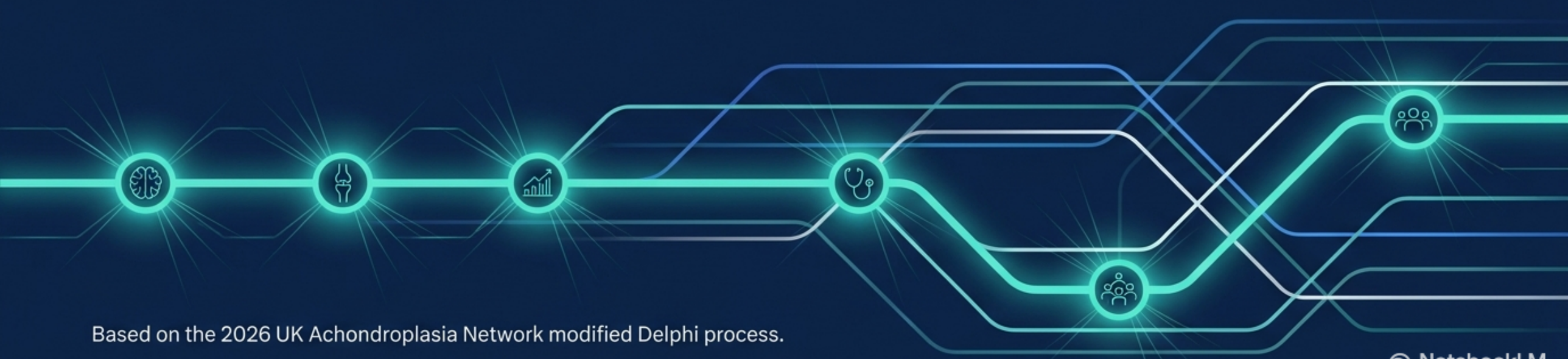
The Anticipatory Blueprint: UK Guidelines for Achondroplasia

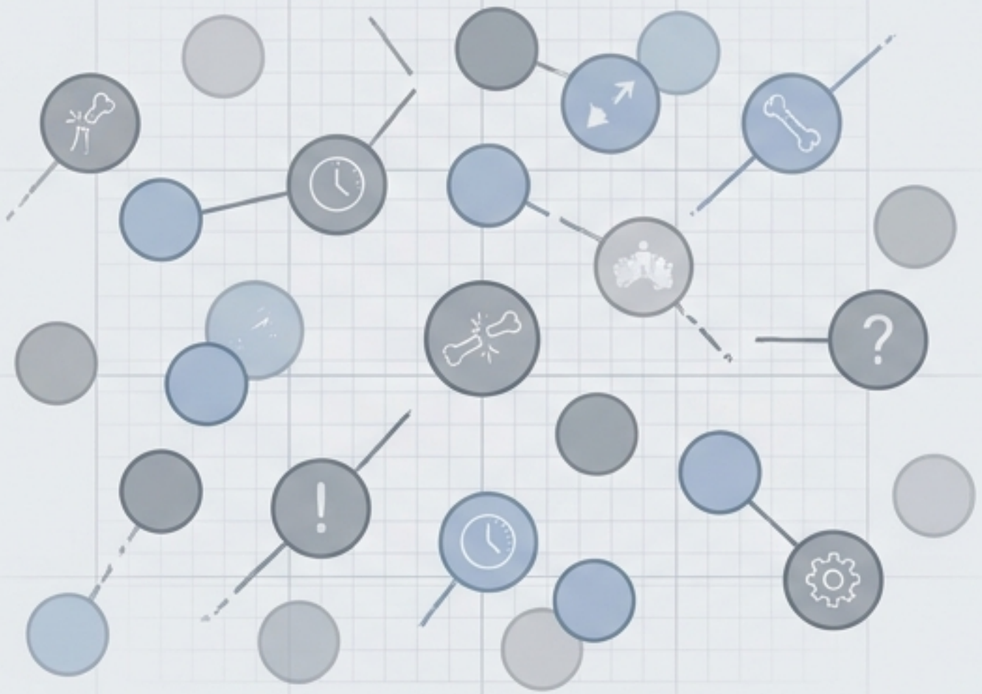
The first multidisciplinary consensus for the standardized care of children and young people



Based on the 2026 UK Achondroplasia Network modified Delphi process.

