

The heritability of aortic dimensions: initial experience in a classical twin study

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Aims and objectives

Changes in aortic diameters can be related to healthy ageing and/or to variety of disorders (such as hypertension, heart failure, dissection or rupture) [1,2,3].

Modern multidetector computed tomography (MDCT) devices are useful in order to identify these disorders, since it provides reliably accurate measurements to visualize the arteries of the body [4].

The extent of genetic and environmental influences over aortic structural parameters is still unclear. The inheritance of the aortic geometry has been in the focus of studies in recent years [5,6]. However, the data regarding aortic diameter and geometrical taper with MDCT in twins is sparse [6].

Therefore our aim was to estimate the inheritance of the aortic diameter, cross sectional area and geometric tapering within a sample of Hungarian twins.

Methods and materials

We measured the aortic parameters of 84 patients who underwent coronary CT angiography (CCTA) between February 2013 and November 2013. Until recently, 42 twin pairs, (21 mono- [MZ] and 21 dizygotic [DZ]) were enrolled in our study. Mean age of MZ and DZ twins was 56.7 ± 8.3 and 57.8 ± 9.7 years.

The subjects were investigated with a 256-slice CT-scanner (Brilliance iCT, Philips Healthcare, Best, The Netherlands). For each participant we determined ascendent and descendent aortic diameters and cross sectional areas.

Ascending aortic measurements were carried out in the level of the aortic annulus, the sinus of Valsava, the sinotubular junction and in the ascending aorta, 10 mm distal from the sinotubular junction. [Fig. 1](#) on page 3

The descendent aortic parameters were measured at 3 different locations. For heritability calculations average of the 3 measurements were used. [Fig. 2](#) on page 4, [Fig. 3](#) on page 5

Geometric tapering was calculated as the difference of the descendent and ascendent parameters. Data analysis was performed using Statistica version 11 (StatSoft, Inc., Tulsa, OK). All continuous variables were expressed as mean $\pm$ SD. All phenotypic variables were screened for outliers and normality. In absence of normality logarithmic

normalization was carried out. Concordance between MZ and DZ pairs were assessed by Pearson correlations.

In this early phase of our investigation comprehensive analysis of A, C and E components of phenotypic variance and covariance would be premature due to the small sample size. Our goal was therefore to approximate the weight of genetic influence solely on the examined phenotypic parameter.

According to Falconer, correlation within MZ twin pairs (r_{MZ}) can be described numerically as:

$$r_{MZ} = A + C \text{ (100\% genetic match)}$$

Correlation within DZ twin pairs (r_{DZ}) can be written as:

$$r_{DZ} = 0.5 \times A + C \text{ (50\% genetic match)}$$

By expressing C from the two equations:

$$r_{MZ} - A = r_{DZ} - 0.5 \times A, \text{ by rearrangement:}$$

$$2 \times (r_{MZ} - r_{DZ}) = A$$

On a population scale, the resulted A value informs us about the relative strength of genetic influence (heritability) determining the particular phenotype in question.

Images for this section:



Fig. 1: Double oblique view of the aortic root indicating the positions where the measurements of the ascending aorta were taken.

