ENDOCRINE • GENETICS • PITUITARY

Acromegaly • Gigantism • Marfan Syndrome

The "Tall & Large" Differential — When Hormones & Genes Go Big

1 Definition / Overview

Three distinct disorders that produce **excessive growth or elongation** — but the cause and danger differ. Mastering the distinction is critical for prioritization on NGN NCLEX.

Acromegaly

GH EXCESS — ADULT

- Excess Growth Hormone **AFTER** epiphyseal plates close
- Almost always caused by a **pituitary adenoma**
- Insidious onset — often diagnosed late (40s–50s)
- Bones thicken/widen; do NOT lengthen

Gigantism

GH EXCESS — CHILD

- Excess GH **BEFORE** epiphyseal plates close
- Same cause: **pituitary adenoma**
- Produces extreme linear (height) growth
- Untreated → may exceed 7–8 feet tall

Marfan Syndrome

GENETIC — CONNECTIVE TISSUE

- **Autosomal dominant** (FBN1 gene mutation)
- Defective **fibrillin-1** → weak connective tissue
- Affects skeletal, cardiovascular, ocular systems
- Aortic dissection = leading cause of death 🚨

2 Pathophysiology (Simplified)

🔶 Acromegaly & Gigantism (same mechanism)

Pituitary adenoma → ↑ Growth Hormone (GH) → Liver releases ↑ IGF-1 → Tissue/bone overgrowth →

Adult = thickening | Child = lengthening

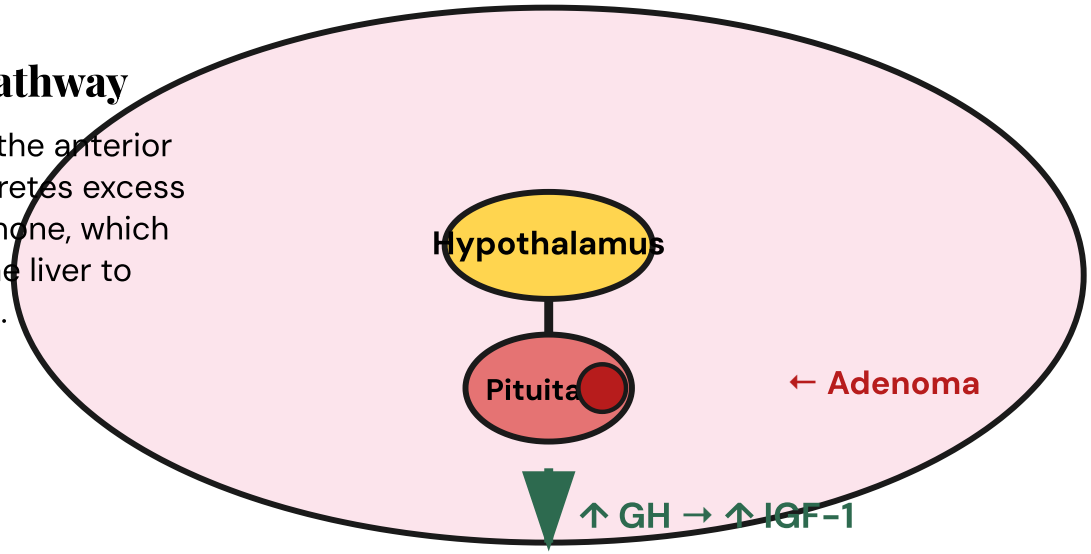
🧬 Marfan Syndrome

FBN1 gene mutation → Defective fibrillin-1 protein → Weak connective tissue → Stretching of aorta, lens, ligaments →

Aneurysm, dislocation, hypermobility

The GH Pathway

Adenoma in the anterior pituitary secretes excess Growth Hormone, which stimulates the liver to release IGF-1.