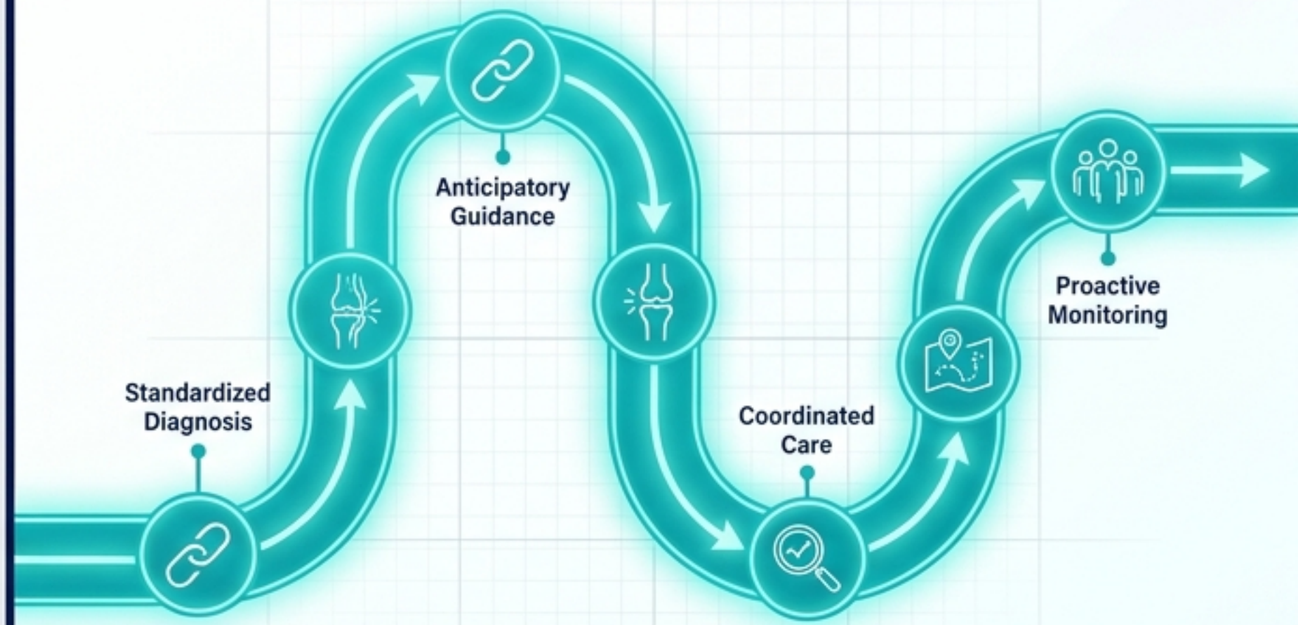
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The Challenge



The UK sees ~**26** new **Achondroplasia (ACH) diagnoses** annually. Historically, multidisciplinary care has lacked national standardization, leading to **preventable morbidity.**

