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IMAGING

IMAGING VIGNETTE: CLINICAL VIGNETTE

Myocardial Infarction and Coronary Anomalies in a 25-Year-Old Neurofibromatosis Type 1 Patient

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ABSTRACT

A 25-year-old male with neurofibromatosis type 1 presented with acute ST-segment elevation myocardial infarction. Coronary imaging revealed an aberrant right coronary anatomy, ectatic coronary arteries, and significant stenosis. Based on previous literature and clinical presentation, this case highlights the potential role of neurofibromatosis in the pathogenesis of coronary artery disease. (J Am Coll Cardiol Case Rep 2024;29:102261) © 2024 The Authors. Published by Elsevier on behalf of the American College of Cardiology Foundation. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).

A 25-year-old male presented to the emergency room with acute anginal symptoms. Ambulance-obtained electrocardiogram showed acute inferoposterolateral ST-segment elevation myocardial infarction (STEMI) (**Figure 1A**). Upon admission heartrate was 74 beats/min, blood pressure 124/80 mm Hg, and O_2 saturation 100% without suppletion. No elevated central venous pressure was observed, and cardiac auscultation was normal. Invasive coronary angiography showed an aberrant origin of the right coronary artery (RCA) originating from the left coronary cusp (LCC) and overall remarkable ectatic remodeling of the coronary arteries (6 mm lumen diameter).

The mid-segment of the RCA had 95% stenosis with plaque rupture (**Figure 1B**), and concomitant coronary artery disease (CAD) in the proximal left anterior descending coronary artery with 95% stenosis (**Figure 1D**). Due to the ectatic nature in combination with angulation and lack of support of several different guiding catheters, performing intracoronary imaging of the RCA was unsuccessful. Successful primary percutaneous coronary intervention (PCI) of both lesions was performed (**Figures 1C and 1E**). Coronary computed tomography angiography (CCTA) confirmed the benign interatrial RCA originating from the LCC and diffuse ectatic CAD (**Figure 1F**). Additional invasive coronary angiograms and images obtained through CCTA can be found in the Supplemental Material (Supplemental Figures 1 to 4, Videos 1 to 5).

Upon admittance, the patient did not use any medication. Post-PCI, the patient received dual antiplatelet therapy, atorvastatin, metoprolol, and ramipril. Cardiovascular risk factors present in this patient were smoking, dyslipidemia, and a positive family history for cardiovascular disease. Furthermore, the patient had been previously diagnosed with neurofibromatosis type 1 (NF1).

NF1 is an autosomal dominantly inherited disease primarily leading to the development of benign neurofibromas. The NF1 gene encodes for neurofibromin, a cytoplasmic antioncogenic protein that inhibits the proliferative function of the RAS-signaling pathway.[1] Loss of inhibition of this pathway is thought to cause

