Table S1: Demographic distribution and risk factors.

|  |  |  |  |
| --- | --- | --- | --- |
|  | | | |
| Parameter | Results  n = 95, study cohort | n = 716, totally screened with fulfilled inclusion / exclusion criteria | p-value |
|  | Mean ± standard deviation | |  |
| Age | 54.9 ± 12.0 | 59.2 ± 8.2 | 0.832 |
|  | Percentage, % | |  |
| Male sex | 70.5% | 68.7% | 0.385 |
| Aortic surgery | 68.4% | 72.1% | 0.153 |
| TEVAR | 31.6% | 27.9% | 0.278 |
| Main diagnosis TAD | 56.8% | 44.6 % | 0.012 |
| Main diagnosis TAA | 41.1% | 52.4% | 0.007 |
| **Cardiovascular risk factors** | | | |
| Arterial hypertension | 83.2% | 80.9% | 0.532 |
| (Ex-)nicotine | 33.7% | 35.1% | 0.874 |
| Hypercholesterolemia | 32.6% | 29.8% | 0.756 |
| Diabetes mellitus | 8.4% | 9.7% | 0.544 |
| CHD (coronary heart disease) | 13.7% | 12.3 % | 0.223 |
| Carotid stenosis or stroke | 5.3% | 6.1% | 0.178 |
| PAD (peripheral arterial  occlusive disease) | 4.2% | 3.6% | 0.159 |
| **Medications** | | | |
| Beta blockers | 94.7% | 92.8% | 0.656 |
| ACE inhibitors/AT1  receptor antagonists | 83.1% | 81.7% | 0.258 |
| Platelet aggregation inhibitors | 42.1% | 40.9% | 0.175 |
| Calcium antagonists | 41.1% | 39.2% | 0.288 |
| Diuretics | 40.0% | 42.4% | 0.731 |
| Oral anticoagulation | 36.8% | 29.7% | 0.021 |
| Other antihypertensive drugs | 21.1% | 22.3% | 0.410 |

Table S2: Distribution of phenotypical characteristics.

|  |  |
| --- | --- |
| **Family History** |  |
| Positive family history of connective tissue or aortic disease  In first- to third-degree relatives | 20% |
| 1st degree relatives with aortic dissection | 2.1% |
| 1st degree relatives with aortic aneurysm | 4.2% |
| **Stature** |  |
| Normal height | 91.6% |
| Normal Relationship from arm span to  body length | 85.3% |
| Ratio of arm span to  body length above to the normal range | 14.7% |
| Tall stature | 6.3% |
| Striae | 5.3% |
| Arachnodactyly | 3.2% |
| Short stature | 2.1% |
| Increased skin elasticity | 1.1% |
| **Skeletal deformities** |  |
| Pes planus | 20.0% |
| Scoliosis | 13.7% |
| Pectus carinatum | 6.3% |
| Pectus excavatum | 4.2% |
| Protrusion acetabuli | 1.1% |
| Pes equinovarus | 0% |
| **Craniofascial anomalies** |  |
| Thin lips | 25.3% |
| Higher palate | 14.7% |
| Dolichocephaly | 5.3% |
| Hypertelorism | 1.1% |
| Cleft palate | 0% |
| Craniosynostosis | 0% |
| Midface hypoplasia | 0% |
| Micrognathia | 0% |
| Uvula bifida | 0% |
| **Joint function** |  |
| Murdoch sign | 9.5% |
| Joint dislocations | 5.3% |
| Steinberg sign | 4.2% |
| Beighton Score 1 | 4.2% |
| Beighton Score 4 | 2.1% |
| Beighton Score 2 | 1.1% |
| **Ophthalmic pathologies** |  |
| Myopia ≥ 3 diopters | 14.7% |
| Enophthalmos | 11.6% |
| Cataract | 7.4% |
| Blue sclera | 7.4% |
| Glaucoma | 5.3% |
| Ablation retinae | 0% |
| Iris hypoplasia | 0% |
| Ectopia lentis | 0% |
| **General diagnoses** |  |
| Hernias | 23.2% |
| Spontaneous pneumothorax | 0% |
| Dura ectasia | 0% |
| **Cardiovascular** |  |
| Bicuspids aortic valve | 17.9% |
| Atrial septal defect | 4.2% |

Figure S1: Distribution of pathology according to age group.

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Figure S2: Distribution of total phenotype score.

**Distribution of total phenotype score**

24.2%

25

15.8%

16.8%

20

13.7%

12.6%

15

9.5%

10

5.3%

5

1.1%

1.1%

,0

0 1 2 3 4 5 6 7 8 9 10 11 12 13

Total score from summed points

Percentage of study cohort