NGN NCLEX 2026

Down Syndrome (Trisomy 21)

SYSTEM: GENETIC / PEDIATRIC / DEVELOPMENTAL · NCJMM CLINICAL JUDGMENT ALIGNED

PEDIATRICS | GENETIC DISORDER | LIFESPAN CARE | FAMILY TEACHING

📚 SOURCE TRANSPARENCY

This topic is not present in Diane's uploaded personal notes. Content is synthesized from standard NCLEX references: *Saunders Comprehensive Review for the NCLEX-RN 2026*, *ATI RN Pediatric Nursing*, *Lippincott Manual of Nursing Practice*, the *American Academy of Pediatrics Clinical Report on Health Supervision for Children with Down Syndrome* (2022), and the *National Down Syndrome Society (NDSS)* clinical guidelines.

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Definition / Overview

WHAT IT IS · WHY IT MATTERS

Down syndrome is a **chromosomal disorder** caused by the presence of a full or partial extra copy of **chromosome 21**. It is the most common chromosomal cause of intellectual disability.

- **Incidence:** ~1 in 700 live births; risk rises sharply with maternal age (especially >35 years).
- **Three genetic types:** Trisomy 21 (nondisjunction, ~95%), Translocation (~3–4%, can be inherited), Mosaicism (~1–2%, milder features).
- **Why it matters for NCLEX:** the nurse must recognize physical features, monitor for associated systemic conditions (cardiac, GI, endocrine, hematologic, cervical-spine), and support the family across the lifespan.

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Pathophysiology

STEP-BY-STEP MECHANISM

An error during meiosis (**nondisjunction**) results in a gamete carrying two copies of chromosome 21. After fertilization, the embryo has **three copies** (trisomy) instead of two, producing systemic effects on growth, development, and organ formation.

1. MEIOSIS ERROR
Chromosome 21 fails to separate during egg or sperm formation (nondisjunction).

→

2. EXTRA CHROMOSOME
Gamete carries 24 chromosomes instead of 23.

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3. FERTILIZATION
Zygote forms with 47 chromosomes (3 copies of #21).

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4. OVEREXPRESSION
Genes on chromosome 21 produce excess proteins → altered development.

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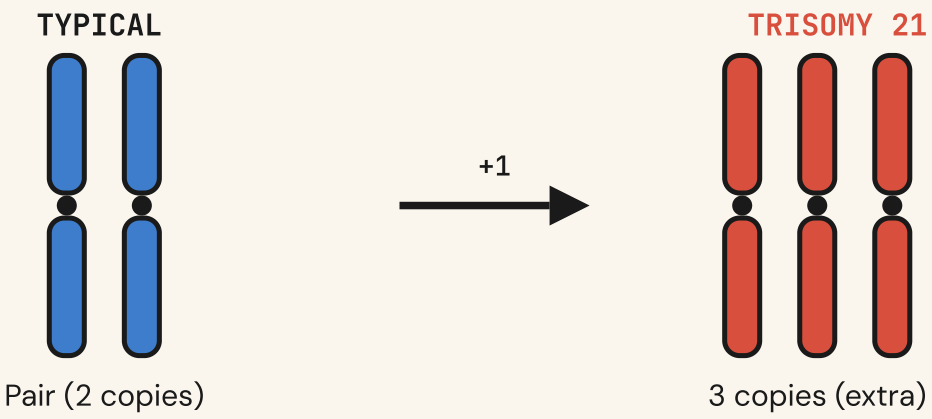
5. MULTISYSTEM EFFECTS
Brain, heart, GI tract, immune & endocrine systems all affected.



