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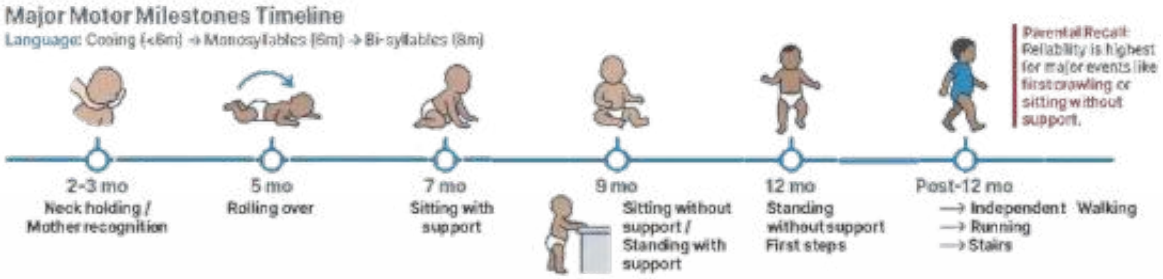
Developmental Milestones & The 5 Domains

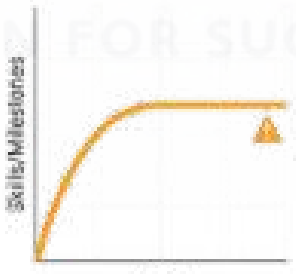


The 5 Clinical Domains

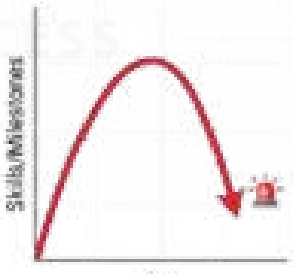
Defining Delay, Stagnation & Regression

Definitions

- **Milestone** missed by 0.5-1 month.
- **Significant Delay:** Milestonemissedby ≥3 months.
- **Global Developmental Delay (GDD):** Significant delay in ≥2 domains (e.g, Motor + Cognitive).



Developmental Stagnation (Plateau) - Red Line.



Developmental Regression (Loss of skills) - **EMERGENCY**

Safety Note: Regression is absolutely devastating and requires emergent evaluation.

Clinical Scenarios: Isolated Motor & Language Delays

Isolated Gross Motor Delay

- **Context:** Finemotor,language, social, cognitive are normal.

Suspected Causes:

- Vitamin D deficiency (Primary concern)
- Bony lesions /Musculoskeletal problems
- Spasticity (e.g., Leukodystrophy)

Isolated Fine Motor Delay

- **Context:** Grossmotor normal (e.g., standing); unable to grasp.

Suspected Causes:

- Extremely rare
- Early onset neuropathies (e.g., rare CMT)

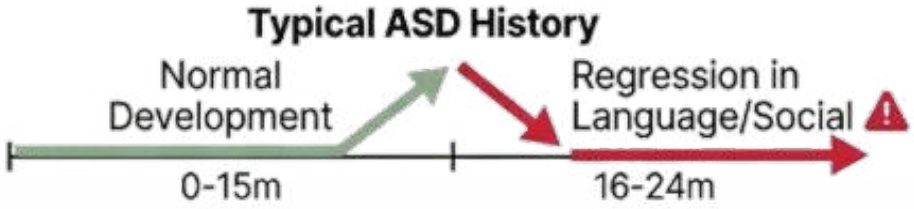
Isolated Language Delay

- **Context:** Nomonosyllables; other domains normal.

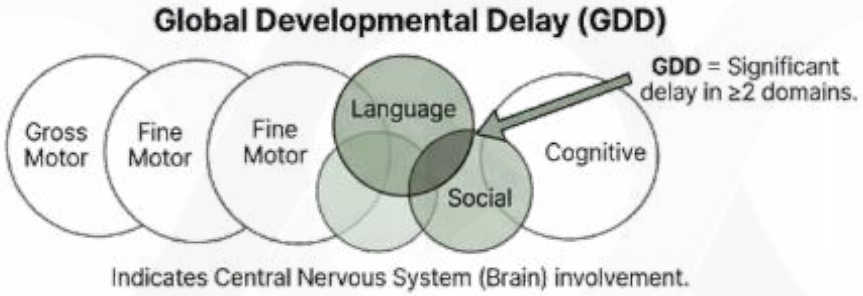
Suspected Causes:

Clinical Scenarios: Social, Cognitive & Global Delay

Autism Spectrum Disorder (ASD) & Cognitive



- **Isolated Cognitive Delay:** Motor/Language normal.
- **Associations:** Learningdisabilities (Dyslexia, mild ID).