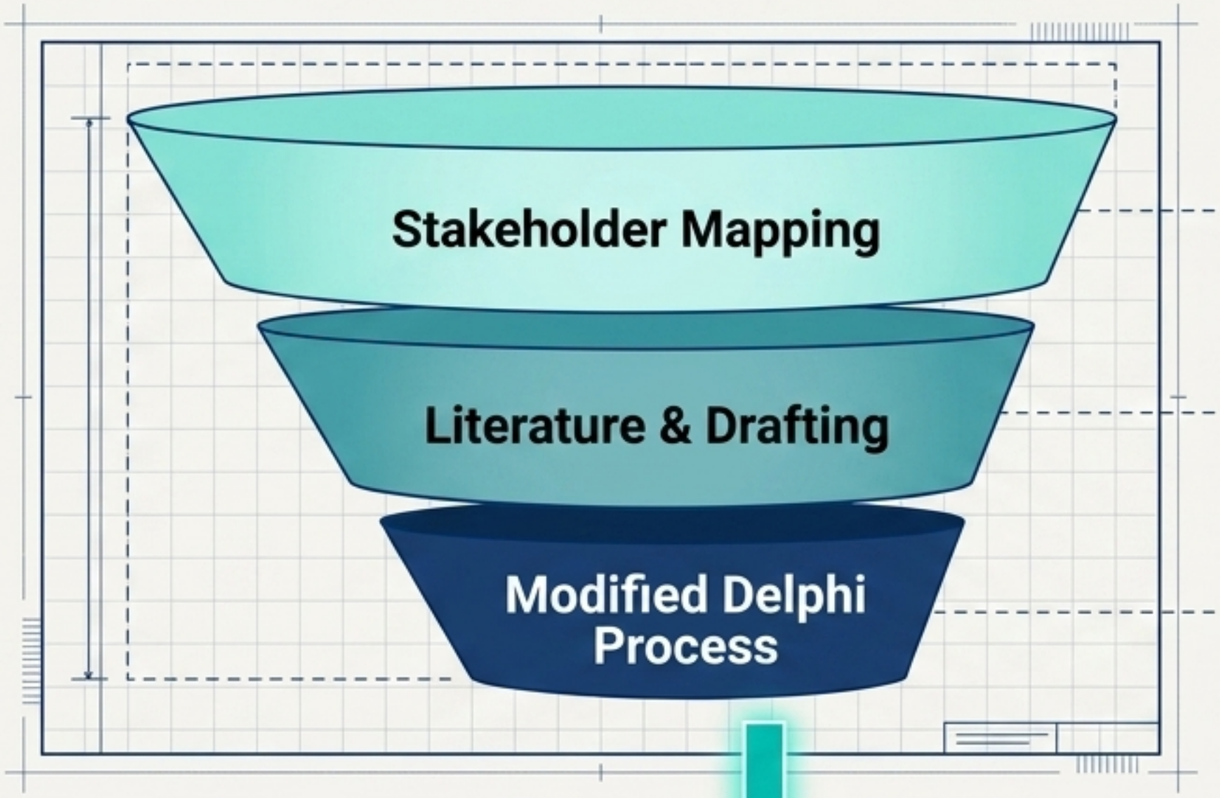
The Solution



A **unified, anticipatory model.**
24 expert consensus statements designed to **shift care from reactive to proactive.**

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Methodological Rigor



Multidisciplinary expert group (27 HCPs) + 2 patient advocacy groups map the care pathway.

Integration of international guidelines with UK healthcare system realities.

25 multidisciplinary UK experts across 16 specialties. Two anonymous voting rounds.

24 Final Statements

Pre-defined consensus of ≥80% agreement (21 statements achieved 100% agreement).

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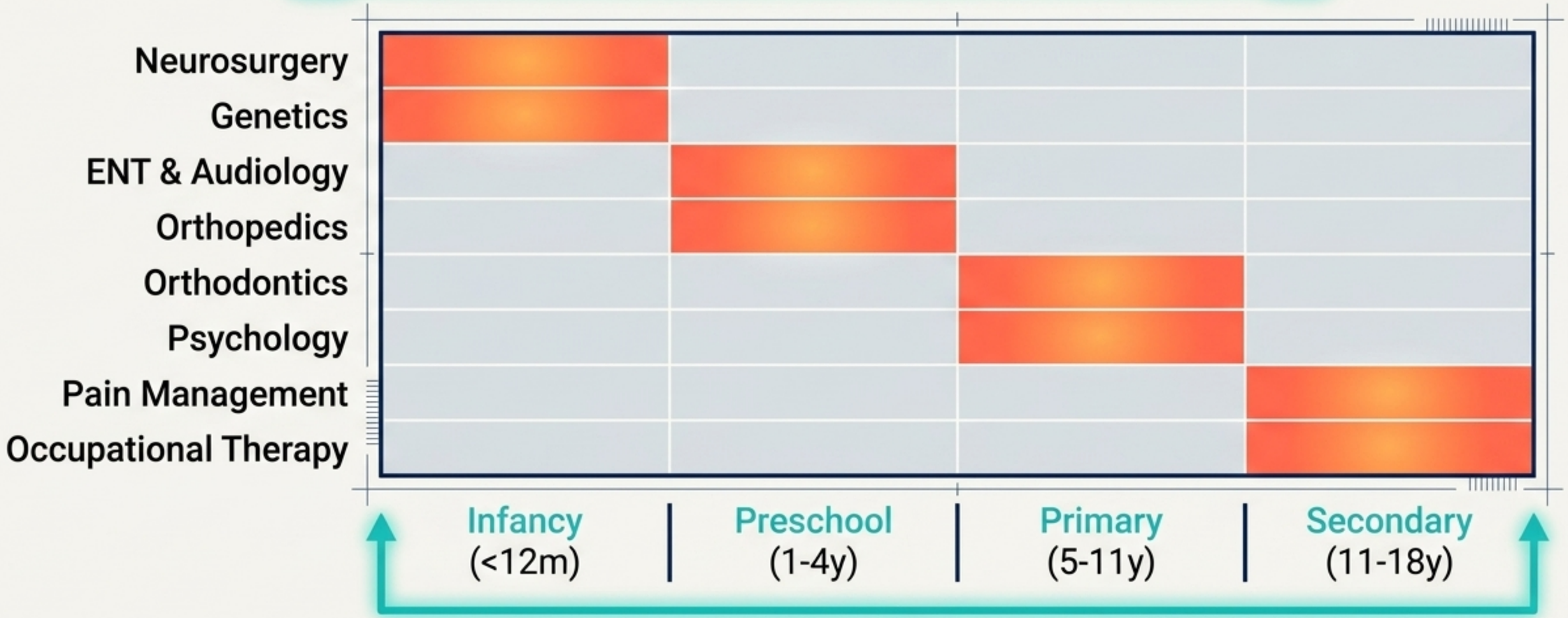
Anatomical Feature vs. Clinical Impact

