GENTAC

CLINICAL EVALUATION – FORM 1

Subject ID Label

1. Enrollment Diagnosis: (YOU MUST CHECK 1 PRIMARY CONDITION; YOU MAY ALSO CHECK 1 SECONDARY CONDITION.)

Diagnosis	1º	2º
a. Marfan syndrome	☐	☐
b. Turner syndrome	☐	☐
c. Ehlers-Danlos syndrome, vascular type	☐	☐
d. Ehlers-Danlos syndrome, other type and with aortic enlargement	☐	☐
e. Loeys-Dietz syndrome	☐	☐
f. FBN1, TGFBR1,TGFBR2, ACTA2 or MYH11 genetic mutation	☐	☐
g. BAV with aortic enlargement, family history not required	☐	☐

Diagnosis	1º	2º
h. BAV with family history	☐	☐
i. BAV with coarctation	☐	☐
j. Shprintzen-Goldberg syndrome	☐	☐
k. Familial Thoracic Aortic Aneurysm and Dissections with aortic enlargement	☐	☐
l. Other aneurysms/dissections of the thoracic aorta in persons < 50 years of age	☐	☐
m. Other congenital heart disease with aortic enlargement	☐	☐
n. 1st degree family member of proband already enrolled in the registry (with approval)	☐	☐

2. Approximate age at earliest diagnosed above condition (physician confirmed): _____ YEARS

3. Ever diagnosed with a thoracic aortic aneurysm, dissection/rupture, or marked tortuosity?
   ☐ Yes
   ☐ No ➔ GO TO Q5
   ☐ Unknown ➔ GO TO Q5

4. Specify the current status of each aortic area (CHECK ALL THAT APPLY IN EACH ROW) and enter date of earliest abnormal:

Area of Aorta	Always Normal	Ever Tortuous	Ever Dilated	Ever Dissected	Unknown	Date of Earliest Abnormality
a. Ascending	☐	☐	☐	☐	☐	
b. Arch	☐	☐	☐	☐	☐	
c. Descending	☐	☐	☐	☐	☐	
d. Abdominal	☐	☐	☐	☐	☐	
e. Sinus of Valsalva	☐	☐	☐	☐	☐	

BASIC MEASUREMENTS (at time of enrollment)

5. Height .....|___|___|___| CM |___| FT |___|___| INCHES
6. Weight .....|___|___|___| . |___| KG |___|___|___| POUNDS
7. Arm Span .....|___|___|___| CM |___|___| INCHES
8. Lower Segment .....|___|___|___| CM |___|___| INCHES
9. Heart Rate .....|___|___|___| BEATS/MINUTE
10. Blood Pressure .....|___|___|___| SYSTOLIC / |___|___|___| DIASTOLIC

DIAGNOSTIC CRITERIA AND ORGAN SYSTEM REVIEW

11. Indicate the historical status of each condition with a check in the appropriate column (YES, NO). Check NO if the patient is not known to have the condition.

A. Musculoskeletal	Yes	No/Unk		Yes	No/Unk
1. Pectus carinatum	☐	☐	11. Osteopenia/osteoporosis	☐	☐
2. Pectus excavatum	☐	☐	12. Congenital bilateral dislocated hips	☐	☐
3. Shield chest, broad with widely spaced nipples	☐	☐	15. Joint pain, arthralgia	☐	☐
4. Recurrent dislocations/sublaxations	☐	☐	16. Thumb sign	☐	☐
5. Joint hypermobility in >1 joint	☐	☐	21. Clubfoot	☐	☐
6. Wrist sign	☐	☐	22. Reduced elbow extension (<170°)	☐	☐
7. Scoliosis that is clinically evident	☐	☐	23. Long fingers, arachnodactyly	☐	☐
8. Abnormal kyphoscoliosis/lordosis	☐	☐	24. Genu recurvatum	☐	☐
9. Pes planus	☐				