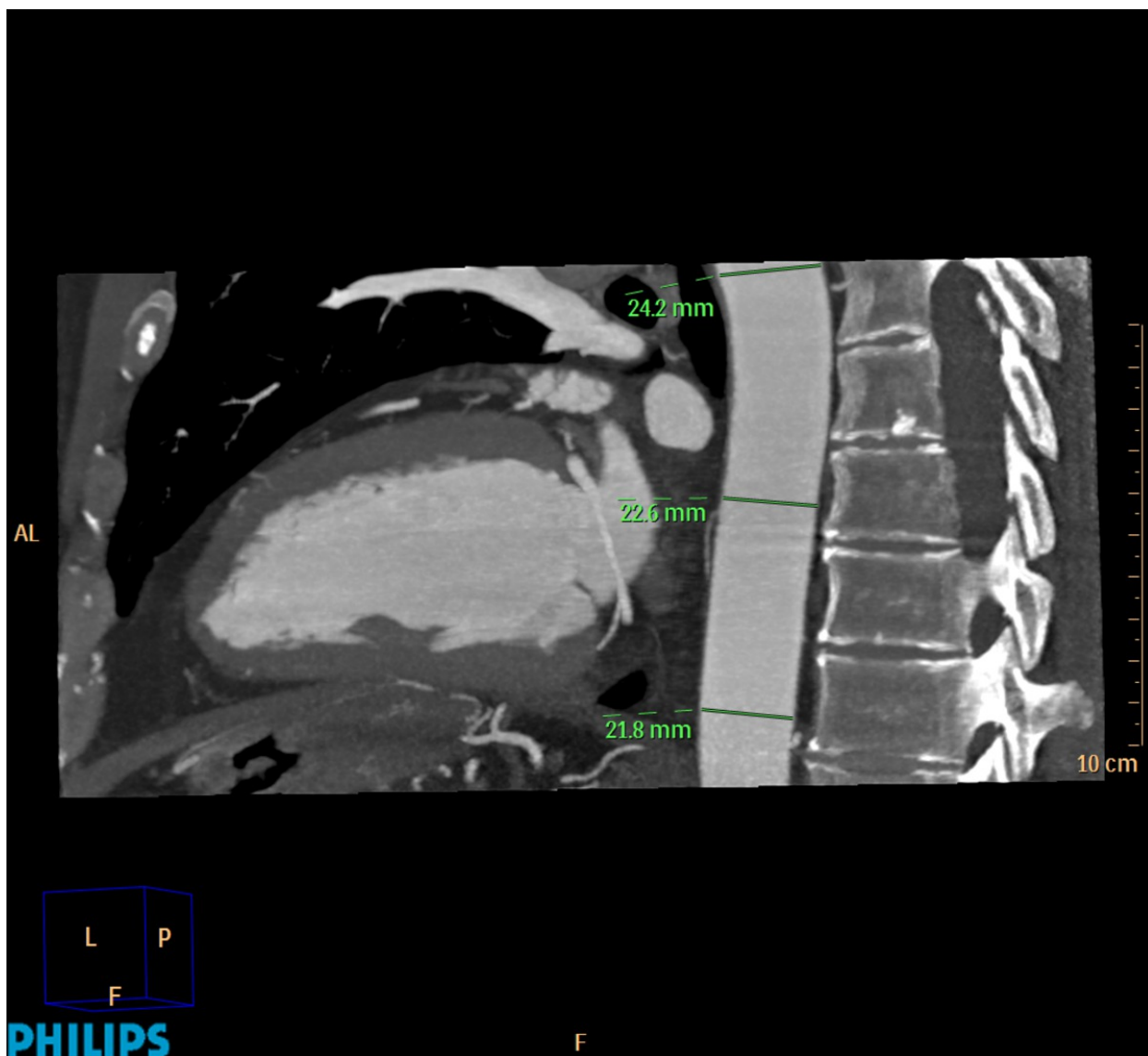


Fig. 2: Sagittal view of the descending aorta. The three measurements were taken at the level of the trachea bifurcation, the aortic annulus and the diaphragm.

