NGN NCLEX 2026 | GENETIC / PEDIATRIC | HIGH-YIELD | MATERNAL-NEWBORN

Edwards Syndrome *(Trisomy 18)*

A severe genetic disorder caused by a third copy of chromosome 18 — recognize the classic constellation, prioritize airway & family-centered palliative care.

INCIDENCE	SEX RATIO	SURVIVAL
~1 in 5,000–6,000 live births	3:1 Female > Male	~50% die < 1 wk · 90–95% < 1 yr

RISK FACTOR
Advanced maternal age

SOURCE TRANSPARENCY

Edwards syndrome was not part of your uploaded notes. Content has been compiled from standard NCLEX references: *Saunders Comprehensive Review for the NCLEX-RN (2026)*, *ATI Maternal Newborn Nursing*, *Lippincott Manual of Nursing Practice*, ACOG / CDC clinical guidance, and the SOFT (Support Organization for Trisomy) family resources.

1 Definition & Overview

WHAT IT IS · WHY IT MATTERS

WHAT IS IT?

A chromosomal disorder where the cells contain **three copies of chromosome 18** instead of two — leading to severe multi-system congenital anomalies.

HOW COMMON?

2nd

Most common autosomal trisomy after Down syndrome (Trisomy 21).

WHY IT MATTERS

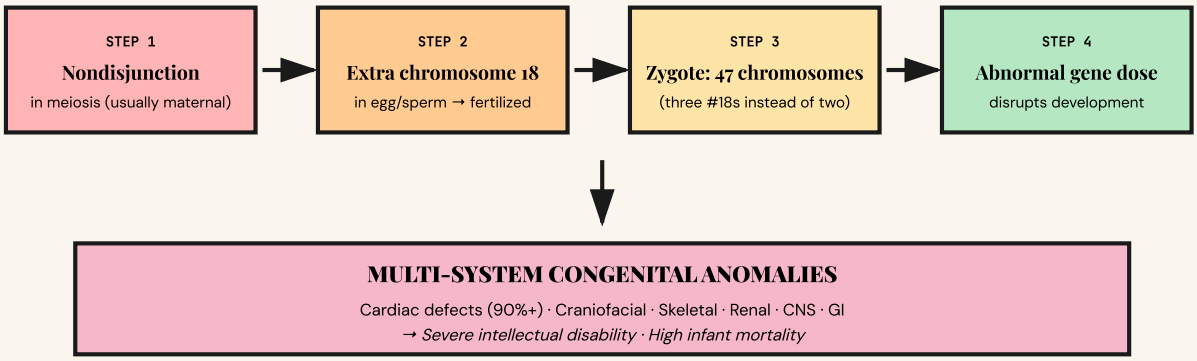
Carries one of the **highest infant mortality rates** of any genetic condition. Care is often focused on **comfort & palliation**, not cure.

WHO'S AFFECTED?

Female infants are affected ~3× more often than males. Risk rises with **advanced maternal age**; most cases are sporadic (not inherited).

2 Pathophysiology (Simplified)

STEP-BY-STEP MECHANISM



FULL TRISOMY 18 (~95%)

Extra chromosome 18 in **EVERY CELL**. Most severe form; nearly all classic features present.

MOSAIC TRISOMY 18 (~5%)

Extra chromosome in **SOME CELLS ONLY**. Highly variable severity; some survive longer.

PARTIAL TRISOMY 18 (RARE)

Only a **PORTION** of an extra chromosome 18 is present. Features depend on which segment is duplicated.

