

NGN NCLEX 2026 • GENETIC / CONNECTIVE TISSUE / CARDIOVASCULAR

Marfan Syndrome

An autosomal-dominant connective-tissue disorder where defective fibrillin-1 weakens elastic tissue throughout the body — most dangerously, the aorta. The NCLEX priority is always the same: protect the aorta, recognize dissection, and reduce cardiovascular stress.

AUTOSOMAL DOMINANT

FBN1 GENE

CHROMOSOME 15

AORTIC RISK

OCULAR • SKELETAL • CARDIAC

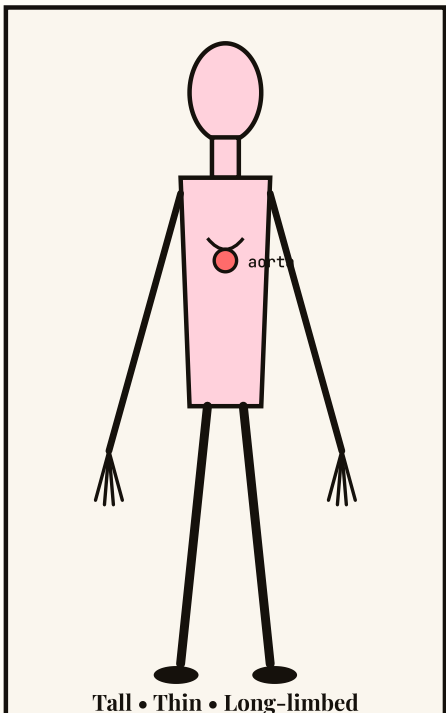


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Definition & Overview

WHAT IT IS • WHY IT MATTERS



- **Genetic, autosomal-dominant connective-tissue disorder** caused by mutation of the **FBN1 gene on chromosome 15**, which codes for **fibrillin-1** — a protein essential to elastic microfibrils.
- Defective fibrillin-1 weakens connective tissue throughout the body, affecting the **cardiovascular, skeletal, ocular, pulmonary, integumentary, and dural systems**.
- **Why it matters:** the #1 cause of death is **aortic aneurysm rupture or dissection**. Early identification + beta-blocker therapy + activity restriction dramatically extends lifespan.
- Prevalence $\approx$ **1 in 5,000**. ~25% of cases are *de novo* mutations (no family history).

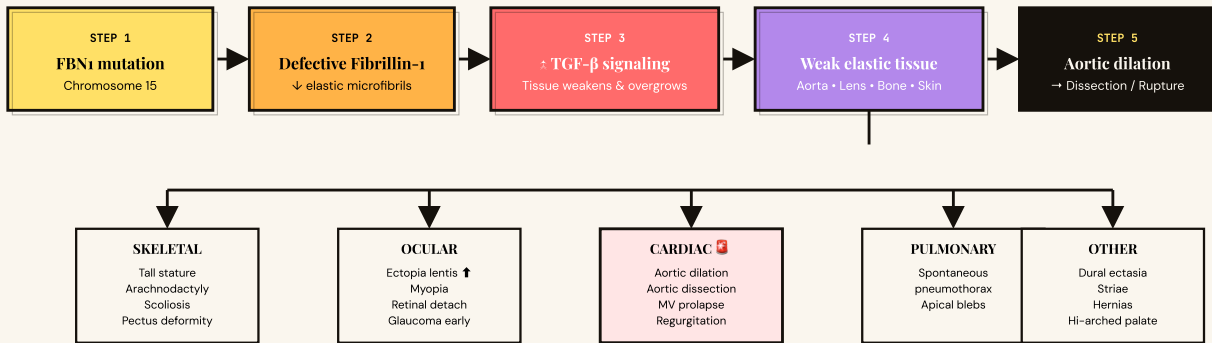
NCLEX PEARL

"**Tall + thin**" on its own is *not* Marfan. Diagnosis requires the **Ghent criteria** — most commonly **aortic root dilation + ectopia lentis**, or **aortic involvement + FBN1 mutation**.