B. Pulmonary	Yes	No/Unk		Yes	No/Unk
1. Pneumothorax	☐	☐	2. Asthma	☐	☐

C. Cardiovascular	Yes	No/Unk		Yes	No/Unk
1. Patent ductus arteriosus	☐	☐	18. Endocarditis	☐	☐
2. Atrial septal defect (ASD)/PFO	☐	☐	19. Atrial arrhythmia	☐	☐
3. Ventricular septal defect (VSD)	☐	☐	20. Cardiac arrhythmia (not atrial)	☐	☐
4. Tetralogy of Fallot	☐	☐	21. Coronary dissection	☐	☐
5. Other congenital heart defect, specify:	☐	☐	22. Coronary ectasia	☐	☐
6. Mitral valve prolapse	☐	☐	23. Carotid dissection	☐	☐
8. Mitral valve regurgitation > trace	☐	☐	24. Symptomatic coronary artery disease	☐	☐
9. Calcified mitral anulus	☐	☐	26. Congestive heart failure/left ventricular function	☐	☐
10. Aortic valve regurgitation	☐	☐	27. Left ventricular dilation	☐	☐
11. Aortic valve stenosis	☐	☐	28. Left ventricular hypertrophy	☐	☐
12. Aortic coarctation, current/repaired	☐	☐	29. Right ventricular hypertrophy	☐	☐
13. Biscuspid Aortic Valve (BAV)	☐	☐	30. Right ventricular dysfunction/failure	☐	☐
14. Tricuspid regurgitation > trace	☐	☐	31. Dilatation of the pulmonary artery	☐	☐
15. Pulmonary stenosis/right ventricular outflow obstruction	☐	☐	32. Other pulmonary artery abnormality	☐	☐
16. Pulmonary regurgitation > trace	☐	☐	33. Hypertension	☐	☐

D. Craniofacial	Yes	No/Unk		Yes	No/Unk
1. High palate/dental crowding	☐	☐	6. Down-slanting palpebral fissures	☐	☐
2. Dolicocephaly	☐	☐	7. Cleft palate	☐	☐
3. Malar hypoplasia	☐	☐	8. Bifid or broad-based uvula	☐	☐
4. Enophthalmos	☐	☐	9. Craniosynostosis	☐	☐
5. Retrognathia	☐	☐			

E. Ocular **Examiner Type:** ☐ Ophthalmologist ☐ Optometrist ☐ Unknown
Slit lamp Exam: ☐ No ☐ Yes ☐ Unknown

	Yes	No/Unk		Yes	No/Unk
1. Lens dislocation (ectopia lentis)	☐	☐	9. Myopia >4 diopters	☐	☐
2. Glaucoma	☐	☐	10. Cataracts, <50 years of age	☐	☐
4. Hypertelorism	☐	☐	11. Iris Flocculi	☐	☐
5. Retinal detachment	☐	☐	12. Blue/grey sclera	☐	☐
6. Amblyopia	☐	☐	13. Iridodenesis	☐	☐
7. Strabismus	☐	☐			

F. Skin/integument	Yes	No/Unk		Yes	No/Unk
1. Skin hyperextensibility	☐	☐	8. Striae atrophicae	☐	☐
2. Wide atrophic scars	☐	☐	9. Hernia, specify type	☐	☐
3. Poor wound healing	☐	☐	10. Recurrent hernia	☐	☐
4. Easy bruising	☐	☐	11. Thin translucent skin	☐	☐
5. Soft, velvety or doughy skin	☐	☐	12. Webbed neck	☐	☐
6. Tissue fragility	☐	☐			

G. Central Nervous and Neurologic	Yes	No/Unk		Yes	No/Unk
1. Lumbosacral dural ectasia	☐	☐	5. Abnormal junction of the brain and medulla (Arnold-Chiari malformation)	☐	☐
2. Hemorrhagic stroke	☐	☐	6. Migraines	☐	☐
3. Ischemic stroke	☐	☐	7. Cerebral aneurysms, specify location:	☐	☐
4. Stroke, type unknown	☐	☐			

H. Vascular and Circulatory	Yes	No/Unk		Yes	No/Unk
1. Early-onset varicose veins, <40 years of age	☐	☐	3. Arterial rupture	☐	☐
2. Arteriovenous malformation	☐	☐	4. Lymphedema	☐	☐
I. Gastrointestinal	**Yes**	**No/Unk**		**Yes**	**No/Unk**
1. Ulcerative colitis	☐	☐	4. Irritable bowel syndrome	☐	☐
2. Ruptured bowel	☐	☐	5. Diverticulosis/diverticulitis	☐	☐
3. Crohn's disease	☐	☐			
J. Reproductive (WOMEN ONLY)	**Yes**	**No/Unk**		**Yes**	**No/Unk**
1. Uterine rupture	☐	☐	4. Uterine fibroids	☐	☐
2. Ovarian failure	☐	☐	5. Premature rupture of membranes	☐	☐
3. Endometriosis	☐	☐			
K. Other Important Health Problem Potentially Related to Diagnosis	**Yes**	**No/Unk**		**Yes**	**No/Unk**
1. Learning disability	☐	☐	3. Other, specify:	☐	☐
2. ADHD	☐	☐	4. Other, specify:	☐	☐
L. Familial History	**Yes**	**No/Unk**		**Yes**	**No/Unk**
1. Immediate family history (parent, child, sibling) of TAA or other major vessel dilatation or dissection	☐	☐	2. Secondary family history (cousin, aunt, uncle, grandparent) of TAA or other major vessel dilatation or dissection	☐	☐