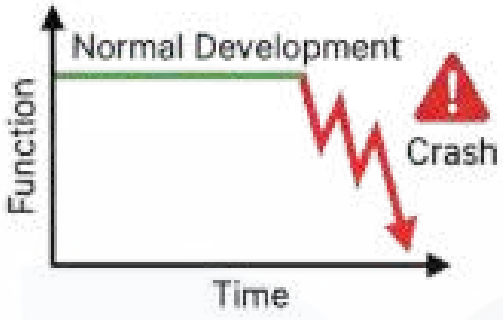
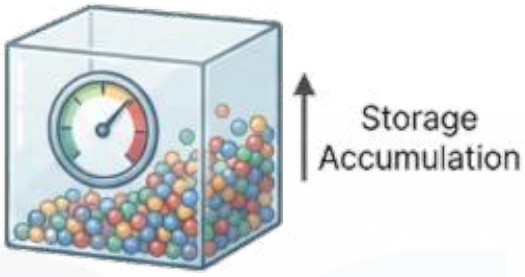


Etiology: Perinatal & Genetic Causes

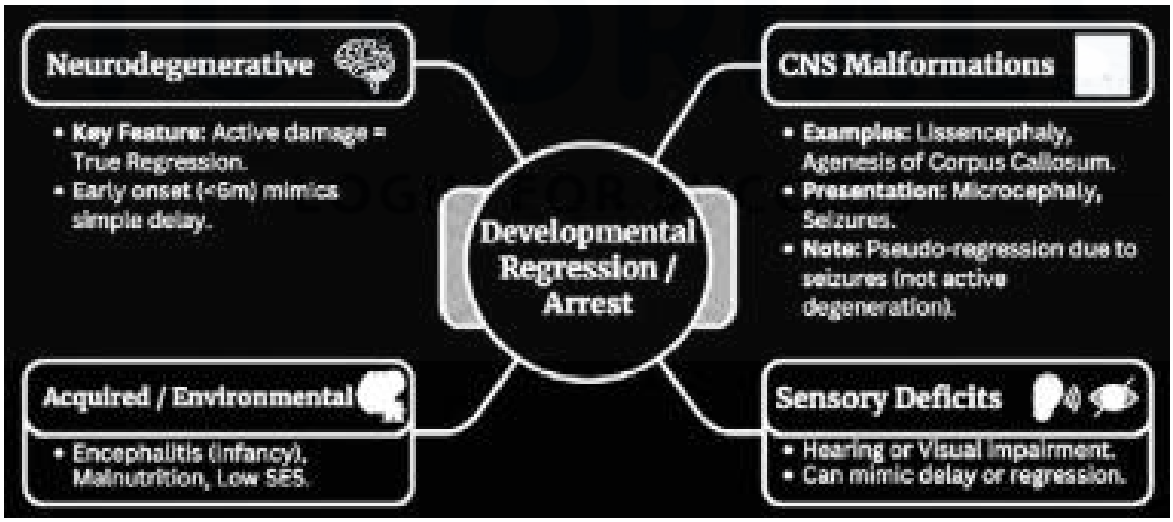
- **History:** Asphyxia, NICU stay, Ventilation, Jaundice, Seizures.
- **Antenatal:** Infections (TORCH, CMV) → Microcephaly.
- **Diagnosis:** MRI confirms injury (e.g., cerebral palsy context).

- **Types:** Syndromic, Chromosomal, Single gene. **Examples:** Down's, Fragile X, Angelman.
- **Clinical Clues:** Dysmorphic features, Hypodontia, Multi-system.
- **Evaluation:** Karyotype, Microarray, Gene panels.