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Pathophysiology

FROM GENE TO AORTIC DISSECTION



MECHANISM BOILED DOWN

Gene defect → bad protein → weak connective tissue → tissues that need elasticity FAIL. The aorta is the highest-stakes failure point because it absorbs every heartbeat of pulsatile pressure.

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Signs & Symptoms

BY SYSTEM • RED FLAGS HIGHLIGHTED

SKELETAL

- **Tall stature** — arm span > height (ratio > 1.05)
- **Arachnodactyly** — long, spider-like fingers
- **Positive thumb sign (Steinberg)** — thumb protrudes past ulnar border when folded across palm
- **Positive wrist sign (Walker-Murdoch)** — thumb and 5th finger overlap when grasping opposite wrist
- **Pectus excavatum** (caved chest) or **pectus carinatum** (pigeon chest)
- **Scoliosis / kyphosis**
- Long narrow face, high-arched palate, crowded teeth
- Joint hypermobility, flat feet (pes planus)

OCULAR

- **Ectopia lentis** — superior/temporal lens dislocation (60% of patients)
- **Severe myopia** (nearsightedness)
- **Retinal detachment**
- Early-onset cataracts and glaucoma
- Blurred vision, "floaters," sudden vision loss

🚩 **Distinguish:** Marfan = lens up; *Homocystinuria* = lens down.

CARDIAC — RED FLAGS

- 🚩 **Aortic root dilation** (#1 monitor)
- 🚩 **Aortic aneurysm / dissection** — leading cause of death
- **Mitral valve prolapse** ± regurgitation — systolic click/murmur
- Aortic regurgitation — diastolic murmur
- Palpitations, dyspnea on exertion

🚩 **Aortic dissection alert:** SUDDEN severe "tearing" chest pain → radiates to back, BP differential between arms (>20 mmHg), pulse deficit, syncope. → 🚩 **This is a surgical emergency.**

PULMONARY / OTHER

- **Spontaneous pneumothorax** — sudden chest pain + dyspnea, especially in tall thin males
- Pulmonary blebs / bullae on imaging
- **Dural ectasia** — low-back pain, headaches, leg weakness
- **Striae** not related to weight gain
- Inguinal / incisional hernias (recurrent)
- Obstructive sleep apnea (high-arched palate)

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Priority Nursing Interventions

ABCS • SAFETY • PRIORITIZATION

AIRWAY / CIRCULATION

- **Assess** for sudden tearing chest/back pain — rule out dissection **FIRST**
- **Monitor** BP *in both arms* — >20 mmHg differential = red flag
- **Auscultate** for new murmurs (MR, AR), S3, S4
- **Maintain** SBP < 120 mmHg goal (per HCP)
- Continuous cardiac monitoring if acute symptoms

ACTIVITY / SAFETY

- **Restrict** contact sports, competitive athletics, weightlifting
- **NO isometric exercise** — spikes intrathoracic pressure
- Allow **moderate aerobic activity** (walking, swimming)
- Fall precautions if visual impairment
- Medical alert bracelet

MONITORING

- **Annual echocardiogram** (more often if aortic root > 4 cm)
- **Annual ophthalmology** exam — slit lamp + dilated fundus
- Yearly orthopedic / scoliosis screen in growing children
- Pulmonary function as indicated
- Genetic counseling for family

PHARMACOLOGY

- **Administer beta-blockers** (atenolol, propranolol) as ordered — slows aortic dilation
- **ARBs (losartan)** — adjunct, blocks TGF- β
- Endocarditis prophylaxis with valve disease (if prior endocarditis or prosthetic valve)
- Avoid stimulants, decongestants, triptans

PSYCHOSOCIAL / EDUC.