Genetics	Anatomical Feature	Functional Impact
FGFR3 Variant	 Smaller, misshapen foramen magnum	Brainstem compression, central apnea, sudden infant death risk.
FGFR3 Variant	 Midface hypoplasia & short, horizontal eustachian tubes	Obstructive sleep apnea, recurrent otitis media, conductive hearing loss.
FGFR3 Variant	 Abnormal endochondral ossification	Disproportionate short stature, tibial bowing, thoracolumbar kyphosis.

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The Multidisciplinary Shift

As the child grows, the center of clinical gravity evolves. Care must anticipate the next developmental hurdle.



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The Diagnostic Pathway

Path A: Family History of ACH

- Pre-conception clinical genetics referral
- 11-14wk Dating Scan & NIPD
- Fetal medicine management

Path B: No Family History

- Standard Dating Scan
- 18-22wk Anomaly Scan
- 3rd Trimester suspicion: short long bones, relative macrocephaly
- Referral to fetal medicine / clinical genetics

⚠ **Postnatal Protocol:** Do not delay specialist referral while awaiting genetic test results; rely on classical clinical/radiological features. Ensure basic life support training before ward discharge.

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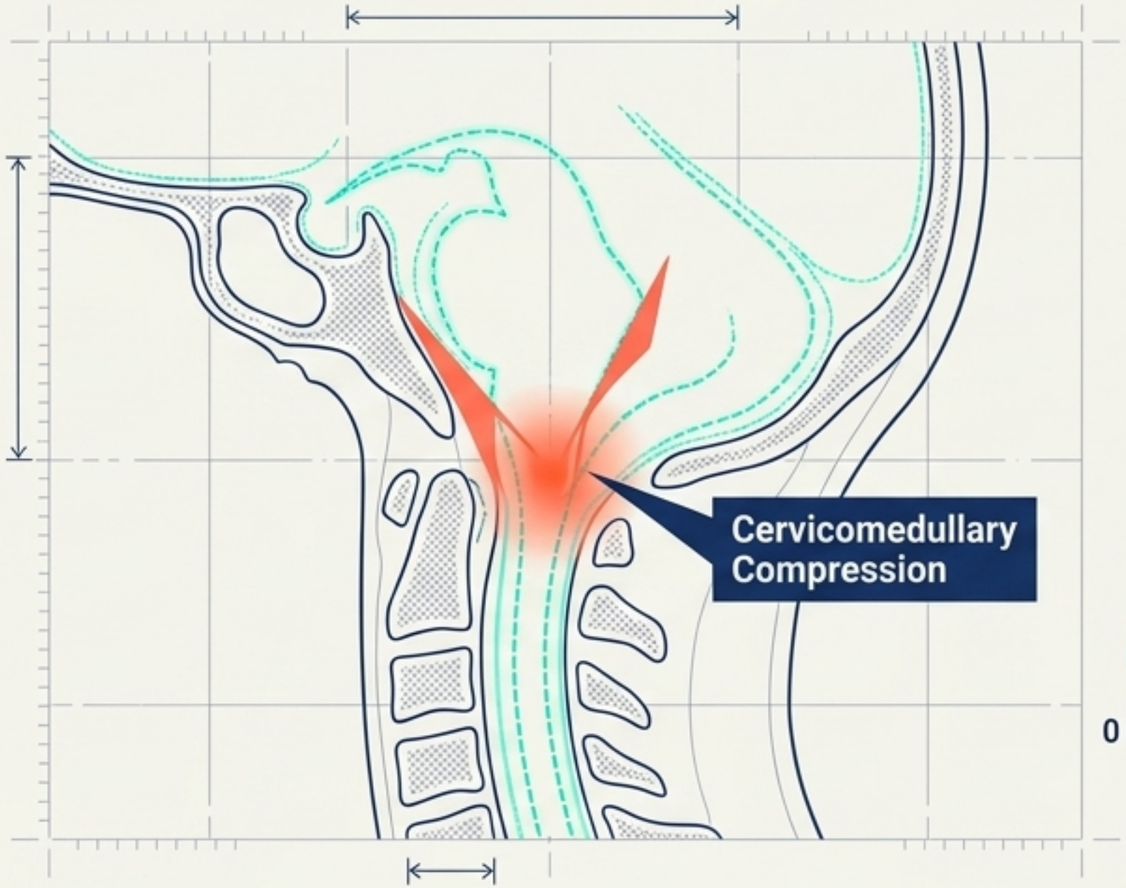
Infancy (<12 Months)

