Multidisciplinary Approach in Loeys-Dietz Syndrome and Complex Aortic Disease

Presenter: Lucas Ribé MD,

Yuki Ikeno, Milan Jaiswal, Rana O Afifi, Akiko Tanaka, Anthony L Estrera, Siddharth K Prakash.

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Department of
Cardiothoracic &
Vascular Surgery

UTHealth
McGovern
Medical School
The University of Texas
Health Science Center at Houston

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Objective

Genetic factors can play a significant role in the development of thoracic aortic disease (TAD).

Around 20% of individuals with a TAD have a first-degree relative with aortic dilatation.

One example is the TGFB2 gene, which is associated with Loeys-Dietz syndrome (LDS).

Since the clinical presentation of these types of diseases can vary between patients, a multidisciplinary care team is important for proper management of the patient.

Methods

The multidisciplinary approach is designed for complex aortic disease patients and developing a patient care plan from the expertise of a panel of specialists.

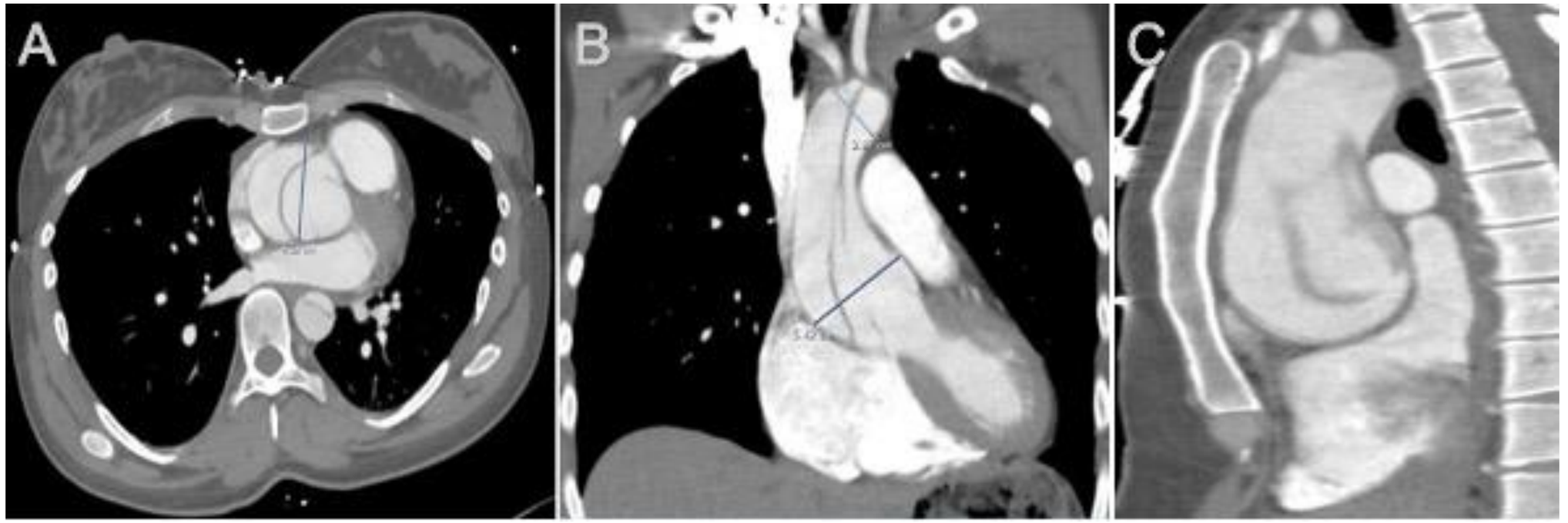
In this report, we highlight the unique management of four individuals with distinct presentations of LDS in the context of aortic disease.