- Genetic counseling — 50% inheritance risk to offspring
- Body-image support (height, deformities)
- Career counseling — avoid high-physical-demand jobs
- Pregnancy is HIGH-risk — preconception cardiology

EMERGENCY: DISSECTION

- **Call rapid response / 911**
- Bed rest, semi-Fowler
- IV access $\times$ 2 large bore
- IV beta-blocker (esmolol) **FIRST** → then nitroprusside for BP
- Type & crossmatch — surgical repair
- NPO, pain control

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Pharmacology

SLOW THE AORTA, LOWER THE LOAD

Atenolol • Propranolol

BETA-BLOCKER • FIRST LINE

MECHANISM: Blocks β_1 receptors $\rightarrow$ $\downarrow$ HR, $\downarrow$ contractility, $\downarrow$ shear stress on aortic wall. **Slows rate of aortic root dilation.**

SIDE EFFECTS: Bradycardia, hypotension, fatigue, bronchospasm (mask hypoglycemia in diabetics).

NURSING: Hold if HR < 60 or SBP < 100 ; teach not to stop abruptly (rebound HTN, tachycardia); take pulse before dose.

Losartan

ARB • ADJUNCT

MECHANISM: Blocks angiotensin II AT1 receptor; **reduces TGF- β signaling** implicated in Marfan tissue overgrowth.

SIDE EFFECTS: Hyperkalemia, hypotension, dizziness, $\uparrow$ creatinine, angioedema (rare).

NURSING: Monitor K^+ and renal function; avoid in pregnancy (teratogenic); rise slowly to prevent orthostasis.

Esmolol • Labetalol (IV)

ACUTE DISSECTION

MECHANISM: Rapid IV beta-blockade for acute aortic dissection — given **BEFORE** any vasodilator to prevent reflex tachycardia.

TARGETS: HR ≤ 60 , SBP 100–120 mmHg.

NURSING: Continuous BP and ECG monitoring; titrate to parameters; have atropine available for bradycardia.

Nitroprusside

VASODILATOR • DISSECTION

MECHANISM: Direct arterial/venous dilation $\rightarrow$ rapid BP reduction. Added **after** beta-blocker in dissection.

SIDE EFFECTS: Cyanide toxicity (prolonged/high-dose), severe hypotension, methemoglobinemia.

NURSING: Use **ONLY** with arterial line monitoring; protect from light; max 10 mcg/kg/min; check thiocyanate levels >48 h.

⚠️ AVOID IN MARFAN

Stimulants (amphetamines, ADHD meds — case-by-case with cardiology), **decongestants** (pseudoephedrine, phenylephrine), **triptans**, **fluoroquinolones** ($\uparrow$ aortic dissection risk), **cocaine**, and **nicotine**. All raise BP or weaken aortic tissue.

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NCLEX Test Traps

WRONG VS. RIGHT THINKING

✗ WRONG

"Patient is tall and thin → must have Marfan."

✓ RIGHT

Diagnosis requires **Ghent criteria**: aortic root dilation/dissection + ectopia lentis, family history, OR FBN1 mutation.

✗ WRONG

Marfan patient with sudden chest pain → suspect MI first.

✓ RIGHT

Sudden **tearing** pain radiating to back in Marfan → suspect **aortic dissection** until proven otherwise. CT angiography of aorta.

✗ WRONG

"Restrict contact sports during growth spurts only."

✓ RIGHT

Contact sports, competitive athletics, isometric exercise, and heavy lifting are **restricted for life**.

✗ WRONG

Lens dislocation is downward (same as homocystinuria).

✓ RIGHT

Marfan = lens **UP and out** (superotemporal).
Homocystinuria = lens **DOWN**.

✗ WRONG

"Give vasodilator first to drop BP in dissection."

✓ RIGHT

Beta-blocker FIRST (esmolol/labetalol), THEN vasodilator.
Vasodilator alone → reflex tachycardia → ↑ shear stress → worsens dissection.

✗ WRONG

Beta-blockers cure Marfan syndrome.

✓ RIGHT

Beta-blockers **slow progression** of aortic dilation; they don't fix the underlying genetic defect.

✗ WRONG

Marfan pregnancy is the same risk as a normal pregnancy.