Anatomy: Pituitary adenoma → ↑GH → ↑IGF-1 (the engine of acromegaly & gigantism)

3 Signs & Symptoms — Side-by-Side

System	Acromegaly	Gigantism	Marfan
Stature	Normal height (plates closed)	Excessive linear growth — very tall	Tall, thin, long limbs (dolichostenomelia)
Hands & Feet	Enlarged, ring/shoe size ↑	Proportionately large	**Arachnodactyly** — long, "spider" fingers
Face	Coarse features, prognathism (jaw protrusion), enlarged tongue, deep voice	Coarsening if untreated into adulthood	Long narrow face, high-arched palate, crowded teeth, micrognathia
Cardiac 🚨	Cardiomegaly, HTN, CHF	Cardiomegaly	**AORTIC ANEURYSM / DISSECTION**, MVP, regurgitation
Visual	**Bitemporal hemianopia** (tumor compresses optic chiasm), HA	Same — visual field defects, HA	**Lens dislocation** (ectopia lentis), myopia, retinal detachment
Skeletal	Joint pain, arthralgia, carpal tunnel	Delayed puberty, joint pain	Pectus excavatum/carinatum, scoliosis, joint hypermobility
Metabolic	Insulin resistance → DM, ↑ sweating	DM, hyperhidrosis	Usually normal
Other	Sleep apnea, organomegaly, ↑ colon polyps	Sleep apnea	**Spontaneous pneumothorax**, dural ectasia

🚨 RED FLAG FINDINGS — Call Provider NOW

- 🚨 **Marfan + sudden severe chest/back pain = aortic dissection until proven otherwise**
- 🚨 **Marfan + sudden dyspnea/unilateral chest pain = spontaneous pneumothorax**
- 🚨 **Acromegaly/Gigantism + new visual field loss or severe HA = tumor expansion / pituitary apoplexy**
- 🚨 **Marfan + sudden vision change or "curtain" = retinal detachment or lens dislocation**

Acromegaly + worsening sleep apnea/airway compromise = enlarged tongue/soft tissue obstruction

4 Priority Nursing Interventions (ABC + Safety)

1. **AIRWAY:** Acromegaly → assess for sleep apnea, enlarged tongue, soft-tissue airway obstruction. Keep oral airway/intubation supplies accessible. Position semi-Fowler's.
2. **CIRCULATION (Marfan priority):** Monitor BP carefully — keep SBP < 120 to reduce aortic wall stress. Auscultate for new murmurs. Check pulses bilaterally. **Assess for chest/back pain — dissection!**
3. **NEURO/VISION:** Perform **visual field testing** in pituitary tumor patients (bitemporal hemianopia). Headache + visual change = report immediately.
4. **POST-OP TRANSSPHENOIDAL HYPOPHYSECTOMY:** Do NOT brush teeth, blow nose, sneeze, cough, bend, or strain ×2 weeks (risk of CSF leak). HOB elevated 30°. Watch for "halo sign" / clear drip = CSF leak (test for glucose).
5. **MONITOR FOR DI:** After hypophysectomy, ADH may drop → polyuria, ↑ serum Na, low urine specific gravity. Strict I&O, daily weights.
6. **ACTIVITY (Marfan):** NO contact sports, weightlifting, isometric exercise. Encourage swimming, walking. Vigorous exertion can rupture aorta.
7. **GLUCOSE MONITORING:** Acromegaly causes insulin resistance — fingersticks, monitor for hyperglycemia.
8. **GENETIC COUNSELING (Marfan):** Autosomal dominant — 50% chance of passing to offspring. Family screening recommended.
9. **SUPPORT & BODY IMAGE:** Disfiguring features can cause severe psychosocial distress. Provide emotional support, refer to support groups.

10 **SAFETY (Marfan):** Eye exams yearly (lens dislocation), echocardiogram yearly, MedicAlert bracelet, avoid LASIK.

🧠 NCJMM Clinical Judgment Framework

1. Recognize Cues

Coarse facial features + ring/shoe size ↑ + visual changes → acromegaly. Tall + arachnodactyly + murmur → Marfan.

