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CARDIOVASCULAR FLASHLIGHT

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Arterial elongation and tortuosity leads to detection of a *de novo TGFBR2* mutation in a young patient with complex aortic pathology

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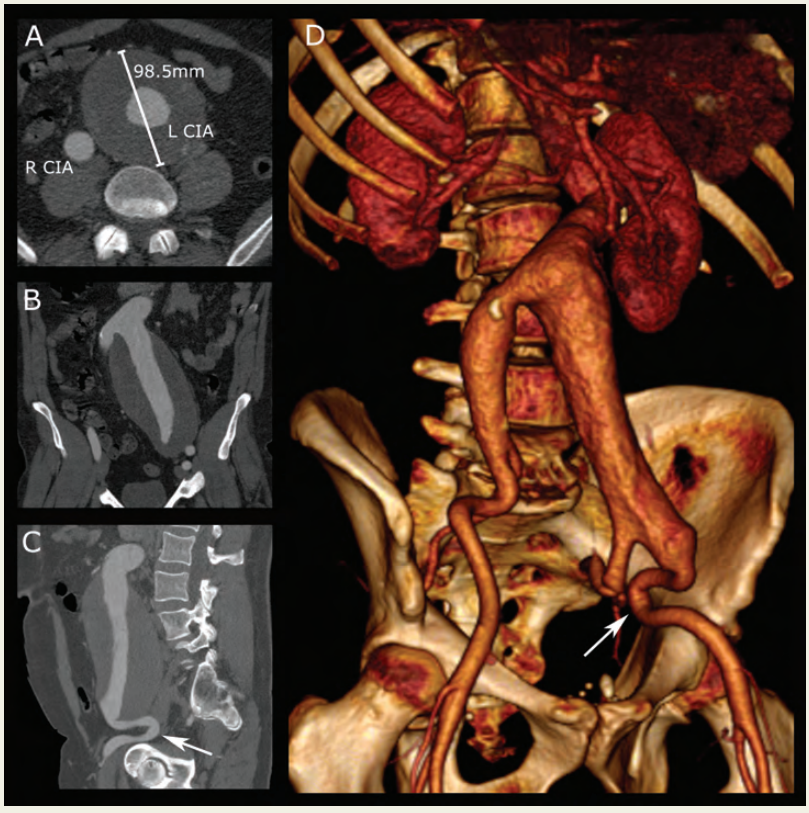

In a 47-year-old muscular-build male admitted with acute abdominal pain imaging revealed a Stanford type-B aortic dissection associated with a pre-existing large aorto-iliac aneurysm (*Panels A* and *B*) and marked iliac-artery elongation and tortuosity (*Panels C* and *D*, arrows). The patient underwent uneventful elective open repair of the aorto-iliac aneurysm. The marked aortic elongation and tortuosity at a young age in this patient prompted referral for genetic counselling after surgery. No characteristic facial or musculoskeletal signs of Loeys-Dietz or Marfan syndrome were present and there was no family history of vascular disease. Nevertheless, DNA analysis showed a '*de-novo*' *TGFBR2* mutation.

Arterial elongation and tortuosity is a main feature in patients with characteristic facial and musculoskeletal appearance of the TGF-β pathway-related genetic aneurysm syndromes. Therefore, our observation expands the phenotypic spectrum of TGF-β pathway-related pathology to patients with severe abdominal and iliacal arterial disease without major dysmorphological characteristics.

We demonstrate the importance of genetic testing in younger patients presenting with complex aortic pathology with marked arterial elongation and tortuosity, even in the absence of characteristic phenotypic features of a genetic aneurysm syndrome or a family history of aortic aneurysms. Correct genetic diagnosis of *TGFBR2*-related aortic pathology is important for clinical management of the patients and for genetic counselling of the family. Since *TGFBR2*-linked genetic aneurysms have an autosomal dominant inheritance, relatives at risk should be offered genetic counselling and pre-symptomatic testing for *TGFBR2* mutations. In this way, carriers of the *TGFBR2* mutation can benefit from screening and timely intervention.

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