



## CLINICAL CHECKLIST AORTIC ANEURYSM

**FOR AORTIC ANEURYSM SCREENING ONE LABORATORY REQUEST AND ONE CLINICAL CHECKLIST SHOULD BE COMPLETED PER PERSON.**

**These forms may be requested by phone or downloaded from the website.**

### Patient information

Name (first, last name) : \_\_\_\_\_  
DOB (dd/mm/yr) : \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_  
Sex : M / F  
Height : \_\_\_\_\_ cm    Weight : \_\_\_\_\_ kg    Blood pressure : \_\_\_\_\_ / \_\_\_\_\_ mmHg  
Referring physician : \_\_\_\_\_  
Referring center : \_\_\_\_\_

#### Sample type :

- EDTA-blood
- DNA
- fibroblasts
- vascular smooth muscle cells
- skin biopsy
- aortic biopsy (physiological medium)
- aortic biopsy (frozen)
- aortic biopsy (paraffin embedded)
- aortic valve (physiological medium)
- aortic valve (frozen)
- aortic valve (paraffin embedded)

Gene defect (if known) : Gene : \_\_\_\_\_ Mutation : \_\_\_\_\_

### Clinical diagnosis

- Marfan syndrome
- Loeys-Dietz syndrome
- vascular Ehlers-Danlos syndrome
- arterial tortuosity syndrome
- Thoracic aortic aneurysm or dissection (TAAD)
  - sporadic case
  - positive family history
- TAAD with patent ductus arteriosus (PDA)
- BAV without aortic aneurysm (BAV only)
  - sporadic case
  - positive family history
- BAV with aortic aneurysm (BAV/TAA)
  - sporadic case
  - positive family history
- other diagnosis : \_\_\_\_\_

Name patient (please complete) : .....

## Phenotypic features (1)

**Cardiovascular** (echo date : \_\_\_\_ / \_\_\_\_ / \_\_\_\_)

### Aortic measurements

Aortic valve annulus : \_\_\_\_\_ mm  
Aortic root (sinus Valsalva) : \_\_\_\_\_ mm  
Sinotubular junction : \_\_\_\_\_ mm  
Ascending aorta : \_\_\_\_\_ mm  
Aortic arch : \_\_\_\_\_ mm  
Descending aorta : \_\_\_\_\_ mm

### Current medications :

Aortic dissection : at age \_\_\_\_\_ yrs

0 Type A

0 Type B

Prior dilation : Y / N / Unknown

Aortic surgery : at age \_\_\_\_\_ yrs

Aortic tortuosity : Y / N / Unknown

Aortic valve : 0 tricuspid 0 bicuspid

0 stenotic - gradient : \_\_\_\_\_ mmHg

0 insufficiency : grade 0 – 1 – 2 – 3 (none-mild-moderate-severe)

Mitral valve : 0 mitral valve prolapse : Y / N / Unknown

0 stenotic - gradient : \_\_\_\_\_ mmHg

0 insufficiency : grade 0 – 1 – 2 – 3 (none-mild-moderate-severe)

Main pulmonary artery :

0 normal

0 dilatation (diameter : \_\_\_\_\_ mm)

0 stenotic : 0 truncal

0 peripheral

Patent ductus arteriosus : Y / N - surgery at age : \_\_\_\_\_ yrs

Atrial septum defect : Y / N - surgery at age : \_\_\_\_\_ yrs

Ventricular septum defect : Y / N - surgery at age : \_\_\_\_\_ yrs

Other congenital heart defect : Y / N / Unknown, if yes : type : \_\_\_\_\_

Other arterial aneurysms : Y / N / Unknown, if yes : location : \_\_\_\_\_

Other arterial dissections : Y / N / Unknown, if yes : location : \_\_\_\_\_

Other arterial stenosis : Y / N / Unknown, if yes : location : \_\_\_\_\_

Other arterial tortuosity : Y / N / Unknown, if yes : location : \_\_\_\_\_

## Craniofacial

Age : \_\_\_\_ Outer canthal : \_\_\_\_ cm Inner canthal : \_\_\_\_ cm Interpupillary : \_\_\_\_ cm