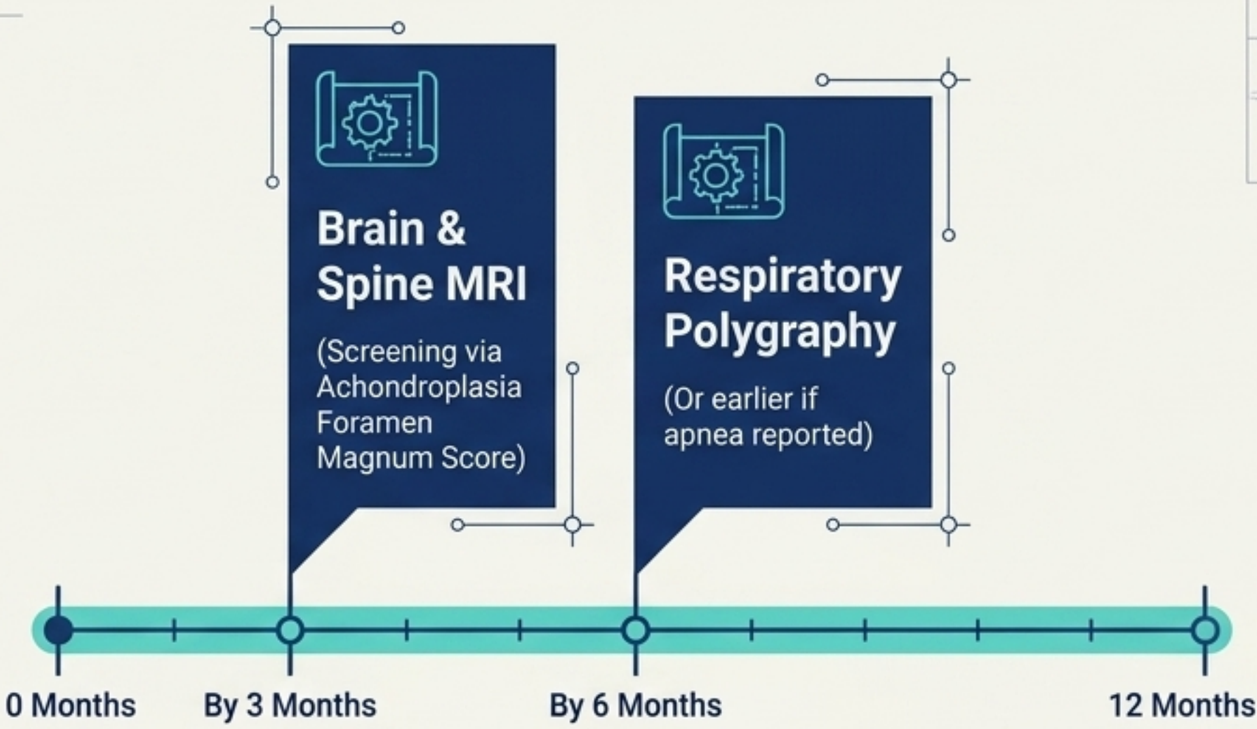
Protecting the Brain & Spine

The first year of life presents acute, potentially life-threatening neurological risks. Monitoring must be intensive, with specialist consultations at least every 6 months.

