Center N°	Center number where the patient is monitored	
Name of the center	Name of the center where the patient is monitored	
Investigator	The investigator who recruited the patient into the study	
Patient ID	Patient ID created automatically thanks to the center N°, the first letter of the first name and first letter of the patient's name.	
HTAD identifier	HTAD identifier	
National rare disease identifier	National rare disease identifier	
National anonymous identifier	National anonymous identifier	
National health identifier	National health identifier	
EUPID identifier	EUPID Pseudonym of the patient	
Date of consent	Date on which the patient signed the consent	DD/MM/YYYY
Last Name	First letter of the patient's last name	
First Name	First letter of the patient's first name	
Date of birth	Patient's date of birth	MM/YYYY
Sex	Patient's sex at birth	Male Female Unknown Foetus unknown
Patient's status	Patient alive, deceased, opted-out or lost. Automatically filled in according to the cessation of participation form.	Alive Dead Lost in follow-up Opted-out
Date of general patient information	Date of general patient information	Yes, No

Information and non-opposition of the participant to the collection of his health data in the Marfan HTAD database	Information and non-opposition of the patient to the collection of his health data in the Marfan HTAD database	Yes, No
Availability of biological samples	Agreement for the reuse of biological samples (carried out exclusively within the framework of treatment) in the context of subsequent research	Yes, No
Biological sample stored in a biobank	Agreement for the conservation of biological samples (carried out exclusively within the framework of treatment) in a biobank	Yes, No
No objection to be contacted for research purposes	Information provided and no objection of the patient to be contacted for a later research objective	Yes, No
No objection to the reuse of health data in subsequent research projects	Information provided and no objection by the patient to the reuse of their health data in the context of subsequent research projects	Yes, No
Contact method	Contact method to be used to contact the patient in the context of subsequent research projects	E-mail
		Phone
		Postal email
Written and signed consent of the patient for the re-use of genetic data for further research	Written and signed consent of the patient for the re-use of genetic data for further research	Yes, No
Written and signed consent of the patient for the reuse of biological samples (carried out exclusively as part of the treatment) in the context of other genetic research	Written and signed consent of the patient for the reuse of biological samples (carried out exclusively as part of the treatment) in the context of other genetic research	Yes, No
Reason for the 1st consultation	The reason for the patient's first consultation	Aortic dissection
		Aortic aneurysm
		Aneurysm/dissection non aortic
		Bicuspid aortic valve
		Mitral surgery
		Mitral vavle prolapse
		Other cardiovascular
		Ophtalmology
		Skeletal
		Pneumothorax
		On his own
		Family history (parent with clear diagnosis)
		Family history (parent with dissection or sudden death)
		Unknown
		Other
Date of first visit	The date of the patient's first visit	DD/MM/YYYY
Clinical diagnosis by expert center	Clinical diagnosis of the patient provided by the expert center	Marfan Syndrome (Ghent 2)
		Loeys Dietz (syndromic)
		Aortic aneurysm/dissection
		Non aortic aneurysm/dissection
		BAV without aneurysm
		Isolated MVP

		Beals (CCA)
		Arterial tortuosity syndrome (ATS)
		Ectopia lentis syndrome
		FBLN4/EFEMP2 related cutis laxa
		Vascular Ehlers Danlos
		Coronary artery dissection
		ELN related cutis laxa
		HCM
		DCM
		LQTS
		No diagnosis
		unaffected
		Other
Other, in clear	Clinical diagnosis of the patient provided by the expert center (except the following diseases: Marfan Syndrome (Ghent 2), Loeys Dietz (syndromic), Aortic aneurysm/dissection, Non aortic aneurysm/dissection, BAV without aneurysm, Isolated MVP, Beals (CCA), Arterial tortuosity syndrome (ATS), Ectopia lentis syndrome, FBLN4/EFEMP2 related cutis laxa, Vascular Ehlers Danlos, Coronary artery dissection, ELN related cutis laxa, HCM, DCM, LQTS)	
Undiagnosed case	Phenotype (HPO) or Genotype (HGVS) in the case the patient is waiting for diagnoses.	Phenotype (HPO)
		Genotype (HGVS)
Rare disease diagnosis (orphanet code)	Orpha code of the rare disease's diagnosis.	
Diagnosis before arrival (BNDMR)	Status of the patient's diagnosis before the consultation	No diagnosis
		Wrong diagnosis
		Diagnosis made
Period of diagnosis	Period in the patient's life when the diagnosis was made	Antenatal
		At birth
		Postnatal
		Undetermined
Age at diagnosis	Patient's age at diagnosis. Automatically filled in according to the period of diagnosis and the date of clinical diagnosis	
Date of clinical diagnosis	Date of clinical diagnosis	DD/MM/YYYY
Date of molecular diagnosis	Date of molecular diagnosis	DD/MM/YYYY
Diagnosis status (BNDMR)	Status of the current patient's diagnosis	Ongoing
		Probable
		Confirmed
		Undetermined
		Not classifiable
Mode of diagnosis (BNDMR)	Mode of diagnosis established for the patient	Caryotype, FISH
		Array-CGH