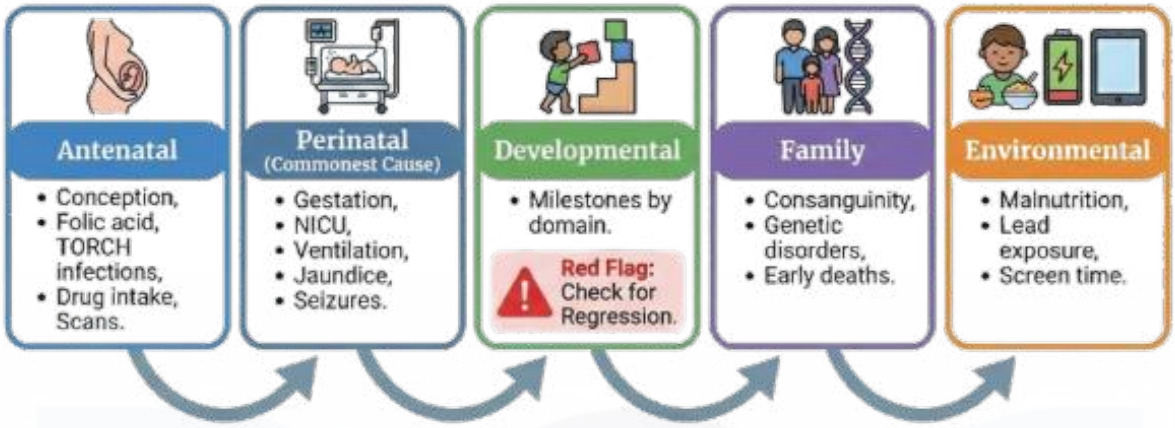
Etiology: Metabolic Disorders (Inborn Errors of Metabolism)

Small Molecule Disorders	Large Molecule Disorders
	
• **Onset**: Early (90% <1 year; usually 1-2 years). • **Presentation:** Acute Decompensation, Regression, Fever with Encephalopathy. • **Clues:** Abnormal Ammonia, Lactate, Ketones.	• **Onset:** Can be late. • **Presentation:** Chronic baseline delay; can have regression. • **Clues:** Dysmorphic features (coarse facies), Hepatomegaly.

Neurodegenerative, Malformations & Acquired Etiologies



Evaluation: Detailed History Framework



Evaluation: Physical Examination & Tone Assessment

General Assessment

- **Head:** Micro/Macrocephaly.
- **Face:** Dysmorphism (Coarse facies).
- **Eyes:** Cataracts, Cherry red spot.
- **Skin:** Café-au-lait spots.
- **Hair:** Lusterless, Flag sign (Nutrition).

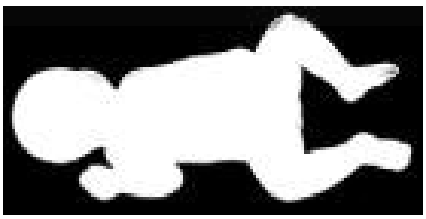
Tone Assessment Matrix	
Central Hypotonia	**Peripheral Hypotonia**
Floppy Strong	Floppy Weak
Normal Reflexes	Decreased Reflexes (e.g., SMA)

Infant Maneuvers

Horizontal Suspension

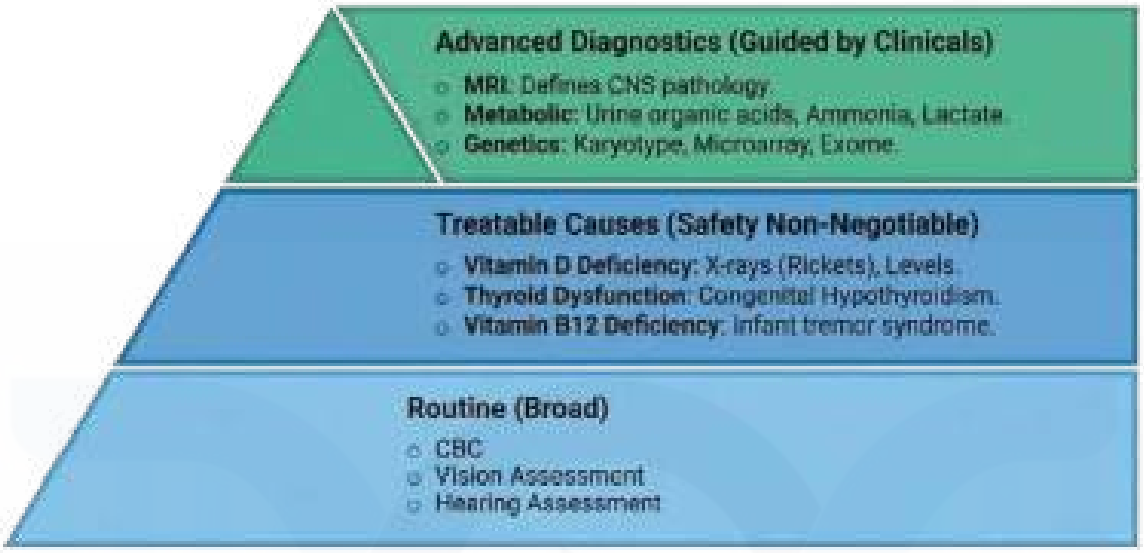
Pull to Sit (Head lag)