CHROMOSOME 21 — TYPICAL PAIR VS. TRISOMY 21

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Signs & Symptoms

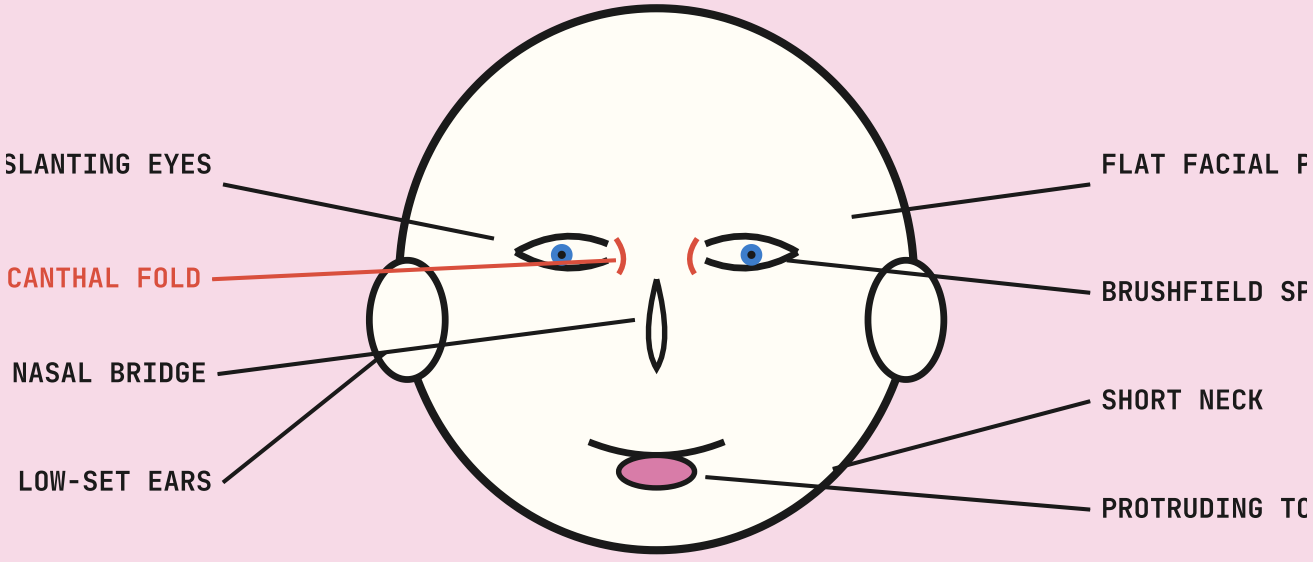
PHYSICAL FEATURES & ASSOCIATED CONDITIONS

🧠 Physical / Facial Features

- Flattened facial profile, small nose with low nasal bridge
- **Epicanthal folds** & upslanting palpebral fissures
- Brushfield spots (white specks on iris)
- Small low-set ears; small mouth with protruding tongue
- Short neck with excess skin at the nape
- **Single transverse palmar crease** (simian crease)
- Short broad hands; wide space between 1st & 2nd toes (sandal-gap)
- Short stature; **generalized hypotonia** ("floppy" infant)

🚑 Associated Systemic Conditions

- **Congenital heart defects (~50%)** — AV septal defect, VSD, ASD, PDA, tetralogy of Fallot
- **GI anomalies** — duodenal atresia, Hirschsprung disease, imperforate anus, GERD, celiac
- **Hypothyroidism** (congenital and acquired)
- **Hearing & vision deficits** — recurrent otitis media, cataracts, strabismus, refractive errors
- **Hematologic/Immune** — ↑ risk leukemia (ALL/AML), recurrent infections
- **Atlantoaxial instability** — C1–C2 subluxation risk 🚑
- **Obstructive sleep apnea** (large tongue, hypotonia)
- **Early-onset Alzheimer's disease** (adulthood)



CLASSIC PHYSICAL FEATURES — MEMORIZE FOR RECOGNITION ITEMS

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Priority Nursing Interventions

ABC FRAMEWORK · SAFETY · NCLEX PRIORITIZATION

🚑 A — Airway

- Suction nares before feedings (mouth breathers due to large tongue & nasal congestion).
- Monitor for **obstructive sleep apnea**; refer for polysomnography by age 4.
- Position upright after feeds (↓ aspiration risk).
- Cool-mist humidifier to loosen secretions.

🚑 B — Breathing

- Auscultate lung sounds q-shift; ↑ risk respiratory infection (immune dysfunction).
- Monitor SpO_2; teach chest physiotherapy as ordered.
- Keep up to date on RSV, influenza, pneumococcal vaccines.

🚑 C — Circulation

- Assess for **congenital heart defect** signs — murmur, cyanosis, poor feeding, diaphoresis with feeds, FTT.
- Daily weights; monitor for HF (tachycardia, tachypnea, hepatomegaly).
- SBE prophylaxis as ordered before dental work.

✅ Feeding & Growth (Infant)

- Use Down-syndrome-specific growth charts.
- Hypotonia + protruding tongue → slow, frequent feeds; use long, straight nipple.
- Burp frequently; allow rest periods.
- Watch for poor suck, choking, FTT — refer to feeding/lactation specialist.
- Screen for **celiac disease** after age 2 if symptomatic.