Case 1

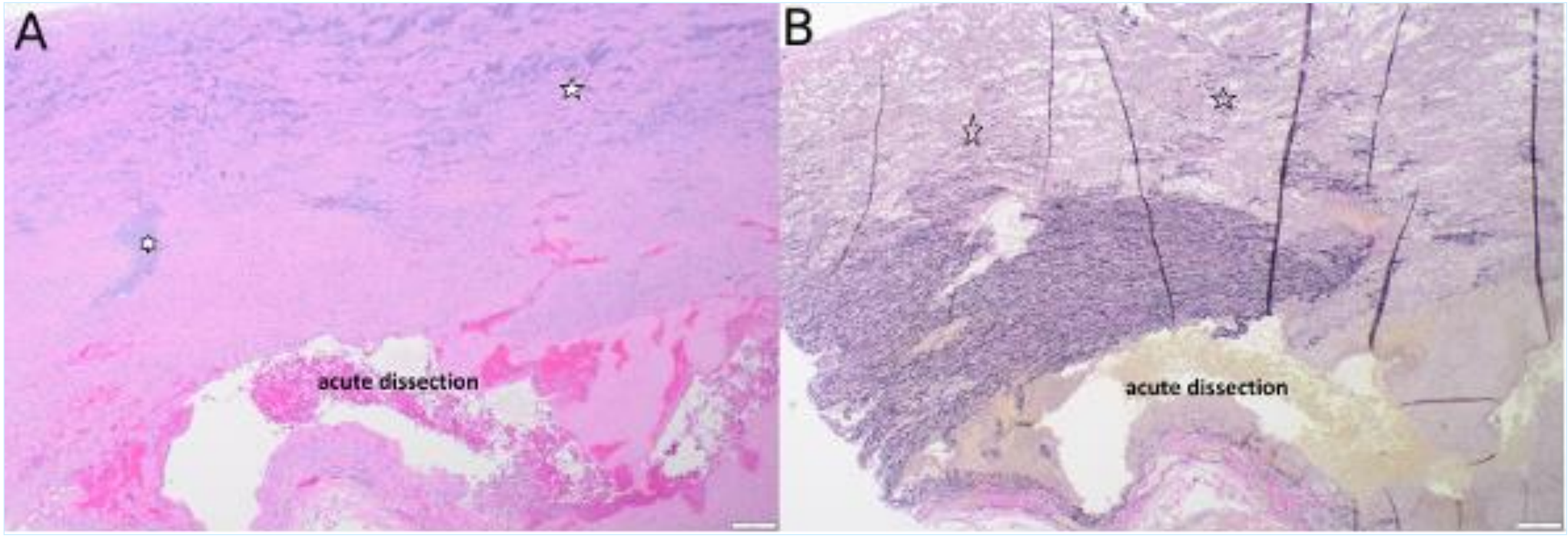
33 years old, Female

- Acute type A aortic dissection with root aneurysm

→ David procedure



Marked elastin fiber damage, mild accumulation of extracellular matrix material, and increased fibrosis in the intimal, subintimal, and medial layer.



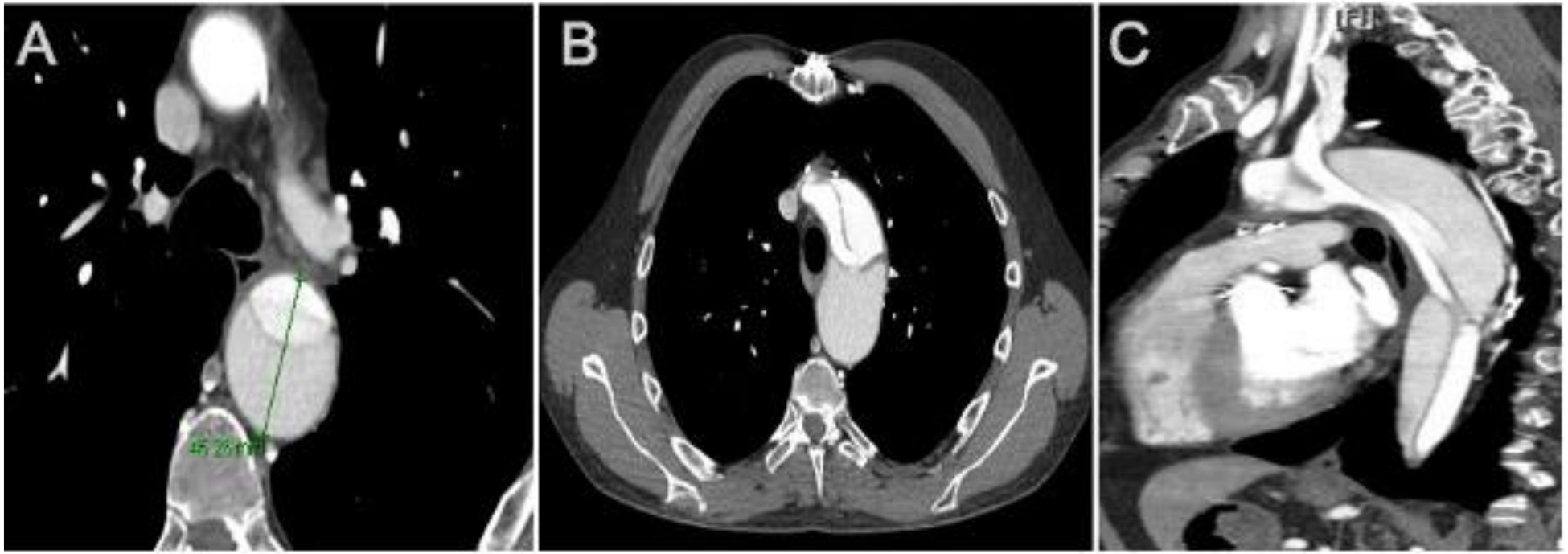
Genetic tests identified a pathogenic variant in **TGFB2 (p.Y126Sfs*19)** that is consistent with LDS.