		Sanger
		NGS
		Other
Period of first symptoms	Period in the patient's life when first symptoms appeared	Antenatal
		At birth
		Postnatal
		Undetermined
Age at onset	Age of onset of the first symtoms. Automatically filled in according to the period of symptoms and the date at first symptoms	
Date at first symptom	Date of onset of patient's first symptoms	DD/MM/YYYY
Commentary	Free field for adding comments on the patient's diagnosis	
Family number in the clinical center	Family number in the clinical center	
Familial form	The patient's disease comes from a familial form	Yes
		No
		Unknown
Family history	Family history of aortic dissection or aortic aneurysm	Dissection
		Aneurysm
Transmission	Paternal, maternal transmission or neomutation	Father
		Mother
		Neomutation
		Unknown
Proband	is the patient the proband	Yes
		No
Identifier (BNDMR)	Proband's identifier	
Relation with proband (BNDMR)	family relation of the patient with the proband	Brother
		Sister
		Mother
		Father
		Grandfather
		Grandmother
DNA available	A genetic sample was taken from the patient	Yes
		No
		Unknown
Date of DNA collection	Date of DNA collection	DD/MM/YYYY
Family N° (lab)	Family number in the laboratory	
Genetic testing	is a genetic testing done in the patient	Performed
		Ongoing
		No
Variant present	A genetic variant was found in the patient	Pathogenic (Classe 4/5)
		VUS (Classe 3)
		None
Genes	Gene identified in the patient	FBN1

		TGFBR1
		TGFBR2
		SMAD2
		SMAD3
		TGFB2
		TGFB3
		ACTA2
		MYH11
		MYLK
		PRKG1
		LOX
		FBN2
		SLC2A10
		FBLN4
		NOTCH1
		SMAD6
		Other
Genetic Diagnosis (HGVS)	Mutation in the patient's DNA. Please use the International classification of mutations (HGVS)	
Mutation protein	Patient's protein mutation.	
Type of mutation	Type of mutation	PTC
		Inframe
		Large rearrangements
If PTC, specify	If PTC, specify	Deletion
		Insertion
		Del/Ins
		Duplication
		Nonsense
		Splicing
If Inframe, specify	If Inframe, specify	Cysteine loss
		Addition of cysteine
		No modification in cysteine content
If large rearrangements, specify	If large rearrangements, specify	Large deletion
		Large duplication
Exons	Exon number	
2nd gene variant	Presence of a second gene variant in the patient	Yes
		No
Genes	Second gene identified in the patient	FBN1
		TGFBR1
		TGFBR2
		SMAD2
		SMAD3

		TGFB2
		TGFB3
		ACTA2
		MYH11
		MYLK
		PRKG1
		LOX
		FBN2
		SLC2A10
		FBLN4
		NOTCH1
		SMAD6
		Other
Genetic Diagnosis (HGVS)	Second mutation in the patient's DNA. Please use the International classification of mutations (HGVS)	
Mutation protein	The patient's second protein mutation	
Type of mutation	Type of mutation	PTC
		Inframe
		Large rearrangements
If PTC, specify	If PTC, specify	Deletion
		Insertion
		Del/Ins
		Duplication
		Nonsense
		Splicing
If Inframe, specify	If Inframe, specify	Cysteine loss
		Addition of cysteine
		No modification in cysteine content
If large rearrangements, specify	If large rearrangements, specify	Large deletion
		Large duplication
Exons	Exon number	
Date of last follow-up	Date of last ophthalmologic follow-up	DD/MM/YYYY
Myopia	The patient has myopia in the right eye	Absent
		Moderate
		Important
		Unknown
Ectopia lentis	The patient has ectopia lentis in the right eye	Absent
		Doubtful
		Presence
		Unknown
Date of diagnosis	Date of diagnosis of ectopia lentis in the patient's right eye	DD/MM/YYYY

Age	Age of diagnosis of ectopia lentis in the patient's right eye. Automatically calculated using the date of birth and the date of diagnosis	
Aphakia	lens removed in the right eye	Yes
		No
Date of ablation	Date of removal of the lens of the right eye	DD/MM/YYYY
Age	Age of removal of the lens of the right eye. Automatically calculated using the date of birth and the date of ablation	
Lens implant	The patient has a lens implant in the right eye	Yes
		No
Date of implant placement	Date of implant placement in the patient's right eye.	DD/MM/YYYY
Age	Age of implant placement in the patient's right eye. Automatically calculated using the date of birth and the date of implant placement.	
Cataract	The patient has the cataract in the right eye	Yes
		No
Date of surgery	Date of cataract surgery on the patient's right eye	DD/MM/YYYY
Age	Age of cataract surgery on the patient's right eye. Automatically calculated using the date of birth and the date of surgery.	
Retinal detachment	The patient has a retinal detachment in the right eye	Yes
		No
Date of first detachment	Date of retinal detachment in the patient's right eye	DD/MM/YYYY
Age	Age of retinal detachment in the patient's right eye. Automatically calculated using the date of birth and the date of retinal detachment.	
Iris flocculi	The patient has the iris flocculi in the right eye	Yes
		No
Myopia	The patient has myopia in the left eye	Absent
		Moderate
		Important
		Unknown
Ectopia lentis	The patient has the ectopia lentis in the left eye	Absent
		Doubtful
		Presence
		Unknown
Date of diagnosis	Date of diagnosis of ectopia lentis in the patient's left eye	DD/MM/YYYY
Age	Age of diagnosis of ectopia lentis in the patient's left eye. Automatically calculated using the date of birth and the date of diagnosis	
Aphakia	The patient has the aphakia in the left eye	Yes
		No