12. Has patient ever been diagnosed with diabetes?
    - ☐ Yes → 12a. What was the age at diagnosis? _____ YEARS
    - ☐ No ➔ GO TO Q13
    - ☐ Don't know ➔ GO TO Q13

    12b. Does the patient receive insulin? ☐ Yes ☐ No ☐ Don't know

13. Patient ever been diagnosed with cancer?
    - ☐ Yes
    - ☐ No ➔ GO TO Q14
    - ☐ Don't know ➔ GO TO Q14

    13a. IF YES: For each primary cancer, complete a row in the table:

	Primary Cancer Type	Cancer Location	Age at Diagnosis	Metastatic?
1.				☐ Yes ☐ No ☐ Unknown
2.				☐ Yes ☐ No ☐ Unknown

14. Have any family members of this patient been enrolled in this Registry (here or at any other participating center)?
    - ☐ Yes
    - ☐ No ➔ GO TO Q15
    - ☐ Don't know ➔ GO TO Q15

    14a. IF YES: For each family member enrolled in this Registry, complete a row in the table:

	GenTAC ID Number	Code	Relationship Code List		
1.			00 = Grandchild	07 = Maternal Aunt	13 = Maternal Niece
2.			01 = Maternal Grandparent	08 = Paternal Aunt	14 = Paternal Niece
3.			02 = Paternal Grandparent	09 = Maternal Uncle	15 = Maternal Nephew
4.			03 = Parent	10 = Paternal Uncle	16 = Paternal Nephew
5.			04 = Whole Sibling	11 = Maternal Cousin	17 = Other
6.			05 = Half Sibling	12 = Paternal Cousin	
			06 = Child		

15. For each drug or category listed, check one box in each row which best describes the status of the drug treatment.

Drug Treatment	Never Prescribed	Past Use Only	Currently Using	Possible Use, Clinical Trial	Unknown	Earliest Start Date Month/Year
a. Beta-Blocker	☐	☐	☐	☐	☐	
b. Losartan	☐	☐	☐	☐	☐	
c. Other ARB	☐	☐	☐	☐	☐	
d. ACE-Inhibitor	☐	☐	☐	☐	☐	
e. Statins	☐	☐	☐	☐	☐	
f. Verapamil or Diltiazem	☐	☐	☐	☐	☐	
g. Other Ca+ channel blocker	☐	☐	☐	☐	☐	
h. Other BP lowering drug	☐	☐	☐	☐	☐	

16. Has patient ever had a surgical procedure related to treatment of their cardiovascular genetic disorder?

☐ Yes
☐ No ➔ GO TO Q17
☐ Don't know ➔ GO TO Q17

16a. IF YES: Indicate the procedures received by checking the "YES" column and entering the corresponding Month and Year, **if readily available** (otherwise leave blank). Complete a Surgical Intervention Form for each procedure *except* for Pacemaker/ICD implantation and Repair of pectus deformity.

Surgical Intervention	Yes	MM/YY	Surgical Intervention	Yes	MM/YY
1. Coronary artery bypass or PTCA grafting	☐		11. Descending thoracic aortic replacement	☐	
2. Aortic valve repair	☐		12. Thoracoabdominal aortic replacement	☐	
3. Isolated aortic valve replacement (non-root)	☐		13. Open arterial bypass	☐	
4. Aortic root replacement (valve-replacing)	☐		14. Repair of pectus deformity	☐	
5. Valve-sparing aortic root replacement	☐		15. Percutaneous/endovascular intervention, non-coronary PTCA	☐	
6. Ascending aortic replacement	☐		16. Aorta to pulmonary shunt	☐	
7. Aortic arch replacement	☐		17. TOF repair	☐	
8. Mitral valve repair or replacement	☐		18. Pulmonary valve replacement	☐	
9. Coarctation repair	☐		19. Tricuspid valve surgery	☐	
10. Pacemaker/ICD implantation	☐		20. Other, specify:	☐	

16b. How many different Surgical Intervention Forms will be completed? _____ ☐ Unknown

17. Source of information for answers recorded on this form (*Check all that apply.*)

☐ Medical record of exams and tests done at your institution
☐ Medical record of exams and tests done elsewhere
☐ Medical record of patient interviews and clinical history
☐ Direct patient interview specifically for this study

18. Year this patient was first seen at your institution: |___|___|___|___| ☐ Patient not seen at this institution

Initials of Abstractor _____

Comments: _____