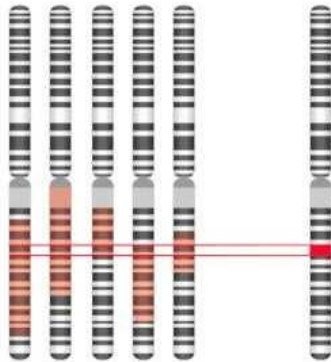




Genetically Personalised Care for Aortic Aneurysms



A study published in *The Annals of Thoracic Surgery* outlines the work of researchers at the Aortic Institute at Yale who have tested the genomes of more than 100 patients with thoracic aortic aneurysms and have provided personalised care. It is believed that their work could lead to the development of a "dictionary" of genes specific to the disease.

It has already been established by experts that thoracic aortic aneurysms run in families and are caused mainly by specific genetic mutations. However, the testing for these mutations are fairly expensive and impractical.

The Aortic Institute collaborated with Allen Bale, M.D. of Yale's Department of Genetics to launch a program to test whole genomes of patients with the condition. The research team applied the Whole Exome Sequencing (WES) technology to more than 100 individuals with aortic aneurysms over a period of three years. It is the first widespread application of WES to this disease.

The researchers found four mutations known to cause thoracic aortic aneurysms. By applying this technology, they were able to identify patients with genetic mutations. They also uncovered 22 previously unknown gene variants that likely also contribute to the condition and were able to tailor treatment according to each patient's genetic profile. Thus, they were able to make personalised aortic aneurysm care a reality. They also offered testing to family members and found genetic mutations in relatives that showed no signs of the disease.

The researchers are confident they can identify more gene variants over time, accumulating a whole dictionary of mutations. Lead author and cardiac surgeon John A. Elefteriades, M.D., director of the institute says, "In a few years, we're going to have discovered many new genes and be able to offer personalised care to an even greater percentage of aneurysm patient."

Source: [Yale University](#)

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