Date of ablation	Date of removal of the lens of the left eye	DD/MM/YYYY
Age	Age of removal of the lens of the left eye. Automatically calculated using the date of birth and the date of ablation	
Lens implant	The patient has a lens implant in the left eye	Yes
		No
Date of implant placement	Date of implant placement in the patient's left eye	DD/MM/YYYY
Age	Age of implant placement in the patient's left eye. Automatically calculated using the date of birth and the date of implant placement.	
Cataract	The patient has the cataract in the left eye	Yes
		No
Date of surgery	Date of cataract surgery on the patient's left eye	DD/MM/YYYY
Age	Age of cataract surgery on the patient's left eye. Automatically calculated using the date of birth and the date of surgery.	
Retinal detachment	The patient has a retinal detachment in the left eye	Yes
		No
Date of first detachment	Date of retinal detachment in the patient's left eye	DD/MM/YYYY
Age	Age of retinal detachment in the patient's left eye. Automatically calculated using the date of birth and the date of retinal detachment.	
Iris flocculi	The patient has the iris flocculi in the left eye	Yes
		No
Date of last follow-up	Date of last follow-up of the patient's cardiological risk factors	DD/MM/YYYY
Treated HT (Hypertension)	The patient is being treated for hypertension	Yes
		No
Number of medicines	Number of treatments taken by the patient for hypertension	
Treated hypercholesterolemia	The patient is being treated for hypercholesterolemia	Yes
		No
Treated Diabetes	The patient is being treated for diabetes	Yes
		No
Type	Type of patient's diabetes	I
		II
Insulin	The patient takes insulin treatment to regulate his level of diabetes	Yes
		No
Tabacco, past or present	Is the patient a smoker or former smoker?	Yes
		No
Exercise: hours/week	Number of hours per week of sport practiced by the patient	
Type of exercise practiced	Type of exercise practiced by the patient	Isometic
		Endurance
		Both

Date of last follow-up	Date of last follow-up of the patient's cardiological complications	DD/MM/YYYY
Complications	The patient has suffered cardiological complications	Yes
		No
		Unknown
First Aortic dissection	The patient had to perform a first aortic dissection	Yes
		No
		Unknown
If yes	Type of first aortic dissection performed on the patient	Ascending aorta
		Ascending aorta + arch
		Ascending aorta + arch + descinding
		Descending thoracic + abdominal
		Abdominal aorta alone
Date	Date of the first aortic dissection performed on the patient	DD/MM/YYYY
Age	Age of patient at first aortic dissection. Automatically calculated using the date of birth and the date of first aortic dissection	
Second Aortic dissection	The patient had to perform a second aortic dissection	Yes
		No
		Unknown
If yes	Type of second aortic dissection performed on the patient	Ascending aorta
		Ascending aorta + arch
		Ascending aorta + arch + descinding
		Descending thoracic + abdominal
		Abdominal aorta alone
Date	Date of the second aortic dissection performed on the patient	DD/MM/YYYY
Age	Age of patient at second aortic dissection. Automatically calculated using the date of birth and the date of second aortic dissection	
Third Aortic dissection	The patient had to perform a third aortic dissection	Yes
		No
		Unknown
If yes	Type of third aortic dissection performed on the patient	Ascending aorta
		Ascending aorta + arch
		Ascending aorta + arch + descinding
		Descending thoracic + abdominal
		Abdominal aorta alone
Date	Date of the third aortic dissection performed on the patient	DD/MM/YYYY
Age	Age of patient at third aortic dissection. Automatically calculated using the date of birth and the date of third aortic dissection	
First Ascending aortic surgery	The patient had to perform a first ascending aortic surgery	Yes
		No
		Unknown

Date	Date of the first ascending aortic surgery performed on the patient	DD/MM/YYYY
Age	Age of patient at the first ascending aortic surgery. Automatically calculated using the date of birth and the date of first ascending aortic surgery	
Reason for surgery	Reason for the first ascending aortic surgery	Aortic dilatation
		Aortic dissection
		Valvular heart disease
Last aortic diameter known	Last aortic diameter known	
Type of aortic surgery	Type of aortic surgery	Supra coronary
		Coronary implantation
Valve surgery type	Valve surgery type	Valve sparing
		Valve replaced
Type of valvular prothesis	Type of valvular prothesis	Bioprothesis
		Mecannical valve
Second Ascending aortic surgery	The patient had to perform a second ascending aortic surgery	Yes
		No
		Unknown
Date	Date of the second ascending aortic surgery performed on the patient	DD/MM/YYYY
Age	Age of patient at the second ascending aortic surgery. Automatically calculated using the date of birth and the date of second ascending aortic surgery	
Reason for surgery	Reason for the second ascending aortic surgery	Aortic dilatation
		Aortic dissection
		Valvular heart disease
Last aortic diameter before surgery	Last aortic diameter before surgery	
Type of aortic surgery	Type of aortic surgery	Supra coronary
		Coronary implantation
Valve surgery type	Valve surgery type	Valve sparing
		Valve replaced
Type of valvular prothesis	Type of valvular prothesis	Bioprothesis
		Mecannical valve
Surgery of cross	The patient had to perform a cross surgery	Yes
		No
		Unknown
Date	Date of the cross surgery performed on the patient	DD/MM/YYYY
Age	Age of the patient at the time of the cross surgery. Automatically calculated using the date of birth and the date of cross surgery	
Surgery type	Type of cross surgery performed on the patient	Thoracotomy
		Endoprothesis
Elephant trunk	Was elephant technique used in the patient ?	Yes
		No

First Descending aortic surgery	a first descending aortic surgery was peformed	Yes
		No
		Unknown
Date	Date of the first descending aortic surgery performed on the patient	DD/MM/YYYY
Age	Age of patient at the first descending aortic surgery. Automatically calculated using the date of birth and the date of first descending aortic surgery	
Surgery type	Type of first descending aortic surgery performed on the patient	Thoracotomy
		Endoprothesis
Second Descending aortic surgery	The patient had to perform a second descending aortic surgery	Yes
		No
		Unknown
Date	Date of the second descending aortic surgery performed on the patient	DD/MM/YYYY
Age	Age of patient at the second descending aortic surgery. Automatically calculated using the date of birth and the date of second descending aortic surgery	
Surgery type	Type of second descending aortic surgery performed on the patient	Thoracotomy
		Endoprothesis
First Mitral surgery	The patient had to perform a first mitral surgery	Yes
		No
		Unknown
Date	Date of the first mitral surgery performed on the patient	DD/MM/YYYY
Age	Age of patient at the first mitral surgery. Automatically calculated using the date of birth and the date of first mitral surgery	
Surgery type	Type of first mitral surgery performed on the patient	Mitral valvuloplasty
		Mechanical valve
		Bioprothesis
Second Mitral surgery	The patient had to perform a second mitral surgery	Yes
		No
		Unknown
Date	Date of the second mitral surgery performed on the patient	DD/MM/YYYY
Age	Age of patient at the second mitral surgery. Automatically calculated using the date of birth and the date of second mitral surgery	
Surgery type	Type of second mitral surgery performed on the patient	Mitral valvuloplasty
		Mechanical valve
		Bioprothesis
Congenital cardiopathy	The patient has a congenital cardiopathy	Yes
		No
		Unknown
Atrial Septal Defect	The patient has an atrial septal defect	Yes