3 Signs & Symptoms

CLASSIC CONSTELLATION · RED FLAGS

★ Classic / Diagnostic Hallmarks

- ★ **Clenched fists** with overlapping fingers (index over middle, pinky over ring)
- ★ **Rocker-bottom feet** (rounded sole)
- ★ Low-set, malformed ears
- ★ Microcephaly & prominent occiput
- ★ Micrognathia (small jaw) & microstomia
- ★ Short sternum
- ★ Low birth weight (< 5.5 lb at term)

Prenatal Indicators

- IUGR — intrauterine growth restriction
- Polyhydramnios (excess amniotic fluid)
- Small placenta
- Abnormal ultrasound: cardiac defects, choroid plexus cysts, clenched fists visible on imaging
- Abnormal quad screen: ↓ AFP, ↓ estriol, ↓ hCG, ↓ inhibin A

Systemic Findings

- **Cardiac (90%+):** VSD, ASD, PDA, tetralogy of Fallot
- **Renal:** Horseshoe kidney, polycystic kidney
- **GI:** Omphalocele, malrotation, esophageal atresia
- **CNS:** Severe intellectual disability, hypotonia → hypertonia, seizures
- Cleft lip / palate (variable)
- Clubfoot, hip dislocation

🚨 RED FLAG Findings

- 🚨 **Apnea episodes** — central respiratory drive failure
- 🚨 **Cyanosis** — cardiac defect / poor perfusion
- 🚨 **Feeding intolerance / aspiration** — poor suck-swallow
- 🚨 **Failure to thrive** — inadequate nutrition + cardiac demand
- 🚨 **Seizure activity**
- 🚨 **Hypothermia** — thermoregulation failure

4 Diagnostics

PRENATAL & POSTNATAL WORKUP

Prenatal Screening & Testing

NIPT / cell-free DNA — non-invasive, screens maternal blood ≥ 10 wk gestation

First-trimester screen — PAPP-A, β-hCG, nuchal translucency (11–13 wk)

Quad screen (15–22 wk) — classic T18 pattern: ALL FOUR markers low

Ultrasound — IUGR, cardiac defects, choroid plexus cysts, clenched fists

CVS (10–13 wk) — diagnostic, invasive

Amniocentesis (15–20 wk) — definitive prenatal diagnosis

Postnatal Confirmation

Karyotype — gold standard / definitive (shows 47,XX,+18 or 47,XY,+18)

FISH — rapid (24–48 hr) chromosomal analysis

Chromosomal microarray (CMA) — detects partial trisomy

Echocardiogram — identify cardiac defects (almost always present)

Renal & abdominal ultrasound

Cranial ultrasound / MRI — CNS anomalies

Hearing screen, ophthalmology consult

5 Priority Nursing Interventions

ABC FRAMEWORK · SAFETY · FAMILY-CENTERED

A

AIRWAY

- → Position to maintain a patent airway (slight head extension; avoid neck flexion)
- → Suction nares/oropharynx PRN — micrognathia & small mouth predispose to obstruction
- → Apnea monitor at bedside continuously
- → Have bag-valve-mask at bedside; know unit resuscitation protocol

B

BREATHING

- → Monitor RR, work of breathing, SpO_2 continuously
- → Administer humidified O_2 per provider order to maintain SpO_2 within ordered range
- → Reposition q2h to optimize lung expansion and prevent atelectasis
- → Recognize central apnea vs. obstructive apnea — both common in T18

C

CIRCULATION

- → Continuous cardiorespiratory monitoring (cardiac defects in > 90%)
- → Assess perfusion: cap refill, color, peripheral pulses, peripheral vs. core temp gap
- → Monitor for signs of CHF: tachypnea, diaphoresis with feeds, hepatomegaly
- → Strict I&O; daily weights; assess for fluid overload

S

SAFETY / THERMOREGULATION / NUTRITION

- → Maintain thermoneutral environment — radiant warmer or isolette; T18 infants poorly thermoregulate
- → Skin care: turn q2h, support pressure points; skin is fragile
- → Position to prevent contractures (clenched fists, clubfoot) — gentle ROM, splinting if ordered
- → Feeding: small frequent feeds; NG / gavage often needed (poor suck-swallow-breathe coordination)
- → Aspiration precautions: HOB elevated during & 30 min after feeds