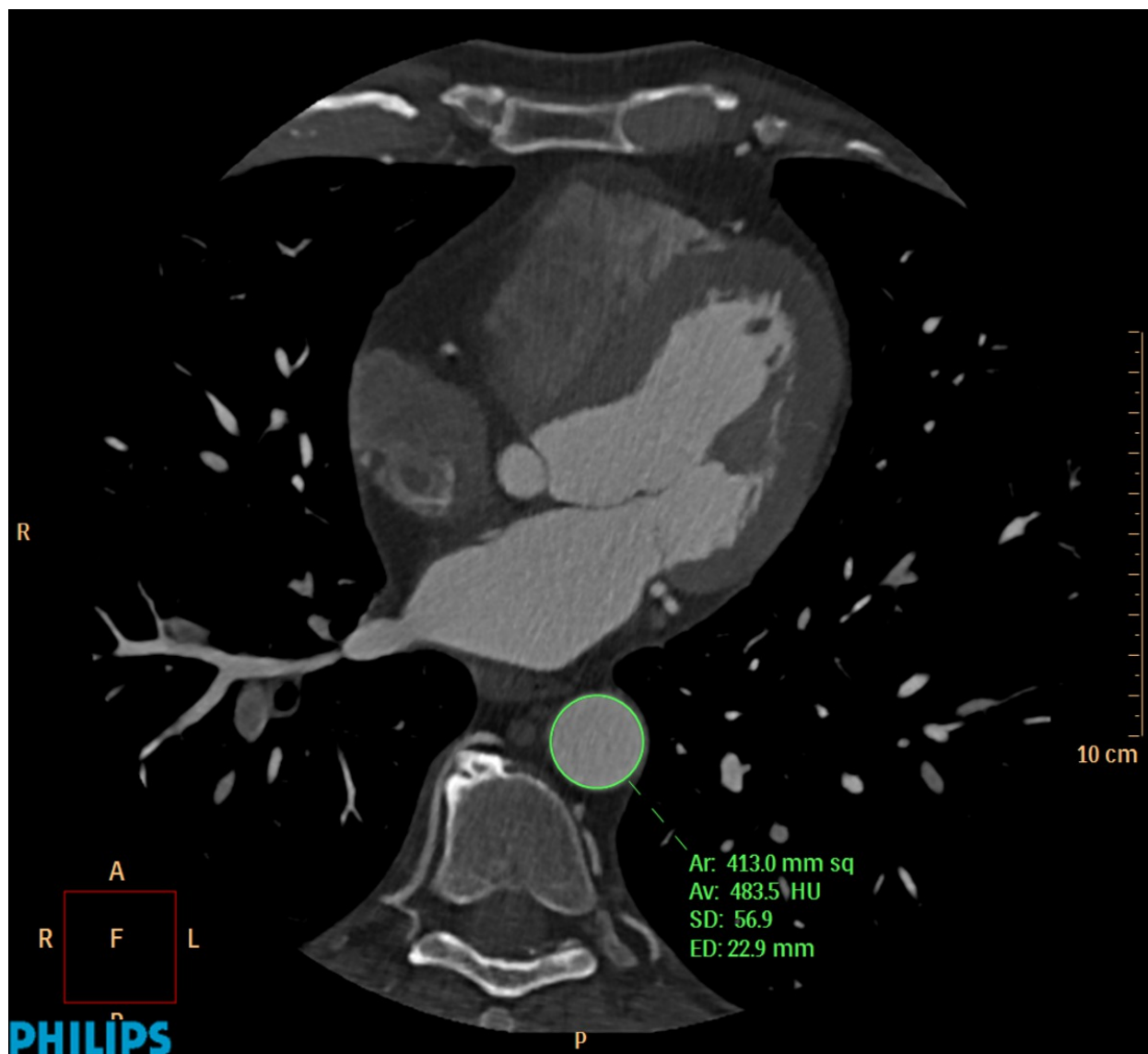


Fig. 3: Measurement of the aortic area and diameter at the level of the aortic annulus.

Results

The mean diameter of the ascending and descending aorta was 29.1 ± 3.8 and 21.4 ± 2.5 mm, respectively.

The mean ascending aorta diameter and area of monozygotic twins were 29.3 ± 3.4 mm and 683.1 ± 171 mm², in dizygotic twins 28.8 ± 4 mm and 664 ± 190 mm², respectively. The mean descending aorta diameter and area of monozygotic twins were 21.3 ± 2.1 mm and 362.7 ± 73.6 mm², in dizygotic twins 21.1 ± 3.4 mm and 359.1 ± 102 mm², respectively. The mean sinotubular junction diameter and area of monozygotic twins were 27.3 ± 2.7 mm and 592.5 ± 118 mm², in dizygotic twins 27.6 ± 3.8 mm and 613 ± 174.2 mm², respectively.

[Table 1](#) on page 7

Highest heritability was found regarding the area of the ascending aorta (0.667) and aortic diameter taper (0.644). [Table 2](#) on page 8

Images for this section:

	Monozygotic twins (n=42)		Dizygotic twins (n=42)	
	Diameter (mm)	Area (mm ²)	Diameter (mm)	Area (mm ²)
Aortic annulus	22.39 ± 2.43	398 ± 87.8	22.6 ± 2.2	405 ± 77.5
Sinus Valsava	30 ± 3.2	770 ± 169.3	30.8 ± 3.9	804.3 ± 211.9
Sinotubular Junction	27.3 ± 2.7	592.5 ± 118	27.6 ± 3.8	613 ± 174.2
Ascending Aorta	29.3 ± 3.4	683.1 ± 171	28.8 ± 4	664 ± 190
Descending Aorta	21.3 ± 2.1	362.7 ± 73.6	21.1 ± 3.4	359.1 ± 102

Table 1: Measured parameters of the ascending and descending aorta.

	rMZ	CI 95%	p-value	rDZ	CI 95%	p-value	H
Ascending aorta diameter	0.541	0.130-0.794	<0.05	0.250	-0.203-0.615	NS	0.582
Ascending aorta area	0.512	0.089-0.778	<0.05	0.179	-0.274-0.657	NS	0.667
Taper diameter	0.538	0.111-0.797	<0.05	0.216	-0.264-0.610	NS	0.644
Taper area	0.506	0.067-0.780	<0.05	0.393	-0.073-0.719	NS	0.226

rMZ: correlation between monozygotic twins

rDZ: correlation between dizygotic twins

CI: 95% confidence interval

H: heritability

NS: non-significant

Table 2: Heritability estimates of aortic parameters.

Conclusion

In this classical twin study we have demonstrated that ascending aortic area and aortic diameter taper have a relatively strong heritability.

In order to quantify the relative influence of additive genetic, common environmental and unique environmental effects on aortic geometry larger sample size is required. By increasing the number of study participants will allow us to set up structural equation models and apply maximum-likelihood-based model-fitting algorithms.

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Personal information

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