		No
		Unknown
Date	Date of diagnosis of the patient's atrial septal defect	DD/MM/YYYY
Age	Patient's age at diagnosis of atrial septal defect. Automatically calculated using the date of birth and the date of diagnosis	
Mode of care	Mode of care of the patient's atrial septal defect	Surgery
		Endovascular
		No treatment
Ventricular Septal Defect	The patient has a ventricular septal defect	Yes
		No
		Unknown
Date	Date of diagnosis of the patient's ventricular septal defect	DD/MM/YYYY
Age	Patient's age at diagnosis of ventricular septal defect. Automatically calculated using the date of birth and the date of diagnosis	
Mode of care	Mode of care of the patient's ventricular septal defect	Surgery
		Endovascular
		No treatment
Aortic Coarctation	The patient has an aortic coarctation	Yes
		No
		Unknown
Date	Date of diagnosis of the patient's aortic coarctation	DD/MM/YYYY
Age	Patient's age at diagnosis of aortic coarctation. Automatically calculated using the date of birth and the date of diagnosis	
Mode of care	Mode of care of the patient's aortic coarctation	Surgery
		Endovascular
		No treatment
Patent Ductus Arteriosus	The patient has a patent ductus arteriosus	Yes
		No
		Unknown
Date	Date of diagnosis of the patient's patent ductus arteriosus	DD/MM/YYYY
Age	Patient's age at diagnosis of the patent ductus arteriosus. Automatically calculated using the date of birth and the date of diagnosis	
Mode of care	Mode of care of the patient's patent ductus arteriosus	Surgery
		Endovascular
		No treatment
Heart transplantation	The patient has a heart transplantation	Yes
		No
		Unknown
Date	Date of heart transplantation	DD/MM/YYYY

Age	Patient's age at the heart transplantation. Automatically calculated using the date of birth and the date of heart transplantation	
Date of last follow-up	Date of last follow-up of the patient's peripheral vascular disease	DD/MM/YYYY
Peripheral vascular disease	The patient has a peripheral vascular disease	Yes
		No
		Unknown
Location	The physical location of the peripheral vascular disease	Cerebral
		Carotid
		Vertebral
		Subclavian
		Axillary
		Mesenteric
		Celiac
		Renal
		Hepatic
		Splenic
		Iliac
		Femoral
		Popliteal
		Other
Side	The side of the peripheral vascular disease	Left
		Right
Type of events	Type of peripheral vascular event	Aneurysm
		Dissection
		Rupture
		Arterial tortuosity
		MoyaMoya type
Date of diagnosis	Date of diagnosis of the peripheral vascular event	DD/MM/YYYY
Imaging technique	Imaging technique used to visualise the peripheral vascular disease	Echography
		CT scanner
		MRI
Surgery	Surgery performed on the patient for the treatment of the peripheral vascular disease	Yes
		No
		Unknown
Mode of surgery	Mode of surgery performed on the patient for the treatment of the peripheral vascular disease	Classic surgery
		Percutaneous surgery
Date of surgery	Date of surgery for peripheral vascular disease	DD/MM/YYYY
Stroke	The patient has suffered a stroke	Yes
		No
		Unknown
Commentary	Free field for adding comments on the patient's peripheral vascular disease	

Treatments	Type of treatment taken by the patient to solve heart problems	Betablocker
		ARA2
		ACEI
		Calcium blocker
		Diuretic including aldactone
		Statins
		Anticoagulation
		Aspirin
If anticoagulants, specify	Type of anticoagulant taken by the patient	VKA
		NOAC
Start date	Start date of treatment	DD/MM/YYYY
Ongoing	Ongoing treatment	Yes
		No
If no, end date :	End date of treatment	DD/MM/YYYY
Date	Date of cardiology examination	DD/MM/YYYY
Height (cm)	Patient's height (cm)	
Weight (kg)	Patient's weight (kg)	
Body surface area	Body surface area with the Dubois and Dubois formula : 0,007184 x Height (cm)^0,725 x Weight (kg)^0,425. Automatically calculated using the patient's height and weight	
Systolic BP (mmHg)	Patient's systolic BP (mmHg)	
Diastolic BP (mmHg)	Patient's diastolic BP (mmHg)	
Sinus rhythm	Sinus rhythm visualised with an electrocardiogram/Holter	Yes
		No
		Unknown
Heart rate (bpm)	Heart rate calculated with an electrocardiogram/Holter	
Holter available	The cardiac holter device was used on the patient	Yes
		No
		Unknown
Aortic valve	Aortic valve structure of the patient visualised by echocardiography	Tricuspid
		Bicuspid
		Unknown
		Bioprothesis
		Mechanical
		Valvular plasty
If BAV	Number and type of raphe of the patient's bicupid aortic valve visualized by echocardiography	0 raphe, vertical opening
		0 raphe, horizontal opening
		1 raphe, LR
		1 raphe, RN
		1 raphe, LN
		2 raphe

