



# MUTATION ANALYSIS OF THE FBN1 GENE IN CZECH PATIENTS WITH AORTIC DILATATION

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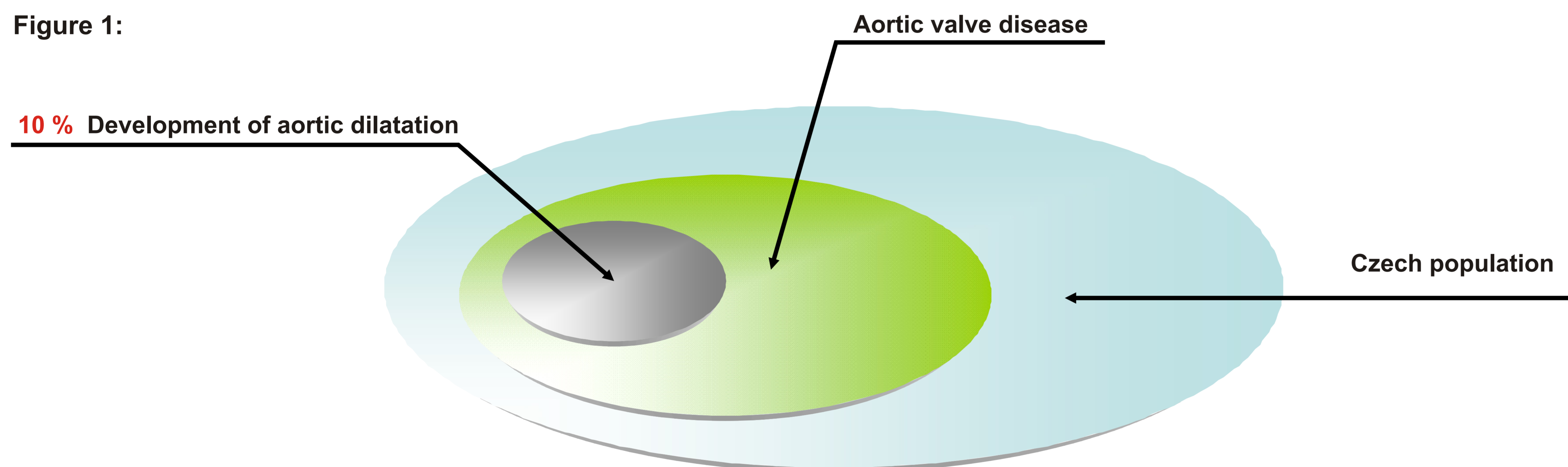
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## INTRODUCTION

- About 10% of patients operated upon for aortic valve disease suffer simultaneously from ascendant aortic dilatation
- Development of dilatation is probably based on pathologically changed aortic wall
- The current study suggests that the role of fibrillin 1 is important in relation to cardiovascular risk associated with aortic dilatation

Genomic DNA was collected from 28 probands diagnosed with aortic valve disorder associated with aortic dilatation

Figure 1:



Aortic dilatation - poststenotic

Years of Age	55±20 (35-75)
Male/Female	22/6

## CLINICAL OBSERVATION

Cause and progression of aortic valve disorder is considered to be the contributory factor for dilatation of primarily changed aortic wall of ascendant aorta. Patients are at risk of fatal complication that correlates with the diameter of dilatated ascendant aorta.

Marfan Syndrome Diagnosis was excluded because of well known symptoms of this disorder

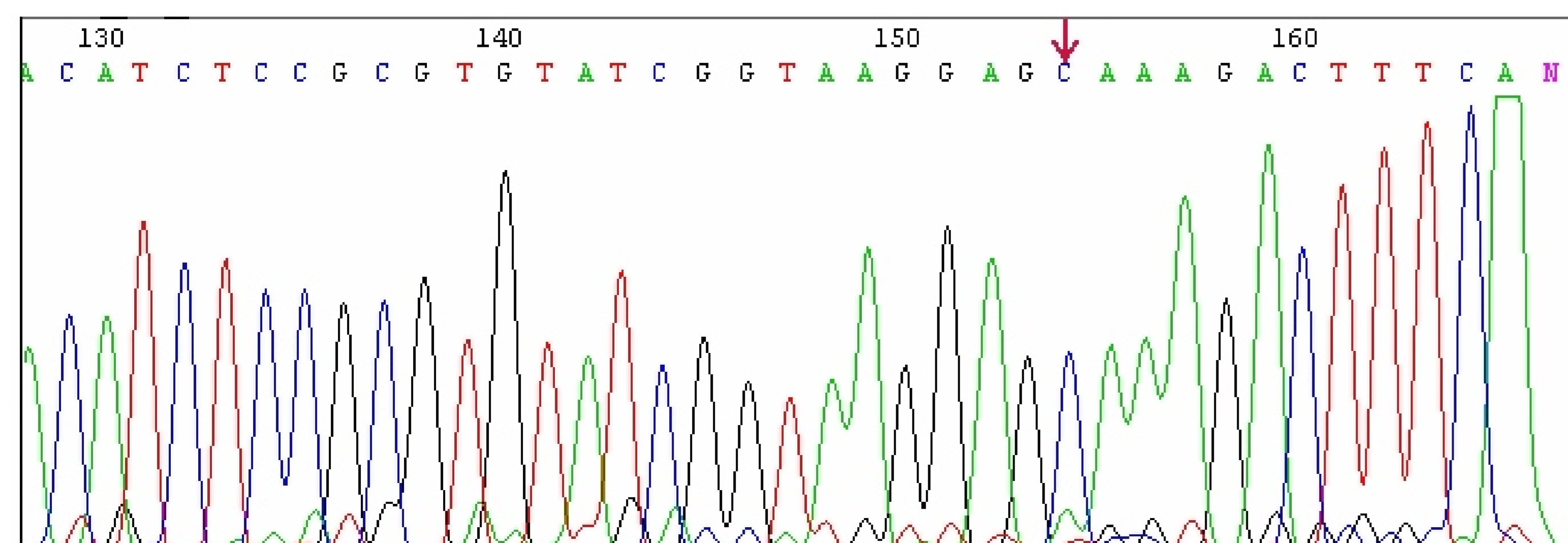
Aortic Stenosis	19
Aortic Regurgitation	9

## METHODS

Genomic DNA was extracted from white blood cells from peripheral blood samples. Exons 26/27 of FBN1 Gene were amplified using primers specific for chosen exons including their intron/exon boundary. All PCR products were sequenced in both directions.

## RESULTS

Figure 2: The location of the insertion is shown by the red arrow



Amplification and sequence analysis of exon 26 and 27 were without changes. Analysis of intronic part situated close to exon 27 showed an insertion of guanine between nucleotide 37,682 and 37,683 of query sequence. We classified this as

**IVS27 37682\_37683insC**

## SUMMARY

The current study suggests that sequence variation in the genes encoding proteins constituting the aortic wall and regulating the turnover of the extra cellular matrix are likely to influence properties of elastic fibers.

This is an initial study and although a causative link has not been shown, these data are very important for further research of the role of fibrillin 1 in relation to cardiovascular risk associated with aortic dilatation.

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