F

FAMILY-CENTERED & PALLIATIVE CARE

→ Provide developmentally supportive, individualized care — encourage parental holding, skin-to-skin

→ Facilitate goals-of-care conversation: comfort care vs. selected interventions

→ Involve neonatal palliative care, social work, chaplaincy, genetic counseling early

→ Connect family with peer support (e.g., SOFT — Support Organization for Trisomy)

→ Bereavement: memory-making (footprints, photos, locks of hair) when appropriate

→ Sibling support & anticipatory grief education

6 Pharmacology

SUPPORTIVE · SYMPTOM-TARGETED

DRUG / CLASS	MECHANISM (SIMPLIFIED)	KEY SIDE EFFECTS	NURSING CONSIDERATIONS
Furosemide (loop diuretic) — for CHF from cardiac defect	Inhibits Na/K/2Cl in loop of Henle → ↓ preload & pulmonary congestion	Hypokalemia, hyponatremia, dehydration, ototoxicity	Monitor K^+, I&O, daily weights, fontanels for dehydration
Digoxin — for CHF / cardiac failure	Positive inotrope; ↑ cardiac contractility, ↓ HR	Bradycardia, N/V, dysrhythmias; narrow therapeutic window (toxic > 2.0 ng/mL)	Apical HR x 1 min before dose; hold if HR < 90–110 (infant); monitor K^+ (hypoK ↑ toxicity)
Phenobarbital / **Levetiracetam** (anticonvulsants)	Suppress abnormal CNS electrical activity	Sedation, respiratory depression, hepatic effects	Monitor LOC, RR, SpO_2; therapeutic levels; have suction ready
Acetaminophen — comfort / fever	Central COX inhibition; antipyretic & analgesic	Hepatotoxicity (overdose)	Weight-based dosing; max 75 mg/kg/day infant
Morphine (opioid) — palliative comfort, dyspnea	μ-opioid agonist; analgesia & ↓ air hunger	Respiratory depression, sedation, constipation, hypotension	Goal is comfort, not sedation; titrate to symptoms; naloxone available; family education
Antibiotics (as needed)	Treat respiratory / UTI / aspiration infections — common	Allergy, organ toxicity (class-dependent)	Cultures before first dose when possible; weight-based dosing

KEY PRINCIPLE · There is no cure or disease-modifying drug for Trisomy 18. All pharmacotherapy is **SUPPORTIVE** — managing CHF, seizures, infections, and comfort. Decisions about which interventions to pursue are made *with the family*, guided by the infant's goals of care.

7 NCLEX Test Traps

WRONG VS. RIGHT THINKING

TRAP

"It's basically the same as Down syndrome — prepare the family for a long life with developmental delays."

CORRECT

Trisomy 18 is far more severe than Trisomy 21. ~50% die in the first week; 90–95% before age 1. Care planning centers on **comfort and palliation**, not long-term developmental services.

TRAP

"Refer the parents to genetic counseling so they understand they passed this to their baby."

CORRECT

~95% of cases are **sporadic nondisjunction**, not inherited. Genetic counseling is offered to discuss recurrence risk in **future pregnancies**, not to assign blame.

TRAP

"Rocker-bottom feet and overlapping fingers — that's Down syndrome."

CORRECT

Those are **classic Trisomy 18** findings. Down syndrome (T21) features include single transverse palmar crease, upslanting palpebral fissures, protruding tongue, and flat nasal bridge — different constellation entirely.

TRAP

"Newborn has murmur and weak cry — first action is to call the cardiologist for surgical evaluation."

CORRECT

Airway first. In an infant with suspected T18, priority is patent airway, then SpO_2 & perfusion. Cardiac consult follows, and surgical decisions are made jointly with the family in light of overall prognosis.

TRAP

"Polyhydramnios + IUGR is just a small baby with too much fluid — reassure the mother."