2. Analyze Cues

GH excess vs connective tissue defect. Cardiac risk = highest priority in Marfan; airway in acromegaly.

3. Prioritize Hypotheses

Marfan + chest pain → aortic dissection #1. Acromegaly + HA + vision loss → tumor expansion.

4. Generate Solutions

BP control, surgical referral, genetic counseling, post-op CSF leak prevention.

5. Take Action

Stop activity, notify HCP, position semi-Fowler's, prepare for emergency intervention.

6. Evaluate Outcomes

Stable VS, no CSF leak, IGF-1 normalizing, aortic root stable on echo, no visual loss.

5 Pharmacology

Octreotide / Lanreotide

SOMATOSTATIN ANALOG

Use: Acromegaly & gigantism — first-line med

MOA: Mimics somatostatin → suppresses GH release

S/E: GI upset, gallstones, bradycardia, hyperglycemia

Nursing: Monitor glucose, gallbladder US, give SQ/IM as ordered

Bromocriptine / Cabergoline

DOPAMINE AGONIST

Use: Acromegaly (adjunct), prolactinomas

MOA: Stimulates D2 receptors → ↓ GH and prolactin

S/E: Orthostatic hypotension, N/V, dizziness

Nursing: Take with food, change positions slowly, fall risk

Pegvisomant

GH RECEPTOR ANTAGONIST

Use: Acromegaly when octreotide fails

MOA: Blocks GH receptor → IGF-1 normalizes

S/E: ↑ LFTs, injection-site reaction

Nursing: Monthly LFTs; SQ daily injection

Beta-Blockers (Atenolol, Propranolol)

MARFAN — CARDIO PROTECTION

Use: #1 drug class for Marfan — slows aortic root dilation

MOA: ↓ HR & ↓ shear stress on aortic wall

S/E: Bradycardia, hypotension, fatigue, bronchospasm

Nursing: Hold if HR < 60 or SBP < 90; do NOT stop abruptly

Losartan / ARBs

MARFAN — ADJUNCT/ALTERNATIVE

Use: Adjunct or alternative to beta-blockers in Marfan

MOA: Blocks angiotensin II → ↓ aortic wall stress; modulates TGF-β

S/E: Hyperkalemia, dizziness, ↓ renal function

Nursing: Monitor K+, BUN/Cr, BP

Hydrocortisone (post-op)

GLUCOCORTICOID REPLACEMENT

Use: After hypophysectomy — ACTH may be lost

MOA: Replaces cortisol

S/E: Hyperglycemia, immunosuppression, mood changes

Nursing: Stress-dose during illness; never stop abruptly

6 NCLEX Test Traps ⚠️

Trap #1: "Tall = Marfan, always"

Acromegaly clients are usually **NOT taller than baseline** because plates are closed. They thicken (jaw, hands, feet). Gigantism = the actually tall GH disorder. Marfan = tall + thin + arachnodactyly + cardiac findings.

Trap #2: Confusing acromegaly with Cushing's

Both can cause facial changes & hyperglycemia, but Cushing's = moon face + buffalo hump + truncal obesity. Acromegaly = jaw protrusion (prognathism) + enlarged tongue + ring/shoe ↑.

Trap #3: Marfan client says "I just want to play football"

NEVER endorse contact sports, isometric exercises, or weightlifting. The aorta can rupture. The "best" answer = swimming, walking, yoga (gentle), low-impact cardio.

Trap #4: Post-op hypophysectomy — "encourage coughing & deep breathing"

WRONG. Coughing, sneezing, blowing nose, and bending are **contraindicated** ×2 weeks (CSF leak risk). Deep breathing alone is fine; incentive spirometry is preferred.

Trap #5: Forgetting DI after hypophysectomy

The pituitary releases ADH. Surgery → DI → polyuria + low specific gravity + hypernatremia. Watch for urine output > 200 mL/hr — that's reportable!