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The FMS Risk Visualizer



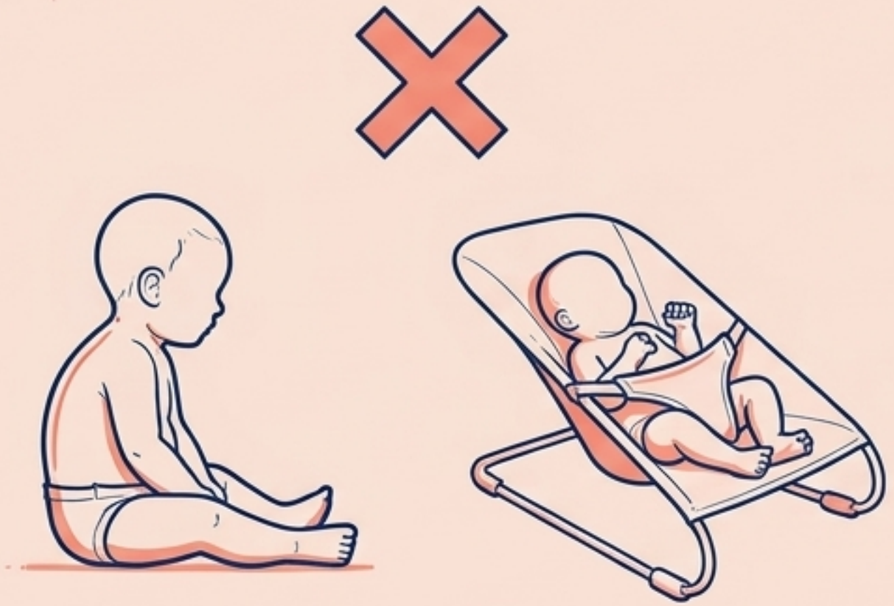


Critical Window Timeline

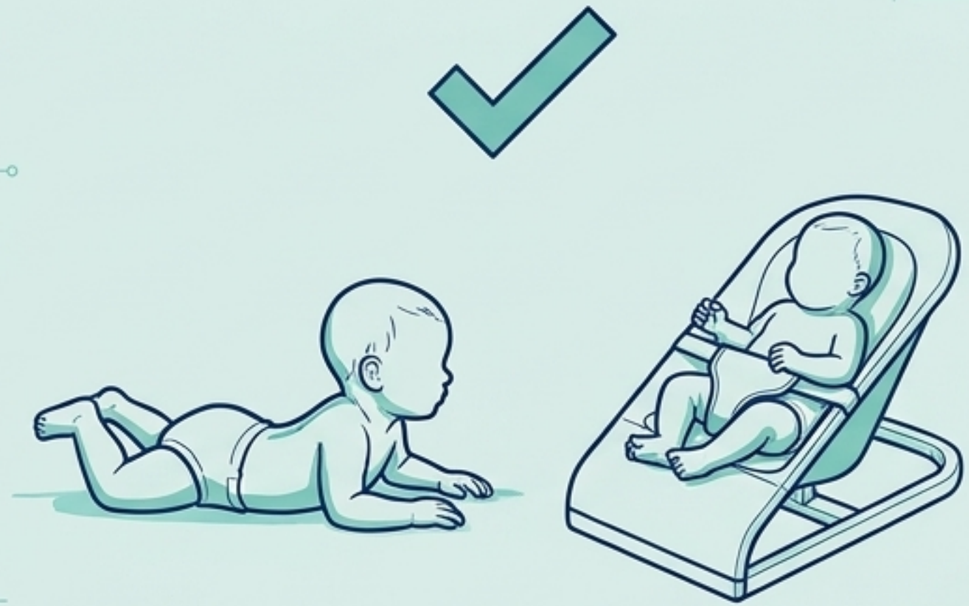
The risk of sudden infant death can reach 7.5% in the first year. Any signs of apnea, hyperreflexia, swallowing difficulty, or rapid head circumference increase require urgent neurosurgical evaluation.

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The Kyphosis Posture Guide



DON'T: Upright sitting before trunk muscular control develops. Avoid slings, baby bouncers, bouncing toys, and trampolines.



DO: Guided 'tummy time', firm truncal support, and supportive seating reclined from the upright position (used only for short periods during feeding).

Insight: Nearly all infants present with a thoracolumbar gibbus. Active postural management minimizes the progression into a fixed deformity that contributes to lifelong morbidity.

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Infancy (<12m) Clinical Dashboard

Growth & Dev

Measure at birth and every 6m. (Must use ACH-specific charts for the ‘red book’).

Neurology

Assess at birth and every 6m.

Spine

Assess at least every 6m.

Hearing / Dental

Newborn hearing test; Audiology evaluation by 1 year. Pediatric dentist review at 1 year.

Psychosocial

Early signposting to ACH support organizations to reduce parental isolation.

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Care Pathway Track

Pre-birth | Infancy | **Preschool** | Primary | Secondary

Preschool (1–4 Years)

Gaining Independence & Function

As neurological risks begin to stabilize, focus shifts to functional independence, sensory input, and preparing the physical environment for exploration.

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The Hearing to Speaking Link

Link 1: Anatomy

Shorter, more horizontal eustachian tubes.

Link 2: Condition

Recurrent Otitis Media with Effusion (OME). Risk is 2x higher in ACH.

Link 3: Outcome

Conductive Hearing Loss.

Link 4: Impact

Speech and Language Delay.

Intervention Alert: Mandatory audiologic evaluations every 6 months. Consider early intervention with hearing aids. Use caution during myringotomy due to potential for a high/exposed jugular bulb.