CORRECT

The combination of **IUGR + polyhydramnios** on prenatal ultrasound is a classic flag for Trisomy 18 (the fetus can't swallow effectively). Recommend further evaluation — NIPT, level II US, amniocentesis as appropriate.

TRAP

"Older paternal age is the major risk factor."

CORRECT

Advanced maternal age is the established risk factor for trisomies (T13, T18, T21) — most nondisjunction events occur in maternal meiosis I.

8 Patient & Family Education

HONEST · COMPASSIONATE · PRACTICAL

Understanding the Diagnosis

- What Trisomy 18 is in plain language
- Difference between full, mosaic, and partial T18
- Realistic prognosis without removing hope
- This is not anyone's fault — usually a random event

Daily Care at Home

- Safe positioning & gentle handling
- Small frequent feeds; recognizing aspiration signs
- Skin care, gentle ROM for clenched fists/feet
- Medication administration & storage

When to Seek Help

- Apnea > 20 sec, color change, limpness
- Fever, poor feeding, fewer wet diapers
- New or worsening seizure activity
- Increased work of breathing, persistent cyanosis

Care Planning & Decisions

- Goals of care: comfort vs. selected interventions
- Advance care planning & DNR discussions
- Hospice / pediatric palliative care options
- Family wishes & values guide all choices

Future Pregnancies

- Recurrence risk is generally low for full T18 (~1%)
- Genetic counseling recommended before conception
- NIPT and detailed US available in next pregnancy

Support & Coping

- SOFT — Support Organization for Trisomy (trisomy.org)
- Peer support & parent-to-parent connection
- Sibling support resources
- Bereavement counseling & memory-making

9 Memory Boosters

MNEMONICS · PATTERN RECOGNITION

E·D·W·A·R·D·S — Trisomy 18 features

- **E** **E**ars low-set & malformed
- **D** **D**efects of the heart (VSD, ASD, PDA)
- **W** **W**ee small mouth & jaw (micrognathia)
- **A** **A**rms with clenched fists, overlapping fingers
- **R** **R**ocker-bottom feet
- **D** **D**eath usually before age 1
- **S** **S**mall for gestational age (IUGR)

"3 Trisomies, 3 letters" — Quick recognition

P **T13 — Patau:** "P" for cleft **P**alate / lip + polydactyly + holoprosencephaly

E **T18 — Edwards:** "E" for low-set **E**ars + clench**E**d fists + rock**E**r-bottom feet

D **T21 — Down:** "D" for **D**own (most common, longest survival, single palmar crease)

Quad Screen Pattern (T18) — "ALL DOWN"

↓ AFP · ↓ Estriol · ↓ hCG · ↓ Inhibin A

Memory: **"Trisomy 18 — everything is low"**

Contrast: T21 (Down) → ↑ hCG & ↑ inhibin A, ↓ AFP & ↓ estriol ("two up, two down")

Priority anchor — "ABC then F"

Airway · **B**reathing · **C**irculation

Then **F**amily — palliative goals, parental presence, bereavement

Remember: in T18, *family-centered care is a priority intervention*, not an afterthought.

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NCJMM Clinical Judgment Scenario

SIX-STEP NGN MODEL · 2026 TEST PLAN

SCENARIO

A female newborn is delivered vaginally at 38 weeks gestation to a 39-year-old G3P3. Birth weight 1.9 kg (4 lb 3 oz). Apgar scores 5 at 1 min, 7 at 5 min. On initial assessment, the nurse notes microcephaly with a prominent occiput, low-set malformed ears, micrognathia, clenched fists with the index finger overlapping the middle and the fifth finger overlapping the fourth, and rocker-bottom feet. A grade III/VI systolic murmur is auscultated. SpO_2 88% on room air. The infant has had two apneic episodes > 20 seconds since birth. Prenatal records note IUGR and polyhydramnios on third-trimester ultrasound.

1 RECOGNIZE CUES

STEP 1

What is relevant?