Mean gradient (mmHg)	Mean gradient at the aortic valve of the patient with an aortic bioprosthesis or a mechanical aortic valve	
Max velocity (m/sec)	Maximum aortic valve velocity of the patient with an aortic bioprosthesis or a mechanical aortic valve	
Aortic regurgitation	Importance of the aortic regurgitation in + (from 0 to 4+)	0
		1+
		2+
		3+
		4+
Telediastolic velocity in the arch	Telediastolic velocity in the arch of the patient with a 1+, 2+, 3+ or 4+ aortic regurgitation	
Aortic annulus (mm)	Patient's aortic annulus diameter (mm) calculated by echocardiography	
Sinus of Valsalva (mm)	Patient's sinus of Valsalva diameter (mm) calculated by echocardiography	
Sinotubular junction (mm)	Patient's sinotubular junction diameter (mm) calculated by echocardiography	
Ascending aorta (mm)	Patient's ascending aorta diameter (mm) calculated by echocardiography	
Aorta arch (before subclavian)(mm)	Patient's aorta arch diameter (before subclavian)(mm) calculated by echocardiography	
Descending thoracic aorta (mm)	Patient's descending thoracic aorta diameter (mm) calculated by echocardiography	
Abdominal aorta (mm)	Patient's abdominal aorta diameter (mm) calculated by echocardiography	
Pulmonary artery (mm)	Patient's pulmonary artery diameter (mm) calculated by echocardiography	
Myxoid aspect	Presence of a myxoid aspect of the patient's mitral valve observed on echocardiography	Yes
		No
		Unknown
Prolapse	The type of mitral valve prolapse of the patient observed on echocardiography	Normal
		Ballooning
		Prolapse
Mitral regurgitation	Importance of the mitral valve regurgitation in + (from 0 to 4+)	0
		1+
		2+
		3+
		4+
LVEDD (mm)	Left ventricular end-diastolic diameter (mm) calculated by echocardiography	
LVESD (mm)	Left ventricular end-systolic diameter (mm) calculated by echocardiography	
LVEF Simpson (%)	Left Ventricular Ejection Fraction by Simpson's method	

LVEF Teicholz (%)	Left Ventricular Ejection Fraction by Teicholz's method. Automatically calculated using the LVEDD and LVESD. Formula : [7 LVEDD^3/(2.4+LVEDD) - 7 LVESD^3/(2.4+LVESD)]/ [7 LVEDD^3/(2.4+LVEDD)] * 100	
Patent Ductus Arteriosus	Presence of a persistence in the patient's ductus arteriosus	Yes No
Other	Free field for adding comments on the patient's echocardiography	
According to Campens	Calculation of the patient's valsava Z-score from echocardiography according to campens. Automatically calculated using the patient's age, the sinus valsava diameter, height and weight. Formula : Man : (Lg10(SoV) - (1.108 + 0.136 x Lg10(Age) + 0.099 x BSA DB))/0.0381 Woman : (Lg10(SoV) - (1.100 + 0.129 x Lg10(Age) + 0.091 x BSA DB))/0.0421 BSA DB = BSA calculated with the Dubois and Dubois formula : 0,007184 x Height (cm)^0,725 x Weight (kg)^0,425 SoV = sinuses of Valsalva.	
According to Roman	Calculation of the patient's valsava Z-score from echocardiography according to Roman. Automatically calculated using the patient's age, the sinus valsava diameter, height and weight. Formula : <20 years : Z = (measured root diameter - 1.02+0.98 x BSA)/ 0.18 20-40 : Z = (measured root diameter - 0.97 + 1.12 x BSA)/ 0.24 >40 : Z = (measured root diameter - 1.92 + 0.74 × BSA)/0.37 BSA calculated with the Dubois and Dubois formula : 0,007184 x Height (cm)^0,725 x Weight (kg)^0,425	
According to Gautier	Calculation of the patient's valsava Z-score from echocardiography according to Gautier. Automatically calculated, for patients under 18 years, using the patient's age, the sinus valsava diameter, height and weight. Formula : Man : Z=[ln(d) - 3.1 - 0.49 x ln(BSA)]/0.1 Woman : Z=[ln(d) - 3.1 - 0.44 x ln(BSA)]/0.09 BSA calculated with the Dubois and Dubois formula : 0,007184 x Height (cm)^0,725 x Weight (kg)^0,425	

According to Devereux	Calculation of the patient's valsava Z-score from echocardiography according to Devereux. Automatically calculated, for patients 15 years and over using the patient's age, the sinus valsava diameter, height and weight. Formula : Man : Z = (diameter of the sinus of Valsava - (2.423 + (age x 0.009) + (BSA x 0.461) - (1 x 0.267)))/0.261 Woman : Z = (diameter of the sinus of Valsava - (2.423 + (age x 0.009) + (BSA x 0.461) - (2 x 0.267)))/0.261 with the diameter of the sinus of Valsava in cm BSA DB = BSA calculated with the Dubois and Dubois formula : 0,007184 x Height (cm)^0,725 x Weight (kg)^0,425	
According to Devereux on size only	Calculation of the patient's valsava Z-score from echocardiography according to Devereux on size only. Automatically calculated using the patient's age, the sinus valsava diameter, height and weight. Z = (diameter of the sinus of Valsava - (1.519 + (age x 0.010) + (taille x 0.010) - (sex x 0.247)))/0.215 with sex =1 for men and sex = 2 for women. The diameter of the sinus of Valsava is in cm.	
According to Campens	Calculation of the patient's tubular aorta from echocardiography according to campens. Man : (Lg10(AA) - (1.033 + 0.188 x Lg10(Age) + 0.070 x BSA))/0.0431 Woman : (Lg10(AA) - (1.033 + 0.188 x Lg10(Age) + 0.070 x BSA))/0.0431 AA = diameter of the ascending aorta; BSA calculated with the Dubois and Dubois : 0,007184 x Height (cm)^0,725 x Weight (kg)^0,425	
According to Gautier	Calculation of the patient's tubular aorta from echocardiography according to Gautier. Homme : Z = [ln(d) − 2.9-0.46 × ln(BSA)]/0.11 Femme : Z = [ln(d) − 2.9-0.46 × ln(BSA)]/0.1 d = diameter of the ascending aorta; BSA calculated with the Dubois and Dubois : 0,007184 x Height (cm)^0,725 x Weight (kg)^0,425	
Other imaging studies performed since last consultation	The patient has had imaging (e.g. MRI or CT scanner) since the last consultation other than echocardiography or ECG/Holter .	Yes No
MRI	The patient has had an MRI since the last consultation	Yes No
Date	Date of the patient's MRI	DD/MM/YYYY
Ascending aorta (mm)	Patient's ascending aorta size (mm) calculated by MRI	
Aorta arch (before subclavian)(mm)	Patient's aorta arch (before subclavian) size (mm) calculated by MRI	