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Preschool (1-4y) Clinical Dashboard

Respiratory Dial



Sleep studies every 6-12 months. Monitor for obstructive sleep apnea (OSA) signs: neck hyperextension, frequent snoring.

Orthopedic Dial



Review every 6-12 months. Lower limb deformities (tibial bowing) become apparent as mobility increases, affecting up to 50% of children.

Therapy Checklist



- Occupational Therapy (OT) for toilet/home adaptations (foldable bottom-wipers, steps).
- Coordinated nursery/school access arrangements prior to primary school entry.

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Primary School (5–11 Years)

Inclusion & Alignment

Complications often stabilize, allowing review intervals to shift to 12-24 months. Care prioritizes social integration, self-esteem, and targeted surgical interventions before growth windows close.

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Critical Intervention Windows

Age 7: Latest point for formal orthopedic evaluation. (Require full-length low-radiation EOS imaging)

Window Closes: Simple surgical interventions (e.g., guided growth for limb alignment) become ineffective past this point.

Window 1: Orthopedic Alignment

Age 5 | Age 6 | **Age 7** | Age 8 | Age 9 | Age 10 | Age 11 | Age 12

Age 10: Formal assessment

Window 2: Orthodontic Correction

Age 5 | Age 6 | Age 7 | Age 8 | Age 9 | **Age 10** | Age 11 | Age 12

Assess for maxillary hypoplasia, crossbites, and crowding. Interceptive management for maxillary arch development begins here.

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Primary School (5-11y) Clinical Dashboard

Pre-birth | Infancy | Preschool | **Primary** | Secondary



Psychology

From age 7, cognitive development allows complex emotional awareness of ACH as a lifelong condition. Consider psychology referral for self-esteem and peer dynamics.



Spine

Monitor for persistent moderate to severe kyphosis; refer to spinal surgeon if unresolved.



Lifestyle

Reinforce healthy weight maintenance to reduce long-term joint and back strain.



School Support

Environmental adjustments (height-appropriate seating) and Individual Education Plans (IEPs).

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Pre-birth | Infancy | Preschool | Primary | **Secondary**

Secondary School (11–18 Years)

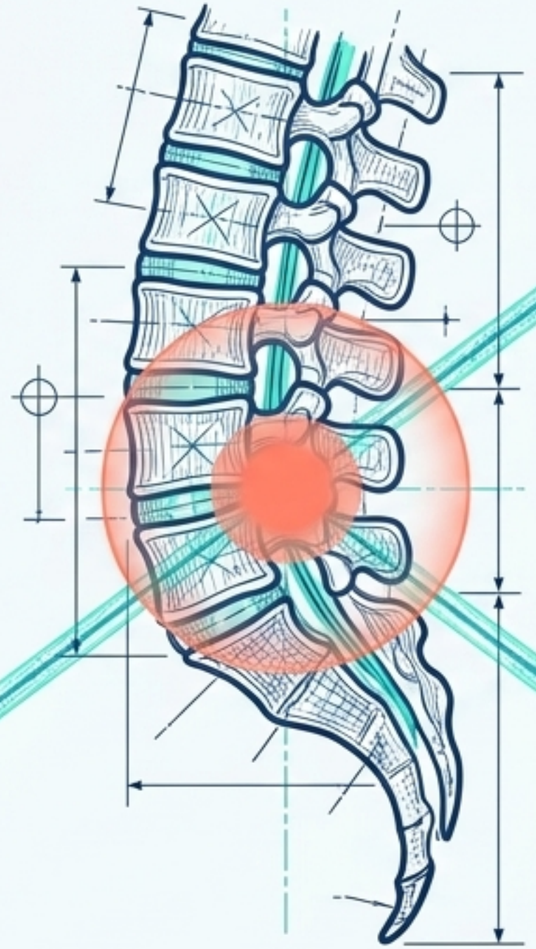
Autonomy & Transition

The transition from pediatric to adult care. The focus shifts to self-advocacy, managing late-physical complications, and vocational planning.

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Adolescent Complications Radar

Pre-birth | Infancy | Preschool | Primary | **Secondary**



Spinal Stenosis

Incidence increases through adolescence. Watch for claudication pain on mobilization (often relieved by squatting) or new loss of bowel/bladder control. Requires urgent neuro-eval/MRI.

Pain & Fatigue

Implement individual pacing, analgesic plans, and mobility aids via specialist physio.

Genetics

Offer genetic counseling from age 13-14 regarding inheritance and reproductive choices.

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Secondary School (11-18y) Clinical Dashboard

Pre-birth | Infancy | Preschool | Primary | **Secondary**



Independence Protocol

Driving vehicle adaptations, adapted hygiene products for menstruation, vocational pathing.



