Neurofibromatosis Type 1 and Unexpected NSTEMI: A Case of RCA Occlusion in a Young Male Patient

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CASE REPORT
A 39-year-old male patient with a past medical history of Neurofibromatosis type 1 and a history of left leg sarcoma treated with radiation presented with chest pain lasting for one day. He reported no other associated symptoms. The pain increased with movement and was relieved by rest.

The patient denied experiencing similar symptoms in the past and is generally physically active. He does not smoke and has no family history of coronary artery disease.

On admission, the patient had high blood pressure measuring 165/100, but other vital signs remained stable. An electrocardiogram (EKG) showed non-specific T wave changes. His troponin-I levels were elevated and peaked at 22.50. The patient was referred for invasive coronary assessment, which revealed a totally occluded right coronary artery (RCA) treated with IVUS-guided PCI.

The patient was started on Aspirin, Plavix, Lisinopril, Metoprolol, and atorvastatin. An echocardiogram showed no remarkable findings, with preserved ejection fraction (EF%). The patient’s condition improved after the procedure and he was discharged home.

RESULTS

DISCUSSION
NF1 1 is typically a hereditary condition caused by mutations in the NF1 gene. The clinical features of NF1 include café-au-lait macules, freckling in the armpit or groin area, neurofibromas, lisch nodules, osseous lesions, and sometimes an optic pathway glioma(1).

Patients with NF1 may also develop arterial disorders such as aneurysms or blockages in the heart or peripheral blood vessels. Vasculopathy in NF1 patients affects vessels in the brain, kidneys, and aorta(2).

Reported cases of myocardial infarction in NF1 patients suggest various mechanisms like blockages, spasms, or aneurysms in the coronary arteries(3,4).

It is believed that the underlying pathological changes responsible for vasculopathies in NF1 involve abnormalities in blood vessel structures, including smooth muscle proliferation, new blood vessel formation, and abnormal intimal proliferation(4).

Echocardiogram studies have revealed that up to 27% of NF1 patients exhibit cardiac anomalies.(1)

RESOURCES/REFERENCES
4. Vasculopathies of neurofibromatosis type 1 (von Recklinghausen Disease) Lie JT. Cardiologu...