✅ Developmental & Safety

- Early intervention (PT, OT, speech) **started as early as possible**.
- Anticipatory guidance — milestones reached but later than typical.
- **Atlantoaxial precautions:** avoid contact sports, trampolines, somersaults until cleared with cervical X-ray.
- Routine vision and hearing screening (every 6 months in infancy).
- Annual TSH; CBC at birth, 6 mo, 12 mo (leukemia surveillance).

⚠️ RED FLAGS — Notify Provider Immediately

- New or worsening **neck pain, torticollis, gait change, weakness, bowel/bladder change** → suspect atlantoaxial subluxation.
- Cyanosis, poor feeding, sweating with feeds in infant → cardiac decompensation.
- **Bilious vomiting**, no meconium, abdominal distension in newborn → duodenal atresia / Hirschsprung.

- Bruising, petechiae, pallor, fatigue → leukemia work-up.

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Pharmacology & Associated Therapies

NO DRUG "TREATS" DOWN SYNDROME — MANAGE COMORBIDITIES

DRUG / CLASS	WHY IT'S USED	KEY NURSING CONSIDERATIONS
Levothyroxine (Synthroid)	Congenital or acquired hypothyroidism (common in DS).	Give morning on empty stomach; monitor TSH q6–12 mo; check growth; teach lifelong therapy.
Digoxin / Furosemide / ACE inhibitor	Heart failure from congenital heart defect.	Apical pulse 1 min before digoxin (hold if <90 infant, <70 child); monitor K^+ with furosemide; watch for dig toxicity.
Antibiotic prophylaxis (amoxicillin)	SBE prevention before dental/invasive procedures in select cardiac lesions.	Confirm allergy history; teach family dental hygiene importance.
Stool softeners / laxatives (docusate, polyethylene glycol)	Chronic constipation (hypotonia, low fiber, hypothyroidism).	Encourage fluids, fiber, activity; do not use stimulants long-term.
Vaccines (RSV/nirsevimab, influenza, pneumococcal, COVID-19)	↑ risk respiratory infection due to immune dysfunction.	Follow standard pediatric schedule + RSV protection in infancy; defer live vaccines if immunocompromised.
Chemotherapy (when applicable)	Acute leukemia (ALL or AML-M7).	Children with DS more sensitive to methotrexate toxicity — close labs, mucositis care.

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NCLEX Test Traps

WRONG-VS-RIGHT THINKING PATTERNS

✘ WRONG THINKING	✓ RIGHT THINKING
"The child has Down syndrome — they need 'special' or 'protected' care; limit normal activities."	Encourage **normal developmental experiences** with adaptations. Only restrict contact sports/somersaults **if atlantoaxial instability** is confirmed.
"Choose the answer that puts the child in a quiet, isolated environment."	Pick interventions that **promote socialization, early intervention, and family-centered care**.
"All children with DS look the same and have the same problems."	Features and severity **vary** (especially in mosaicism). Always assess the individual child's findings.
"Floppy infant — that's just hypotonia, monitor and reassess later."	Hypotonia + poor feeding + cyanosis with feeds = **screen for congenital heart disease NOW**. Cardiac assessment is priority.
"Speak to the parents about the child only — don't address the child."	Speak **directly to the child** in age-appropriate language; respect dignity. Use **person-first language** ("child with Down syndrome," not "Down's child").
"Newborn with DS is vomiting — likely overfeeding."	**Bilious vomiting** in a DS newborn = duodenal atresia until proven otherwise. NPO, NG decompression, surgical consult.
"All children with DS will look the same as adults — no new health surveillance needed."	Lifelong surveillance: **annual thyroid, vision/hearing, cervical-spine review, dementia screening after age 40**.

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Patient & Family Education

LIFESPAN TEACHING PRIORITIES

👶 Newborn / Infant Period

- Allow parents time to grieve, ask questions, and bond — avoid sharing diagnosis when parents are alone or distressed.
- Use **person-first language**: "your baby with Down syndrome."
- Encourage feeding success: positioning, smaller more frequent feeds, lactation referral.
- Teach signs of cardiac trouble — sweating with feeds, blue spells, poor weight gain.
- Provide local Down syndrome support group resources & early-intervention referral.

👧 Childhood / School Age

- Promote independence in ADLs; routines and visual schedules help.
- IEP (Individualized Education Program) — partner with school.
- Dental hygiene critical (delayed eruption, crowding, periodontal disease).
- Skin care — dry skin prone to cracking; moisturize daily.
- Routine screenings: thyroid yearly, hearing/vision every 1–2 years, sleep study by age 4.