Trap #6: "Marfan + sudden chest pain — give nitroglycerin"

WRONG priority. Sudden tearing chest/back pain in Marfan = **aortic dissection**. Do NOT delay — call provider/RRT immediately. Nitro is not the priority; surgical evaluation is.

Trap #7: "Octreotide is curative"

It is not. Surgery (transsphenoidal hypophysectomy) is the **first-line definitive treatment** for the adenoma. Meds and radiation are for non-surgical candidates or residual disease.

Trap #8: Visual changes ≠ "ophthalmology referral, routine"

Bitemporal hemianopia = pituitary tumor compressing optic chiasm. Sudden visual loss + HA = **pituitary apoplexy** = neurosurgical emergency.

7 Patient & Family Education

Acromegaly / Gigantism

- ✓ Lifelong monitoring of GH and IGF-1 levels
- ✓ Annual colonoscopy (↑ colon polyp risk)
- ✓ Sleep study — high apnea risk
- ✓ Diabetes self-monitoring & diet
- ✓ Soft-tissue changes are partially reversible after treatment
- ✓ Report severe HA, vision changes, or sudden weakness

Post-Hypophysectomy

- ✓ NO nose blowing, sneezing through nose, brushing teeth ×2 wk
- ✓ Use soft toothettes; rinse mouth gently
- ✓ Avoid bending at waist, straining, lifting > 5–10 lb
- ✓ Report clear nasal drainage (CSF leak)
- ✓ Take lifelong hormone replacement as prescribed
- ✓ "Stress-dose" steroids during illness/surgery

Marfan Syndrome

- ✓ Annual echocardiogram + ophthalmology exam
- ✓ NEVER skip beta-blocker — reduces aortic risk
- ✓ NO contact sports, weightlifting, scuba diving
- ✓ Wear MedicAlert bracelet
- ✓ Pregnancy = high-risk; pre-conception cardiac eval
- ✓ Report sudden chest/back pain, dyspnea, or vision change immediately

Genetic Counseling (Marfan)

- ✓ Autosomal dominant — 50% inheritance per child
- ✓ First-degree relatives should be screened
- ✓ Genetic testing available (FBN1)
- ✓ Family planning conversations recommended
- ✓ Connect with The Marfan Foundation for support

8 Memory Boosters & Mnemonics

"BIG HANDS" — Acromegaly Findings

B — Bitemporal hemianopia (tumor compresses chiasm)

I — Insulin resistance / hyperglycemia

G — Growth Hormone elevated; Goiter possible

H — Headache, Hypertension, HF

A — Arthralgia, Apnea (sleep)

N — Nose / jaw enlarged (prognathism)

D — Diabetes, Deep voice

S — Sweating, Soft-tissue swelling, Shoe/ring size ↑

"MARFAN" — Key Features

M — Mitral valve prolapse / Murmur

A — Aortic aneurysm / dissection 🚨

R — Retinal detachment / lens dislocation

F — Fibrillin-1 defect (FBN1 gene)

A — Arachnodactyly (long spider fingers)

N — Narrow long face, high palate

Pattern Recognition Tips

Adult + facial coarsening + ring won't fit: Acromegaly

Child + extreme growth spurt + tall parents NOT: Gigantism

Tall + thin + heart murmur + lens problem: Marfan

Sudden tearing chest/back pain in Marfan: Aortic dissection — emergency!

Clear drip from nose post-pituitary surgery: CSF leak — test for glucose

Polyuria + low SG + ↑ Na post-pituitary surgery: DI

"Steinberg & Walker-Murdoch" — Marfan Bedside Tests

Steinberg Thumb Sign: Thumb folded across palm extends beyond ulnar border of hand → positive

Walker-Murdoch Wrist Sign: Thumb & pinky overlap when wrapped around opposite wrist → positive

Both indicate excessive limb/digit length consistent with Marfan

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