✓ RIGHT

Pregnancy is **HIGH-risk** — hemodynamic strain ↑ dissection risk. Pre-conception aortic root assessment; surgery often advised if root ≥ 4.5 cm.

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Patient & Family Education

LIVING SAFELY WITH MARFAN



Activity

- **Avoid:** football, basketball, wrestling, weightlifting, push-ups, sit-ups (isometrics), competitive sports
- **Encourage:** walking, swimming, light cycling, golf, doubles tennis at low intensity
- Stop activity if dizzy, chest pain, palpitations



Medication Adherence

- Take beta-blocker EVERY day — never stop abruptly
- Check pulse and BP daily; report HR < 60 or SBP < 100
- Avoid OTC decongestants (Sudafed, Afrin)
- Tell every provider you have Marfan before any new prescription



When to Call 911

- **Sudden severe chest or back pain** — tearing/ripping quality
- **Sudden shortness of breath** (possible pneumothorax)
- **Sudden vision change** — flashes, floaters, curtain (retinal detachment)
- Fainting / loss of consciousness
- One-sided weakness, slurred speech



Family / Genetic

- 50% chance of passing FBN1 to each child
- First-degree relatives need screening: echocardiogram + slit-lamp + FBN1 testing
- Genetic counseling before pregnancy
- Pregnancy planned with high-risk OB AND cardiology



Health Maintenance

- Annual echocardiogram (more often if root ≥ 4 cm)
- Annual ophthalmology dilated exam
- Routine dental care — endocarditis prophylaxis only per current AHA criteria (prior endocarditis, prosthetic valve, valve repair material)
- Medical alert bracelet "MARFAN — AORTIC RISK"



Daily Lifestyle

- No smoking / vaping (pneumothorax risk)
- Avoid caffeine excess
- Eye protection for sports (LASIK contraindicated; risk of detachment)
- Treat sleep apnea aggressively
- Mental health support — anxiety/depression common

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Memory Boosters

MNEMONICS • PATTERN RECOGNITION

PRIMARY MNEMONIC — "M.A.R.F.A.N"

M

Mitral Valve
Prolapse

A

Aortic
Aneurysm

R

Retinal
Detachment

F

Fibrillin-1
Defect

A

Arachnodactyly

N

No Strenuous
Activity



"Spider Fingers"

Arachno- = spider. Long, slender fingers + thumb sign + wrist sign = classic Marfan hand.

Steinberg thumb sign: Make a fist around your own thumb — if the thumb tip pokes past the pinky side of the hand, it's positive.

Walker-Murdoch wrist sign: Grasp your wrist with the opposite hand — thumb and 5th finger should overlap if positive.



Lens Direction = Diagnosis

Marfan = lens **UP** (think M is high on the page).

Homocystinuria = lens **DOWN** (think H is low).

Both cause ectopia lentis but the direction is the boards' favorite distractor.



"Beta Before Vaso"

In aortic dissection: **Beta-blocker FIRST**, then vasodilator.

Vasodilator alone = reflex tachycardia = MORE shear stress = WORSE dissection.



Arm Span > Height

Normal: arm span $\approx$ height (ratio < 1.0).

Marfan: arm span $> 1.05 \times$ height.

Plus reduced upper-to-lower segment ratio (< 0.85 in adults) — legs disproportionately long.



"No I.C.E." Activities

Isometric exercise (weightlifting)

Contact sports (football, hockey)

Extreme exertion (sprinting, max-effort)



Annual "Eyes & Echo"

Two annual must-dos:

- **Echocardiogram** — aortic root + valves
- **Eye** exam — dilated, slit-lamp

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NCJMM Clinical Judgment Scenario

SIX-STEP NEXT GEN NCLEX WALKTHROUGH

CASE

Demographics: 22-year-old male, 6'5", weight 145 lb (BMI 17.2), with known Marfan syndrome diagnosed at age 14.

Presentation: Arrives in ED reporting sudden onset of severe "ripping" chest pain that radiates to his upper back. Pain started 30 minutes ago while lifting a bag of mulch in the garden. He is diaphoretic and anxious.