Low birth weight at term · microcephaly with prominent occiput · low-set ears · clenched fists with overlapping fingers (**classic**) · rocker-bottom feet (**classic**) · cardiac murmur · SpO_2 88% · recurrent apnea · prenatal IUGR + polyhydramnios · advanced maternal age.

2 ANALYZE CUES

STEP 2

What do the cues mean together?

The constellation — micrognathia, low-set ears, overlapping clenched fists, rocker-bottom feet, IUGR, polyhydramnios, cardiac defect, advanced maternal age — strongly suggests **Trisomy 18 (Edwards syndrome)**. The murmur signals a likely congenital heart defect (most often VSD). SpO_2 88% and apnea indicate **active respiratory compromise**.

3 PRIORITIZE HYPOTHESES

STEP 3

What is the priority?

Highest: ineffective breathing pattern / risk for apnea — immediately life-threatening. **Next:** impaired gas exchange related to suspected cardiac defect. **Then:** confirm Trisomy 18 diagnosis and initiate family-centered goals-of-care planning. Long-term concerns (feeding, thermoregulation, attachment) follow.

4
GENERATE SOLUTIONS

STEP 4

What could be done?

Place on continuous cardiorespiratory + apnea monitor · administer humidified blow-by O_2 per protocol · position to optimize airway · obtain stat echocardiogram · draw karyotype / FISH for diagnostic confirmation · notify provider & neonatology · consult neonatal palliative care, social work, and genetic counseling · support parental presence and skin-to-skin if stable · prepare family for diagnostic conversation.

5
TAKE ACTION

STEP 5

What to do *now*?

1. Reposition for patent airway and apply continuous SpO_2 + apnea monitoring. **2.** Initiate humidified O_2 per provider order, titrating to ordered SpO_2 goal. **3.** Stabilize thermoregulation under radiant warmer. **4.** Obtain IV access; draw chromosomal studies and routine labs. **5.** Notify provider; request stat echocardiogram. **6.** Bring parents to the bedside; provide honest, compassionate explanation; arrange palliative care consult.

6
EVALUATE OUTCOMES

STEP 6

Did it work?

SpO_2 maintained within the ordered range with supplemental O_2 · apnea episodes resolve or are promptly responded to · infant remains normothermic · cardiac defect identified on echo and care plan adjusted accordingly · karyotype confirms diagnosis · family verbalizes understanding and participates in goals-of-care planning · interdisciplinary palliative care plan documented. Reassess each shift and modify priorities as the infant's condition and family's wishes evolve.

11

Compare & Contrast: The 3 Trisomies

T13 · T18 · T21 AT A GLANCE

	TRISOMY 13 — PATAU	TRISOMY 18 — EDWARDS	TRISOMY 21 — DOWN
INCIDENCE	~1 in 10,000–16,000	**~1 in 5,000–6,000**	~1 in 700 (most common)
SURVIVAL	~80% die < 1 yr	**90–95% die < 1 yr**	Often into adulthood
CLASSIC FEATURES	Cleft lip/palate, polydactyly, holoprosencephaly, microphthalmia, scalp defects (cutis aplasia)	**Clenched fists with overlapping fingers, rocker-bottom feet, low-set ears, micrognathia, prominent occiput**	Upslanting palpebral fissures, flat nasal bridge, single palmar crease, protruding tongue, hypotonia
CARDIAC DEFECTS	~80% (VSD, PDA)	**> 90% (VSD most common)**	~40–50% (AV canal defect classic)
QUAD SCREEN	Variable	**↓ AFP · ↓ Estriol · ↓ hCG · ↓ Inhibin A (all low)**	↓ AFP · ↓ Estriol · ↑ hCG · ↑ Inhibin A
MEMORY HOOK	"P" — **P**atau, **P**alate, **P**olydactyly	**"E" — Edwards, low-set Ears, clenchEd fists**	"D" — **D**own, **D**evelopmental delay, longest survival