Descending thoracic aorta (mm)	Patient's descending thoracic aorta size (mm) calculated by MRI	
Abdominal aorta (mm)	Patient's abdominal aorta size (mm) calculated by MRI	
Diameter 1	Technique for measuring max diameters: Diameter perpendicular to the axis of the largest vessel, diameters at the level of the sinuses of valsalva are from cusp to cusp. Calculation of max diameter 1 from an MRI.	
Diameter 2	Technique for measuring max diameters: Diameter perpendicular to the axis of the largest vessel, diameters at the level of the sinuses of valsalva are from cusp to cusp. Calculation of max diameter 2 from an MRI.	
Diameter 3	Technique for measuring max diameters: Diameter perpendicular to the axis of the largest vessel, diameters at the level of the sinuses of valsalva are from cusp to cusp. Calculation of max diameter 3 from an MRI.	
CT scanner	The patient has had an CT scanner since the last consultation	Yes
		No
Date	Date of the patient's CT scanner	DD/MM/YYYY
Ascending aorta (mm)	Patient's ascending aorta size (mm) calculated by CT scanner	
Aorta arch (before subclavian)(mm)	Patient's aorta arch (before subclavian) size (mm) calculated by CT scanner	
Descending thoracic aorta (mm)	Patient's descending thoracic aorta size (mm) calculated by CT scanner	
Abdominal aorta (mm)	Patient's abdominal aorta size (mm) calculated by CT scanner	
Diameter 1	Technique for measuring max diameters: Diameter perpendicular to the axis of the largest vessel, diameters at the level of the sinuses of valsalva are from cusp to cusp. Calculation of max diameter 1 from an CT scanner.	
Diameter 2	Technique for measuring max diameters: Diameter perpendicular to the axis of the largest vessel, diameters at the level of the sinuses of valsalva are from cusp to cusp. Calculation of max diameter 2 from an CT scanner.	
Diameter 3	Technique for measuring max diameters: Diameter perpendicular to the axis of the largest vessel, diameters at the level of the sinuses of valsalva are from cusp to cusp. Calculation of max diameter 3 from an CT scanner.	
Date of examination	Date of orthopaedic rheumatological examination	DD/MM/YYYY
Height (cm)	Patient's height (cm)	
Weight (kg)	Patient's weight (kg)	

BMI	Patient's BMI (Body Mass Index). Automatically calculated using the patient's height and weight. Formula : weight/height² (kg/m)	
Z score / Height	Patient's Zscore/Height only for patients under 18 years of age	
Arm span (cm)	Patient's arm span (cm)	
Calculation (Arm span / Height)	Automatically calculated using the patient's height and arm span.	
Lower segment (cm)	Patient's lower segment (cm)	
Upper segment (cm)	Patient's upper segment (cm)	
Calculation (lower / upper segment) (cm)	Automatically calculated using the patient's upper and lower segment	
Date of last follow-up	Date of last follow-up of the patient's orthopaedic and rheumatological history	DD/MM/YYYY
Surgery for cervical instability	The patient had an operation for cervical instability	Yes
		No
		Unknown
Date	Date of the patient's operation for cervical instability	DD/MM/YYYY
Age	Age of the patient at the time of his operation for cervical instability. Automatically calculated using the patient's date of birth and the date of the surgery.	
Bone fracture	The patient had a bone fracture	Yes
		No
		Unknown
Recurrent sprains	The patient has recurrent sprains	Yes
		No
		Unknown
Recurring dislocations	The patient has recurrent dislocations	Yes
		No
		Unknown
Chronic fatigue	The patient has chronic fatigue	Yes
		No
		Unknown
Joint pain	The patient has joint pain	Yes
		No
		Unknown
Migraine	The patient has migraine	Yes
		No
		Unknown
Headache increased by orthostatism	The patient has headache increased by orthostatism	Yes
		No
		Unknown
Eosinophyllous esophagitis	The patient has eosinophyllous esophagitis	Yes
		No

		Unknown
Inflammatory bowel disease	The patient has a inflammatory bowel disease	Yes
		No
		Unknown
Food allergy	The patient has a food allergy	Yes
		No
		Unknown
Severe allergy	The patient has a severe allergy	Yes
		No
		Unknown
Cancer	Free field for adding comments on the patient's cancer	
Commentary	Free field for adding comments on the patient's orthopaedic and rheumatological history	
Date of last follow-up	Date of last follow-up of patient's limbs, thorax and face	DD/MM/YYYY
Pectus	The patient has a deformity of the thorax called pectus	No
		Mild
		Carinatum
		Excavatum
Surgery for pectus	The patient had surgery for his thoracic deformity (pectus)	Yes
		No
		Unknown
Date	Date of patient's pectus operation	DD/MM/YYYY
	The age of the patient at the time of his thoracic surgery for the treatment of pectus	
Thumb sign	The thumb is going beyond the auricular when the hand is closed	Yes
		No
		Unknown
Wrist sign	Ability to cross the last phalanger of the auricular and the last phalanger of the thumb across the wrist	Yes
		No
		Unknown
Beighton Score	Calculation of Score according to Beighton's rule	
Hyperlaxity	If the patient has a Beighton score greater than 5 and is under 18 years of age then the patient has hyperlaxity. If the patient has a Beighton score greater than 4 and is over 18 years of age then the patient has hyperlaxity. Otherwise he is not hyperlaxed. Automatically filled in using the patient's Beighton score and patient's age.	Yes
		No
		Unknown
Elbow extension degree	What is the degree of extension of the patient's elbows?	> 170°
		< 170°