Case 2

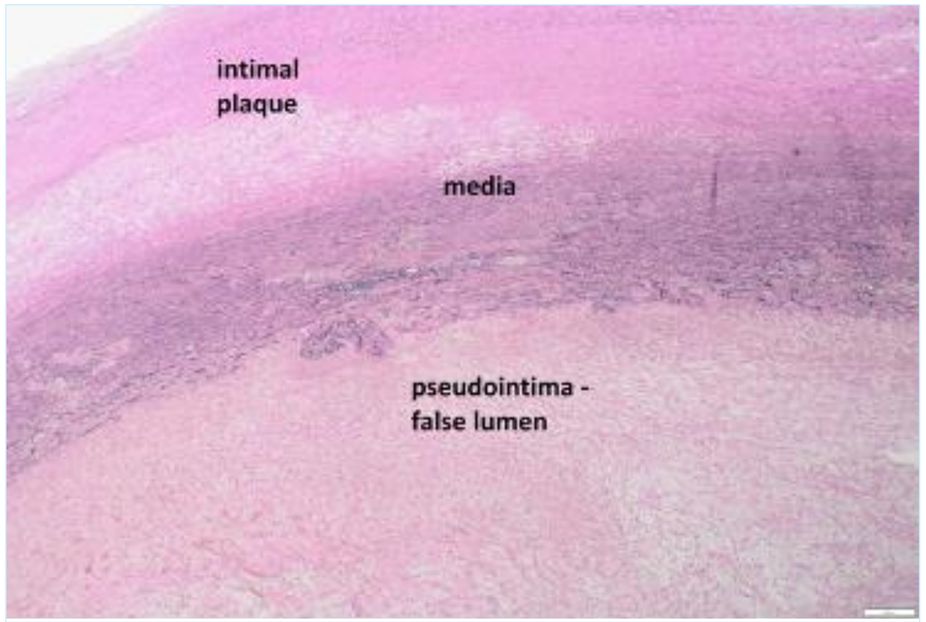
33 years old, Male

- Acute type A aortic dissection with root aneurysm

→ Modified Bentall procedure



Medial degeneration with focal necrosis and disruption of elastic fibers is also present.



Within 3 years, the diameter of the brachiocephalic artery (2.9 cm), aortic arch (4.2 cm), and proximal descending thoracic aorta (5.4 cm) had significantly increased.

Genetic tests identified a pathogenic variant in **TGFB2 (p.R330C)** that is consistent with LDS.

Case 3

38 years old, female

- Chronic type A aortic dissection with root aneurysm

→ Aortic valve, root, and ascending replacement with a porcine bioprosthetic valve.

Follow-up surveillance

Both of her sister's children were diagnosed with aortic aneurysm at age 10 and 15. Both have undergone repair for pectus excavatum, and one of them survived a Type A dissection at age 23, when his aortic root was measured at 5.4 cm.

Genetic testing revealed a pathogenic variant in **TGFB2 (p.C380F)**, consistent with the diagnosis of **LDS.**

Case 4

53 years old, female

- Chronic type A aortic dissection with root aneurysm

- At age 43, moderately dilated aortic root aneurysm (4.6 cm) with severe AR

→ Modified Bentall Procedure

Follow-up surveillance

By 3 years, images showed a progressive increase in the maximum **thoracoabdominal aortic diameter to 4.1 cm.**

Genetic testing: this patient is the sister of the case 3 and carries the same pathogenic variant of **TGFB2 (p.C380F).**

Conclusions

These cases illustrate the various clinical manifestations of LDS and the high risk of developing subsequent arterial disease after the index event.

Although several of these patients were being observed carefully and were on appropriate medical therapies, they nonetheless developed progressively enlarging aneurysms or new dissections.

LDS patients require lifelong management and follow-up to prevent deaths due to recurrent aortic events.