Frog Leg Posture

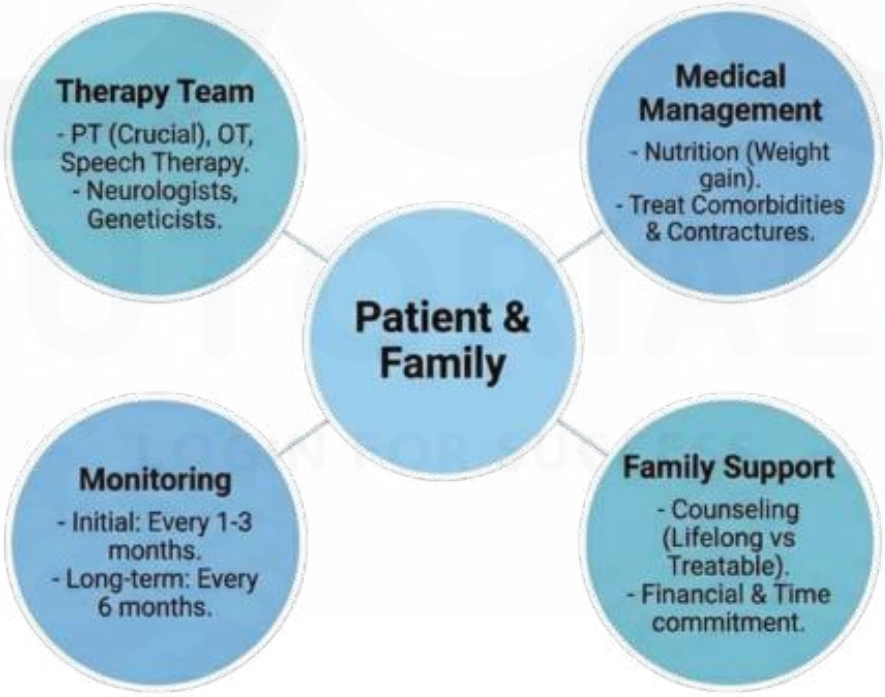


Frog Leg Posture (Sign of severe hypotonia/SMA)