Pes Planus	The patient has pes planus	No
		Yes
		Yes with hindfoot deformity
		Unknown
Camptodactyly	The patient has camptodactyly	Yes
		No
		Unknown
Facial dysmorphism	The patient has facial dysmorphism	Yes
		No
		Unknown
Dolichocephaly	The patient has dolichocephaly	Yes
		No
Downslanting palpebral fissures	The patient has downslanting palpebral fissures	Yes
		No
Enophthalmia	The patient has enophthalmia	Yes
		No
Retrognathia	The patient has retrognathia	Yes
		No
Malar hypoplasia	The patient has malar hypoplasia	Yes
		No
Hypertelorism	The patient has hypertelorism	Yes
		No
		Unknown
Blue sclerotic	The patient has blue sclerotic	Yes
		No
		Unknown
Dental malocclusion	The patient has dental malocclusion	Yes
		No
		Unknown
Ogival palate	The patient has ogival palate	Yes
		No
		Unknown
Craniosynostosis	The patient has craniosynostosis	Yes
		No
Uvula	Shape of the uvula	Normal
		Bifid
		Wide
		Wide with raphe
		Short
Osteoarthritis	The patient has osteoarthritis	Yes
		No
		Unknown
Date	Patient's osteoarthritis symptom onset date	DD/MM/YYYY

Hand	The patient has osteoarthritis in the hands	Yes
		No
Cervical	The patient has osteoarthritis in the cervical	Yes
		No
Lumbar rachis	The patient has osteoarthritis in the lumbar rachis	Yes
		No
Hip	The patient has osteoarthritis in the hip	Yes
		No
Knee	The patient has osteoarthritis in the knee	Yes
		No
Other	The patient has osteoarthritis elsewhere than in the knees, hands, cervical, lumbar rachis and hip	Yes
		No
Date of last follow-up	Date of last follow-up of patient's spine and hip	DD/MM/YYYY
Scoliosis	The patient has a scoliosis	Yes
		No
		Unknown
Angle	Angle of the patient's scoliosis	
Surgery	Surgery of the patient's scoliosis	Yes
		No
Date	Patient's scoliose surgery date	DD/MM/YYYY
Age	Age of the patient at the time of the scoliosis surgery	
Kyphosis	The patient has kyphosis	Yes
		No
		Unknown
Spondylolisthesis	The patient gas spondylolisthesis	Yes
		No
		Unknown
Acetabular protrusion (stage)	The patient has an acettabular protrusion (stage)	No
		Stage 1
		Stage 2 and +
		Unknown
Date of examination	Date of patient's skin and integument examination	DD/MM/YYYY
Stretch marks	The patient has stretch marks	None
		Stretch marks
		Micro striae
Shoulders	Stretch marks or micro striae are on the patient's shouder	Yes
		No
Breast	Stretch marks or micro striae are on the patient's breast	Yes
		No
Belly	Stretch marks or micro striae are on the patient's belly	Yes
		No

Hip	Stretch marks or micro striae are on the patient's hip	Yes
		No
Buttock	Stretch marks or micro striae are on the patient's buttock	Yes
		No
Thigh	Stretch marks or micro striae are on the patient's thigh	Yes
		No
Lumbar or back	Stretch marks or micro striae are on the patient's lumbar or back	Yes
		No
Hernias	The patient has hernias	Yes
		No
Inguinal	The patient has an inguinal hernia	Yes
		No
Hiatus hernia	The patient has an hiatus hernia	Yes
		No
Ombilic	The patient has an ombilic hernia	Yes
		No
On scar	The patient has a hernia on a scar	Yes
		No
Surgery	The patient had surgery to treat the hernia	Yes
		No
		Unknown
Date	Date of the patient's hernia surgery	DD/MM/YYYY
Age	Age of the patient at the time of hernia surgery. Automatically calculated using the patient's date of birth and the date of the surgery.	
Recurrence	The hernia reappeared after surgery	Yes
		No
		Unknown
Cutaneous hyperlaxity	The patient has a cutaneous hyperlaxity	Yes
		No
		Unknown
Thin, translucent skin	The patient has a thin skin and translucent	Yes
		No
		Unknown
Large scar	The patient has large scar	Yes
		No
		Unknown
Livedoid vasculitis	The patient has livedoid vasculitis	Yes
		No
		Unknown
Velvety skin	The patient has velvety skin	Yes
		No
		Unknown