Downslanting palpebral fissures : Y / N / NE

Hypertelorism : Y / N / NE Blepharophimosis : Y / N / NE

Post. rotated ears : Y / N / NE Crumpled ears : Y / N / NE

Cleft palate : Y / N / NE High palate : Y / N / NE

Bifid uvula : Y / N / NE Broad uvula : Y / N / NE

Malar hypoplasia : Y / N / NE Beaked nose : Y / N / NE

Retrognathia : Y / N / NE Micrognathia : Y / N / NE

Dolichocephaly : Y / N / NE Long face : Y / N / NE

Craniosynostosis : Y / N - closed sutures : \_\_\_\_\_ Cranial asymmetry : Y / N

Other : \_\_\_\_\_

## Ophthalmological

Exotropia : Y / N / NE - surgery at age : \_\_\_\_\_

Esotropia : Y / N / NE - surgery at age : \_\_\_\_\_

Proptosis : Y / N / NE

Blue sclerae : Y / N / NE

Lens (sub)luxation : Y / N / NE Scleral/corneal fragility : Y / N / NE

Retinal detachment : Y / N / NE Tortuosity retinal arteries : Y / N / NE

Keratoconus/globus : Y / N / NE Retinal degeneration : Y / N / NE

Myopia : Y / N / NE - diopters : L : - \_\_\_\_\_ R : - \_\_\_\_\_

Other : \_\_\_\_\_

Name patient (please complete) : .....

## Phenotypic features (2)

### Skeletal

Age : \_\_\_\_ Height : \_\_\_\_\_ cm Armspan : \_\_\_\_\_ cm US : \_\_\_\_\_ cm LS : \_\_\_\_\_ cm  
Armspan/height : \_\_\_\_\_ US/LS : \_\_\_\_\_

Arachnodactyly :	Y / N / NE	Joint hypermobility :	Y / N / NE
Wrist sign :	Y / N / NE	Beighton score :	____ / 9
Thumb sign :	Y / N / NE	Thumb abduction :	L / R
Camptodactyly :	Y / N / NE	Finger extension :	L / R
Clinodactyly :	Y / N / NE	Elbow extension :	L / R
Flat feet :	Y / N / NE	Knee extension :	L / R
Club feet :	Y / N / NE	Hands on ground :	Y / N
Pectus excavatum :	Y / N / NE	Pectus carinatum :	Y / N / NE
Joint contractures :	Y / N / NE - joints : elbow, shoulder, knee, other : _____		
Scoliosis :	Y / N - surgery at age : _____		
Cervical instability :	Y / N / NE	Kyphosis :	Y / N / NE
Spondylolisthesis :	Y / N / NE	Osteoporosis :	Y / N / NE
Osteo-arthritis :	Y / N / NE - location : _____		
Protrusio acetabuli :	Y / N / NE		
Other :	_____		

### Skin

Hyperextensibility :	Y / N	Velvety texture :	Y / N	Translucency :	Y / N
Striae :	Y / N	Delayed wound healing :	Y / N	Easy bruising :	Y / N
Widened, atrophic scars :	Y / N - if yes, location : _____				
Herniae :	Y / N - type : _____				

### Other organ systems

Pneumothorax :	Y / N / unknown	Lung emphysema :	Y / N / unknown
Dural ectasia :	Y / N / unknown	Gastro-intestinal rupture :	Y / N / unknown
Uterine rupture :	Y / N / unknown	Intestinal diverticulae :	Y / N / unknown
Acute myocardial infarct :	Y / N / unknown	Cerebrovascular infarction :	Y / N / unknown

## Pedigree

Consanguinity : Y / N / unknown - if yes : degree : \_\_\_\_\_