👦 Adolescence / Adulthood

- Address puberty, sexuality, consent, and contraception openly and at appropriate level.
- Encourage **employment, social activity, independent living** per ability.
- Annual cardiac, thyroid, and mental health assessment (↑ depression risk).
- Plan transition to adult primary care.

👨‍👩‍👧‍👦 Family Support

- Respite care, sibling support, financial/legal planning (guardianship, special-needs trust).
- Genetic counseling for parents — recurrence risk depends on type (translocation has higher risk).
- Encourage realistic optimism — many adults with DS lead full, meaningful lives.

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Memory Boosters

MNEMONICS & PATTERN RECOGNITION

CHILD

- **C** ardiac defects (50%)
- **H** ypotonia + Hypothyroidism
- **I** ntellectual disability
- **L** eukemia risk + Low-set ears
- **D** uodenal atresia + Developmental delay

FACES

- **F** lat facial profile
- **A** lmond-shaped (upslanting) eyes + epicanthal folds
- **C** rease — single transverse palmar (simian)
- **E** ars low-set / small
- **S** hort neck, short stature, sandal-gap toes

"21 = 3 copies"

21 reminds you of chromosome 21.

3 copies = Trisomy. Three is the magic number → "Trisomy 21."

World Down Syndrome March **21** is **Day** (3/21 = 3 copies of 21).

ATLANTOAXIAL — "Neck NO-GOs"

- **N** o trampolines
- **O** mit somersaults & diving
- **G** ymnastics & contact sports — clear with X-ray first
- **O** bserve for neck pain, weakness, gait change → notify provider

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NCJMM Clinical Judgment Scenario

SIX-STEP NEXT-GENERATION NCLEX WALKTHROUGH

CASE

A 6-month-old infant with Down syndrome (trisomy 21) is brought to the pediatric clinic by his mother for a routine well-baby visit. Birth weight was 3.2 kg; current weight is 5.4 kg (below the 5th percentile on DS growth chart). Mother reports the baby "tires out easily" during bottle feeds, sweats around the forehead, and has been less active for 2 weeks. On assessment: HR 168, RR 62, SpO_2 92% on room air, mild circumoral cyanosis, a III/VI holosystolic murmur at the LLSB, and palpable liver edge 2 cm below the costal margin.

STEP 1

Recognize Cues

Relevant cues: tachycardia (168), tachypnea (62), SpO_2 92%, circumoral cyanosis, diaphoresis with feeds, holosystolic murmur LLSB, hepatomegaly, poor weight gain on DS-specific chart. **History:** Down syndrome (↑ cardiac defect risk).

STEP 2

Analyze Cues

The cluster — murmur + feeding intolerance + tachypnea + hepatomegaly + failure to thrive — is the classic presentation of a **congenital heart defect with developing heart failure**. Most likely defect in DS: AV septal defect or VSD with left-to-right shunt and pulmonary overcirculation.

STEP 3

Prioritize Hypotheses

Highest priority: congestive heart failure secondary to a congenital heart defect. **Lower-priority differentials:** respiratory infection, anemia, hypothyroidism contributing to fatigue. Cardiac assessment cannot be deferred — perfusion is at risk.

STEP 4

Generate Solutions

Anticipate: cardiology referral, echocardiogram, ECG, chest X-ray, BNP and CBC, oxygen support if SpO_2 falls below 90% (cautious in left-to-right shunts), small frequent feeds with high-calorie formula, weigh daily, diuretic (furosemide) and possibly digoxin and ACE inhibitor per cardiology, surgical consult for definitive repair.

STEP 5

Take Action

Now: place on cardiorespiratory monitor and pulse oximetry, elevate head of crib, cluster care to minimize energy expenditure, obtain IV access, notify pediatric cardiology and provider, prepare family with calm clear teaching, hold oral feeds pending evaluation if respiratory distress worsens, document accurate weight.

STEP 6

Evaluate Outcomes

Expected improvement: HR <160, RR <60, SpO_2 ≥95%, no diaphoresis with feeds, weight gain of 20–30 g/day, no worsening hepatomegaly, family verbalizes understanding of plan. **Failure cues:** worsening cyanosis, lethargy, falling

SpO_2, decreased UOP → escalate to higher level of care.

DOWN SYNDROME (TRISOMY 21) · NGN NCLEX 2026 · DIANE'S VISUAL STUDY LIBRARY
SOURCES: SAUNDERS 2026 · ATI PEDIATRIC NURSING · LIPPINCOTT · AAP HEALTH SUPERVISION REPORT 2022 · NDSS