Acrogeria	The patient has acrogeria	Yes
		No
		Unknown
Atrophic scars	The patient has atrophic scars	Yes
		No
		Unknown
Easy bruising	The patient has easy bruising	Yes
		No
		Unknown
Delayed healing	The patient has delayed healing	Yes
		No
		Unknown
Date of last follow-up	Date of the patient's last pneumological follow-up	DD/MM/YYYY
Spontaneous pneumothorax	The patient had a spontaneous pneumothorax	Yes
		No
		Unknown
Type	Type of spontaneous pneumothorax that occurred in the patient	Right
		Left
		Both sides
		Unknown
Date of onset	Date of patient's spontaneous pneumothorax	DD/MM/YYYY
Treatment	Type of treatment for the patient's pneumothorax	Exsuflation
		Surgery
		Unknown
Date of last follow-up	Date of last monitoring of the patient's nervous system	DD/MM/YYYY
Dural ectasia	The patient has a dural ectasia	No
		Yes
		Doubtful
		Unknown
Diagnosis based on	Tool used to diagnose the patient as having dural ectasia	X ray
		MRI
		CT scanner
		Unknown
Peripheral neuropathy	The patient has a peripheral neuropathy	Yes
		No
		Unknown
Diagnosis based on	Tool used to diagnose the patient as having peripheral neuropathy	Physical examination
		EMG
		Nerve biopsy
		Cutaneous biopsy

Date of interview	Date of the patient's interview about his children in the case of a man or about her pregnancies in the case of a woman	DD/MM/YYYY
Number of pregnancies / Number of children	Number of pregnancies (woman) / Number of children (man)	
Prenatal diagnosis	Diagnosis made prior to birth (preimplantatory, or prenatal)	PID
		PND
		Not choosen
		Not possible
		Not available in the center
Prenatal diagnosis result	Result of the prenatal diagnosis. Is the child a mutation carrier?	Non mutation carrier
		Mutation carrier
Child Ranking (N° of child)	Child Ranking (N° of child)	
Pregnancy Ranking (N° of pregnancies)	Pregnancy Ranking (N° of pregnancies)	
Marfan diagnosis known at the beginning of pregnancy	Had the patient been diagnosed with Marfan's disease at the beginning of pregnancy ?	Yes
		No
Type of pregnancy	Number of fetuses in the patient's womb	1
		>1
Beta-Blocker treatment during pregnancy	The patient is taking beta-blocker treatment during her pregnancy	Yes
		No
Maximum aortic diameter at the beginning of pregnancy (mm)	Maximum aortic diameter of the patient at the beggining of pregnancy (mm)	
Ovarian stimulation	The patient has had ovarian stimulatio	Yes induction
		Yes for intrauterine insemination
		Yes in vitro fertilization
		No
Aortic event during pregnancy	The patient had an aortic event during her pregnancy	Yes
		No
Type of events	Type of aortic event during the patient's pregnancy	Dissection type A
		Dissection type B
		Surgery
		Other
Date of onset	Timing of the patient's pregnancy at the time of the aortic event	1° trimester
		2° trimester
		3° trimester
		post partum (<6 months)
Pregnancy reached full-term	Did the patient have a full term pregnancy?	Yes
		No
Term of the pregnancy	Patient's term of pregnancy	1st trimester
		2nd trimester
		3rd trimester
		Unknown
Reason for early termination	Reason for patient's early termination	Miscarriage/foetal death in utero
		Elective abortion

		Therapeutic abortion
Medical termination of pregnancy related with Marfan syndrome	Medical termination of pregnancy related with Marfan syndrome	Yes
		No
Reason	Are the causes of pre-term termination due to the mother or the fœtus ?	Mother
		Foetus
Date of the Delivery	Date of patient's delivery	DD/MM/YYYY
Term of the Delivery (amenorrhea weeks)	Patient's term of delivery	
Prematurity (between 26 and 37 amenorrhea weeks)	Mother's reason for premature delivery	Spontaneous
		For maternal cardiac problem
		For obstetrical problems (RCIU/PE)
Delivery mode	The patient's mode of delivery	Vaginal
		Vaginal with help
		Scheduled Caesarean section
		Caesarean section during labor
Epidural administration	The patient had an epidural	Yes
		Refused
		Not possible dural ectasia
Breast-feeding	The patient is breastfeeding her child	Yes
		No
If not breast-feeding	Reason for mother's refusal to breastfeed	Mother choice
		Impossible due to BB
Aortic diameter of the mother within 6 months after delivery (mm)	Aortic diameter of the mother within 6 months after delivery (mm)	
Continuation of Beta-Blocker treatment after delivery	Patient continues beta-blocker treatment after delivery	Yes
		No
Obstetric complications	Type of obstetrical complications that occurred in the patient	Premature delivery threats
		Premature rupture of membranes
		Fetal growth restriction
		Preeclampsia
		Other
The patient is still followed-up?	The patient is still followed-up?	Yes
		No
If no, specify the reason for premature termination	Reason for discontinuation of patient follow-up in the study	Death of the patient
		Patient's refusal to continue collecting data and / or samples (patient data / samples collected up to this date may be used)
		Refusal of the patient to continue collecting his data and written request made by the patient for the deletion of all his data and / or samples
		Decision of the investigator
		Lost of follow-up
Cause of death	Cause of patient's death	Cardiovascular
		Non cardiovascular

		Unknown
		Other
If cardiovascular	What was the cardiovascular cause of death of the patient?	Aortic dissection
		Sudden cardiac death
		Heart failure
		Post surgery (within 30 days)
If non cardiovascular	What was the non cardiovascular cause of death of the patient?	Cancer
		Infection
If other	Cause of death of the patient other than cardiovascular, infection or cancer	
Death due to rare disease: (BNDMR)	Is the patient's death due to his rare disease?	Yes
		No
Date of death	Date of the patient's death	DD/MM/YYYY
If decision of the investigator, specify the reason	Reason for investigator's decision to discontinue patient follow-up	
Date end of study	The end of study date corresponds to the date of death. The study end date corresponds to the date of the patient's refusal to continue collecting data and / or samples (patient data / samples collected up to this date may be used). The end of the study date corresponds to the date of the patient's refusal to continue collecting his data and a written request made by the patient for the deletion of all his data and / or samples. The study end date corresponds to the investigator's decision date. The end of study date corresponds to the date of the latest news.	DD/MM/YYYY