Obesity Management

Use ACH-specific weight charts. Obesity deeply exacerbates joint pain and apnea.

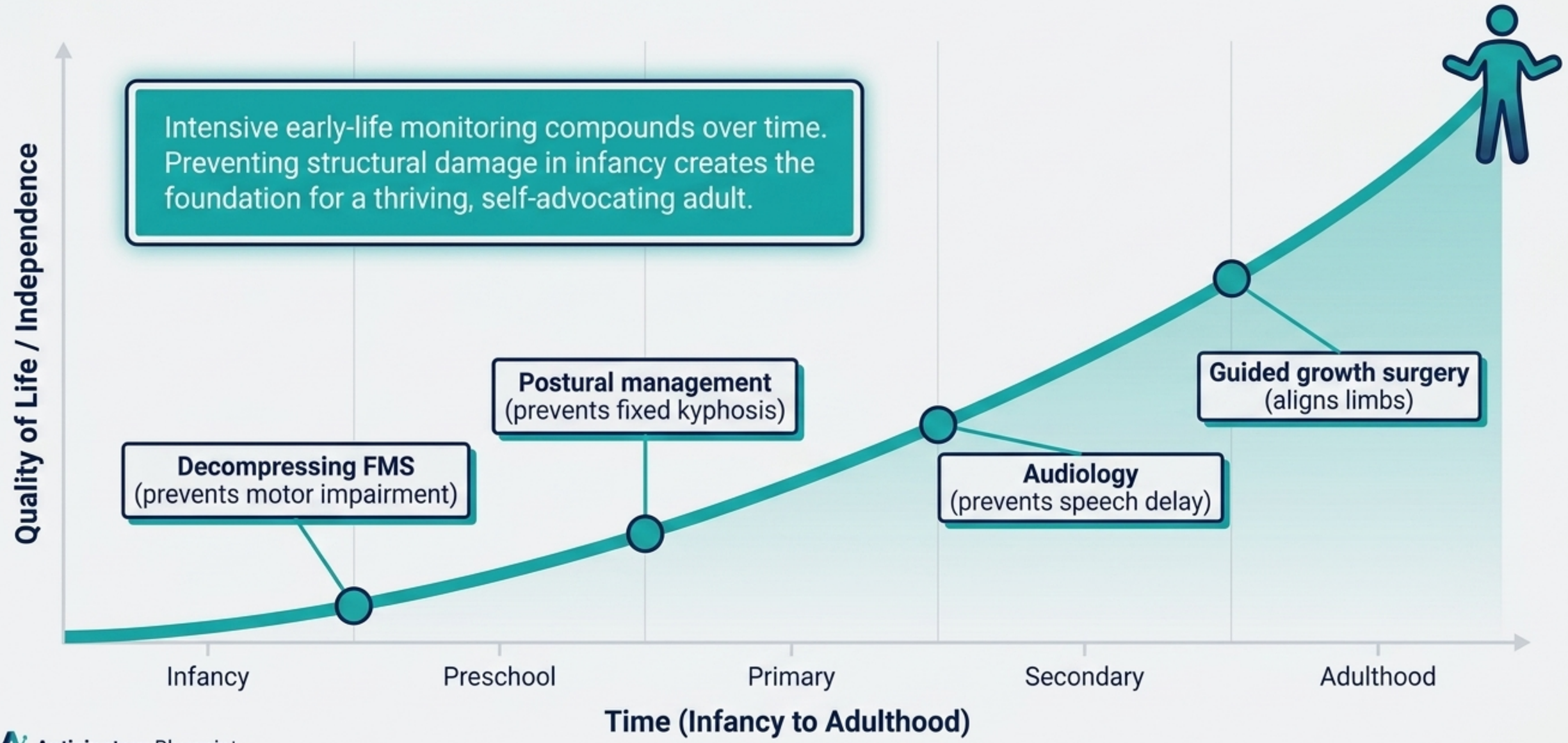


Transition Protocol

Handing over autonomy to the young person. By age 18, transfer to an adult center of expertise. Ensure they know key symptoms to report to general practitioners.

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The Lifelong Impact of Anticipatory Care



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The Horizon



Emerging Tech

Introduction of C-type natriuretic peptide (CNP) analogues (e.g., vosoritide) and FGFR3 inhibitors aiming to increase stature and reduce complications.



System Need

Prescription must remain within specialist multidisciplinary ACH centers. A nationally agreed 'minimal dataset' is required to monitor real-world safety and efficacy of these new technologies in the UK.

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