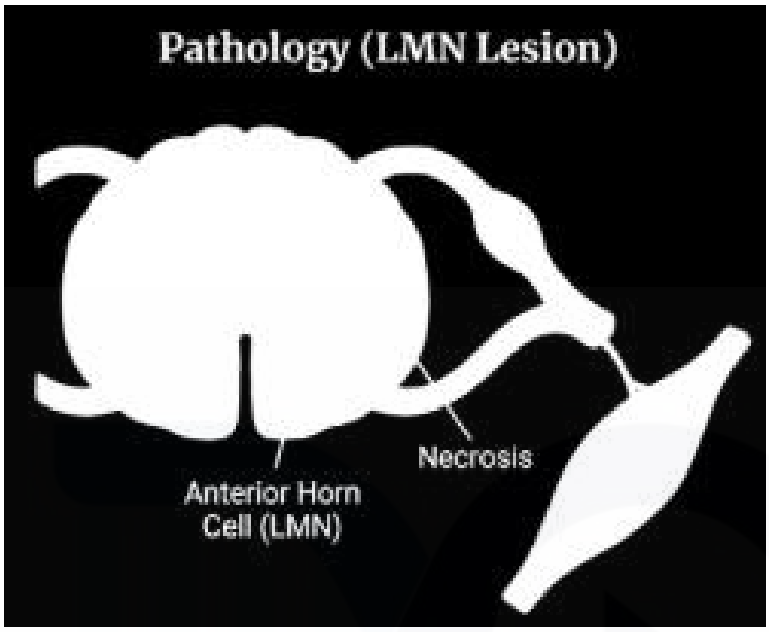
Investigations: Baseline & Advanced



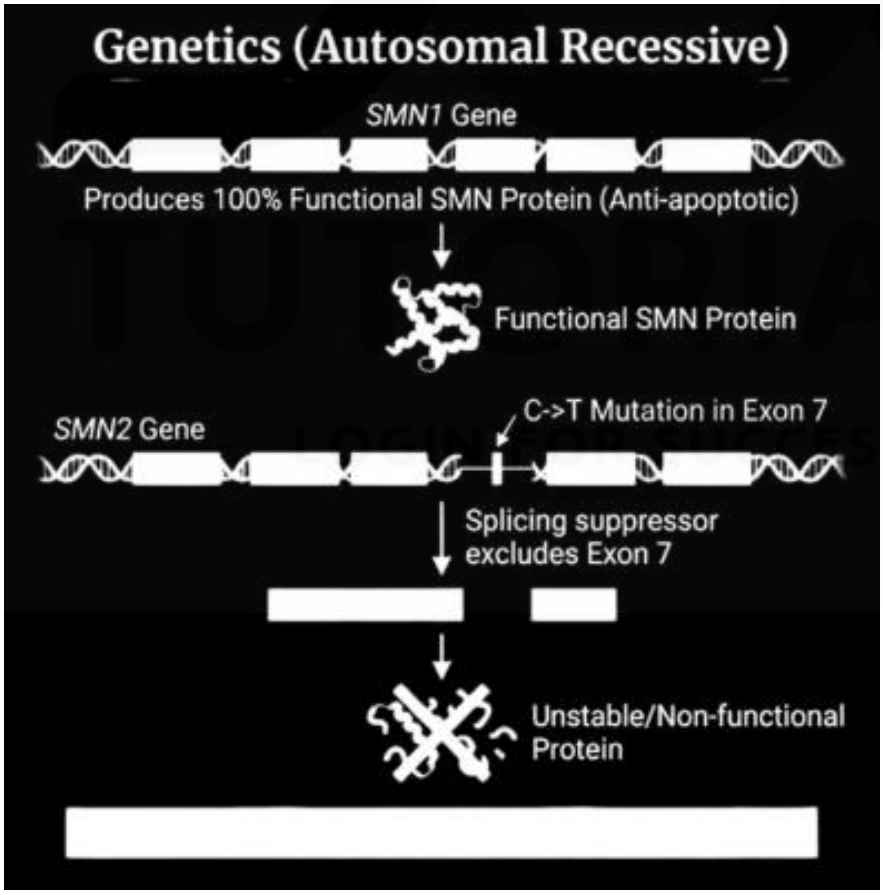
Management of Developmental Delay



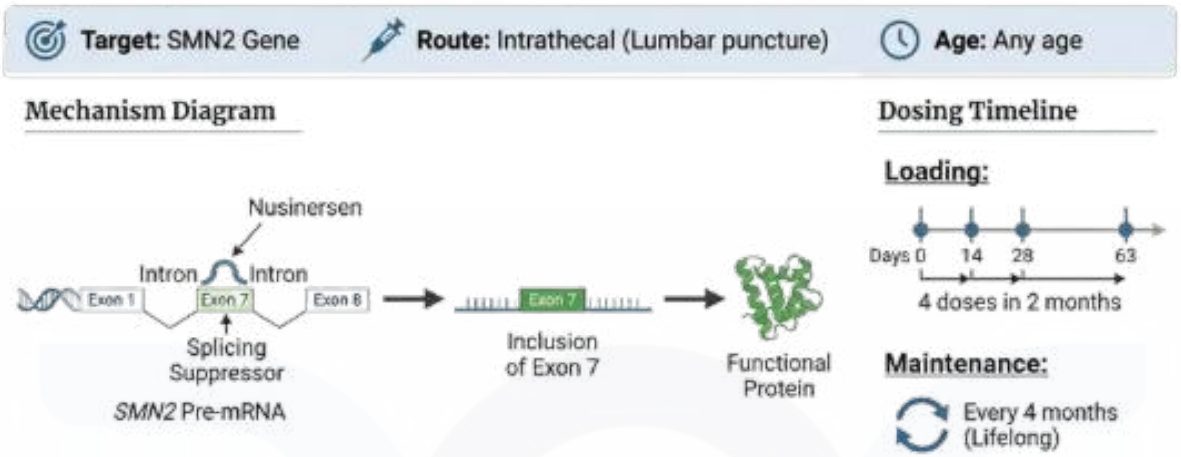
Spinal Muscular Atrophy (SMA): Pathology & Genetics



Progressive apoptosis of Anterior Horn Cells Flaccid paralysis, Loss of reflexes.



SMA Therapy: Nusinersen (Spinraza)



SMA Therapy: Onasemnogene abeparvovec (Zolgensma)

SMA Therapy: Risdiplam (Evrysdi) & Summary

Risdiplam Profile

- **Mechanism:** SMN2 Splicing Modifier (Promotes Exon 7 inclusion).
- **Route:** Oral (Daily/Lifelong).
- **Age:** ≥2 months (No upper limit).
- Very well tolerated.

Comparison Table

	Nusinersen	Zolgensma	Risdiplam
Target	SMN2	SMN1	SMN2
Mechanism	Blocks Suppressor	Gene Replacement	Splicing Modifier
Route	Intrathecal	IV (Single Dose)	Oral (Daily)

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