Abstract

Homocystinuria is a rare, autosomal recessive, inborn error of metabolism, first described in 1962. It's characterized by the deficiency of the enzyme cystathionine beta-synthase (CBS), which converts homocysteine to cystathionine. As a result, patients with homocystinuria have significant elevations in blood and urine homocysteine levels. Untreated patients will develop multiple system manifestations, including eye involvement, musculoskeletal involvement, developmental delay with or without other neurological symptoms, and cardiovascular involvement like arterial or venous thrombotic events resulting in acute coronary syndrome and stroke. Early diagnosis and management of this patient are very important to prevent these complications.

Our patient had been diagnosed with classical homocystinuria at 10 years old. He was also diagnosed with B/L early nephrocalcinosis at 10 years of age when he was evaluated for back pain. His sister is also a diagnosed patient with homocystinuria.In this admission, our patient was treated for non-ST elevation MI. The coronary angiogram was normal and didn't show evidence of coronary occlusion. Hence, we considered this case as a MI with non-obstructive coronary arteries (MINOCA).