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ABBREVIATIONS AND ACRONYMS

CAD = coronary artery disease

CCTA = coronary computed tomography angiography

LCC = left coronary cusp

NF1 = neurofibromatosis type 1

PCI = percutaneous coronary intervention

RCA = right coronary artery

STEMI = ST-segment elevation myocardial infarction

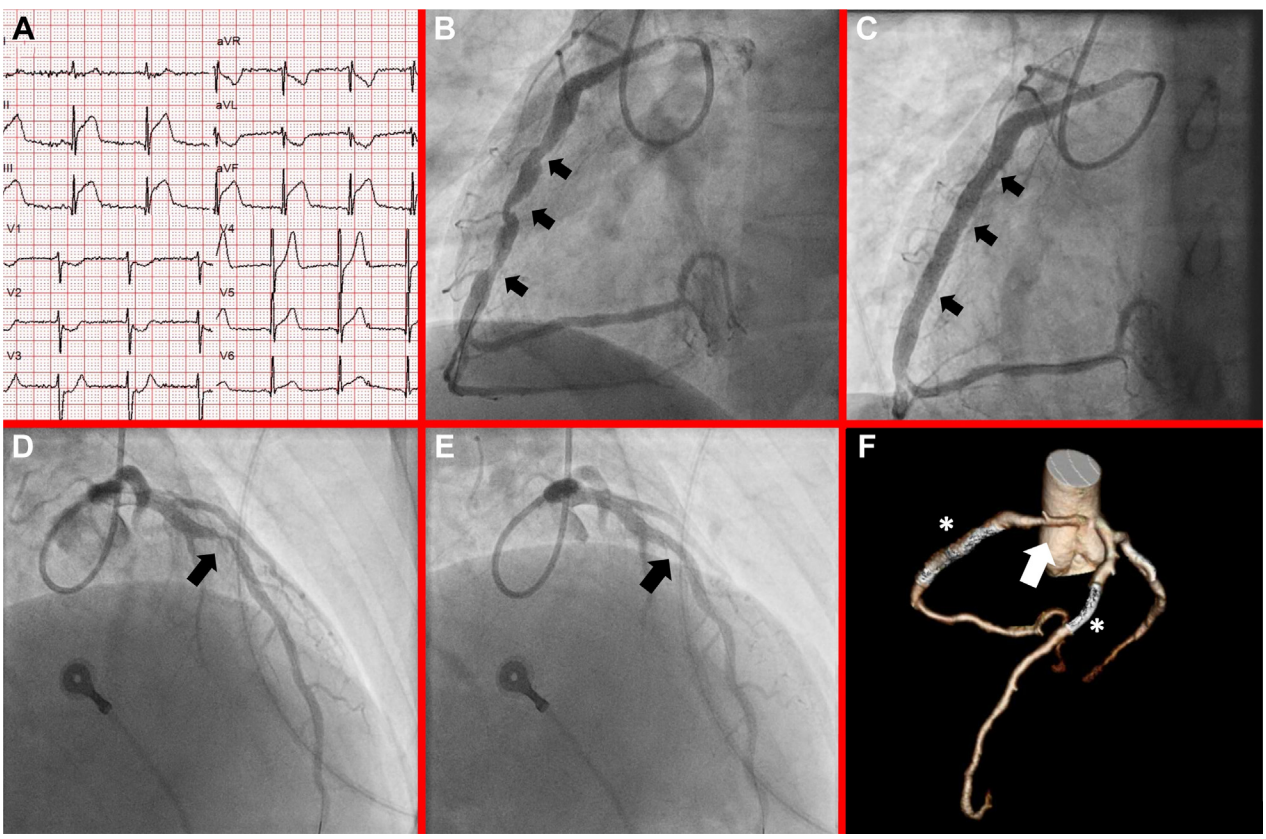
abnormal smooth muscle cell proliferation and migration in response to vascular injury in the intimal layer of arterial vessels, causing diverse NF1-associated vasculopathies that can arise in any arterial vessel.[1,2]

Several case reports have shown an association between NF1 and obstructive and/or aneurysmal CAD (ACAD).[3] In a case series published in 2022, Corona-Rivera et al[3] presented an overview of 17 published cases of NF1 patients with ACAD.[3] ACAD was diagnosed in patients younger than the age of 40 years in 11 of these 17 cases, the youngest being 13 years old at the time of diagnosis.[3] Most of the patients (76%) presented with anginal symptoms, similarly to the patient currently presented.[3]

Even considering the multiple cardiovascular risk factors, the extent and degree of CAD and remodeling is remarkable at this young age. Given the observational evidence from numerous published case reports and pathophysiological insights, it seems plausible that NF1 has played a key role in the pathogenesis of this very early-onset and severe CAD.[1,3]

This case describes a rare occurrence of STEMI and ectatic CAD, in concomitance with aberrant coronary anatomy in a young individual with NF1, emphasizing the potential interplay between NF1 and traditional cardiovascular risk factors in the pathogenesis of CAD.

FIGURE 1 Electrocardiogram and Coronary Imaging of an Individual With NF1 Presenting With Anginal Symptoms



(A) The electrocardiogram acquired in the ambulance shows ST-segment deviations in the inferior, posterior, and lateral leads corresponding to an inferoposterolateral ST-segment elevation myocardial infarction. Coronary angiography (CAG) revealed an obstructed and ectatic right coronary artery (RCA) originating from the left coronary cusp (B) and an ectatic left anterior descending coronary artery (LAD) with significant stenosis (D). Successful percutaneous coronary intervention (PCI) was performed (C, E). The black arrows indicate sites of significant stenosis in the RCA and LAD before (B, D) and after PCI (C, E). The aberrant anatomy of the RCA observed during CAG was confirmed by coronary computed tomography angiography (CCTA) (F). The white arrow indicates the interatrial part of the RCA (F). Stents placed during PCI are visible in the RCA and proximal LAD on CCTA, indicated by white asterisks (F).

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KEY WORDS computed tomography, coronary angiography, coronary vessel anomaly, genetic disorders, genetics, myocardial infarction, neurofibromatosis, neurofibromatosis type 1, percutaneous coronary intervention, risk factor

APPENDIX For supplemental figures and videos, please see the online version of this paper.