. Unifying dermatologic and radiologic observations. On examination, the patient had café-au-lait macules (A) and axillary freckling (B). Ultrasound showed a 9-cm cystic right suprarenal mass (C), also identified on magnetic resonance imaging (D) as a mass with intermediate T2 hyperintense signal.

Received: 8 January 2024. **Editorial Decision:** 13 February 2024. **Corrected and Typeset:** 1 April 2024

confirmed a 7.1-cm pheochromocytoma. With genetic testing, a likely pathogenic frameshift variant in NF1 was detected in the heterozygous state (NF1:NM_001042492.1; c.[7545_7546insT; 7549C>G; 7553C>T] p.[Pro2516Serfs*20; Arg 2517Gly; Ala2518Val]). This case emphasizes the importance of recognizing features of NF1, an autosomal dominant condition with an estimated prevalence of 1 in 2000 to 1 in 3000, such as iris hamartomas, café-au-lait macules, and skinfold freckling, to facilitate early diagnosis and also that pheochromocytomas can be successfully managed during pregnancy with medical therapy with favorable outcomes (1, 2).

Funding

No public or commercial funding.

Disclosures

None declared.

Informed Patient Consent for Publication

Signed informed consent obtained directly from the patient.

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