Vital signs: HR 118, RR 26, BP **184/108 (right arm), 142/86 (left arm)**, SpO₂ 94% on room air, T 98.7 °F.

Exam: Tall, thin build with arachnodactyly. New early diastolic murmur at the right sternal border. Diminished left radial pulse compared to right. Lung sounds clear bilaterally. Recently stopped his atenolol "because it made him tired."

STEP 1 — RECOGNIZE CUES

What stands out?

- Known Marfan syndrome — high baseline aortic risk
- Sudden severe **tearing/ripping** chest pain → back radiation
- Onset during isometric activity (lifting)
- **BP differential > 20 mmHg** between arms
- Diminished left radial pulse (pulse deficit)
- New diastolic murmur (possible aortic regurgitation)
- Beta-blocker non-adherence
- Tachycardia and tachypnea

STEP 2 — ANALYZE CUES

What do they mean together?

The combination of Marfan + isometric trigger + tearing pain + BP differential + pulse deficit + new diastolic murmur is the **classic pattern of acute aortic dissection**. The new murmur suggests dissection has extended into the aortic root, causing aortic regurgitation. Beta-blocker non-adherence removed the only protection slowing aortic wall stress, and the lifting episode acutely spiked his BP and shear forces.

STEP 3 — PRIORITIZE HYPOTHESES

Most likely → least likely

- **#1 Acute aortic dissection (Stanford Type A)** — HIGHEST priority, time-critical, fits every cue
- **#2 Spontaneous pneumothorax** — possible in tall thin male, but breath sounds clear and no tracheal shift
- **#3 Acute MI** — possible but Marfan + tearing pain + BP differential makes dissection far more likely
- **#4 Pulmonary embolism** — less consistent with the tearing pain and BP findings

STEP 4 — GENERATE SOLUTIONS

What needs to happen?

- Activate rapid response / cardiothoracic surgery team STAT

- Obtain CT angiography of chest/aorta (or TEE if unstable)
- Place two large-bore IVs; type & crossmatch 4 units PRBCs
- Start continuous cardiac, BP (arterial line preferred), and SpO₂ monitoring
- IV **esmolol** drip to achieve HR ≤ 60 and SBP 100–120
- Add **nitroprusside** AFTER beta-blockade for BP control
- NPO, bed rest semi-Fowler, IV pain control (morphine/fentanyl)
- Prepare for emergency surgical repair
- Notify family; obtain consent

STEP 5 — TAKE ACTION

Implement in priority order

1. **Call rapid response / cardiothoracic surgery** — this is a surgical emergency
2. Apply continuous cardiac monitoring; obtain 12-lead ECG
3. Establish 2 large-bore IVs; draw labs (CBC, BMP, type & crossmatch, troponin, BNP, d-dimer)
4. **Initiate IV esmolol** first per orders — HR/BP titration
5. Add nitroprusside only AFTER HR controlled
6. Place patient in semi-Fowler, NPO, bed rest
7. Administer IV opioid analgesia per orders (pain ↑ catecholamines)
8. Transport to CT angiography or directly to OR per surgeon
9. Communicate clearly with family; provide emotional support

STEP 6 — EVALUATE OUTCOMES

Did it work?

- **Expected outcomes:** HR ≤ 60, SBP 100–120, pain reduced, BP differential narrowing, no extension of dissection on serial imaging, hemodynamic stability for OR
- **Worsening signs to escalate:** Widening pulse pressure, new neuro deficits (carotid extension), oliguria (renal artery extension), abdominal pain (mesenteric extension), syncope, cardiac tamponade signs (Beck's triad: ↓BP, ↑JVD, muffled heart sounds)
- **Post-op evaluation:** Surgical repair complete, normal sinus rhythm, urine output ≥ 0.5 mL/kg/hr, neurologic baseline intact, pain controlled, family informed and supported
- **Long-term:** Re-education on lifelong beta-blocker adherence, activity restrictions, follow-up echocardiogram schedule, genetic counseling reinforcement