



PG Residency

Pediatrics

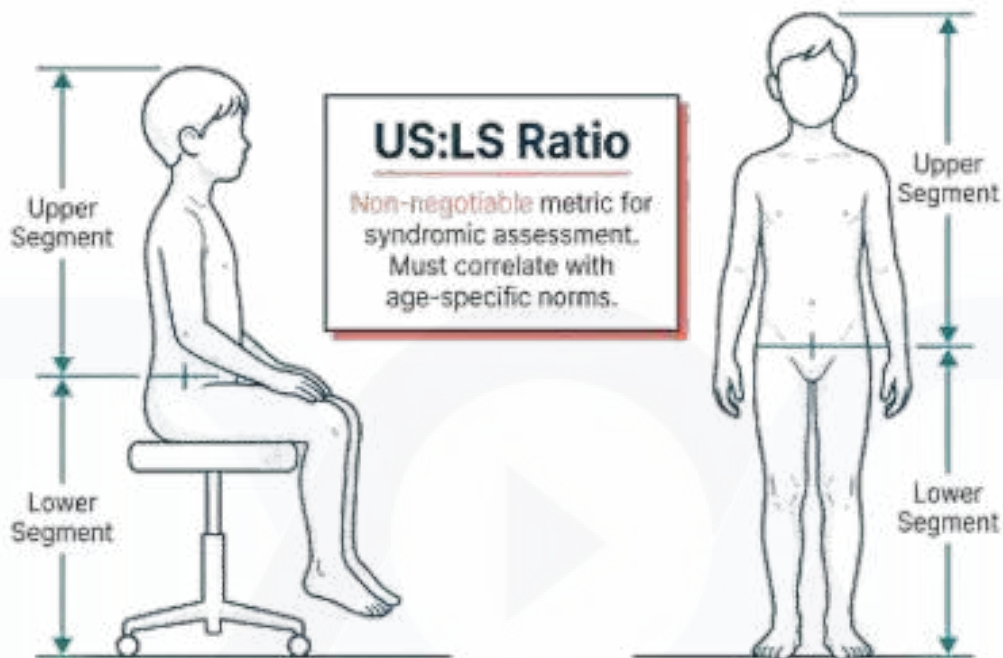
Genetics & Syndrome

Smart Notes. Better Scores.



General Assessment & Anthropometry

The Foundation of Dysmorphic Examination



Clinical Checklist

1. Consciousness & Orientation

- Assess: Conscious? Cooperative? Alert?
- Verify orientation to time, place, and person.

2. Vital Signs

- Correlate abnormalities directly with clinical condition.

3. Anthropometric Assessment

- Malnutrition Staging: Acute, Chronic, or Acute-on-chronic.
- US:LS Ratio Rule: Essential for all dysmorphic children. Document even if normal.

4. General Signs (Rapid Screen)

- Pallor, Icterus, Cyanosis, Clubbing, Lymphadenopathy, Edema, Raised JVP.



DocTutorials

High-Yield Videos

Structured Syllabus Videos



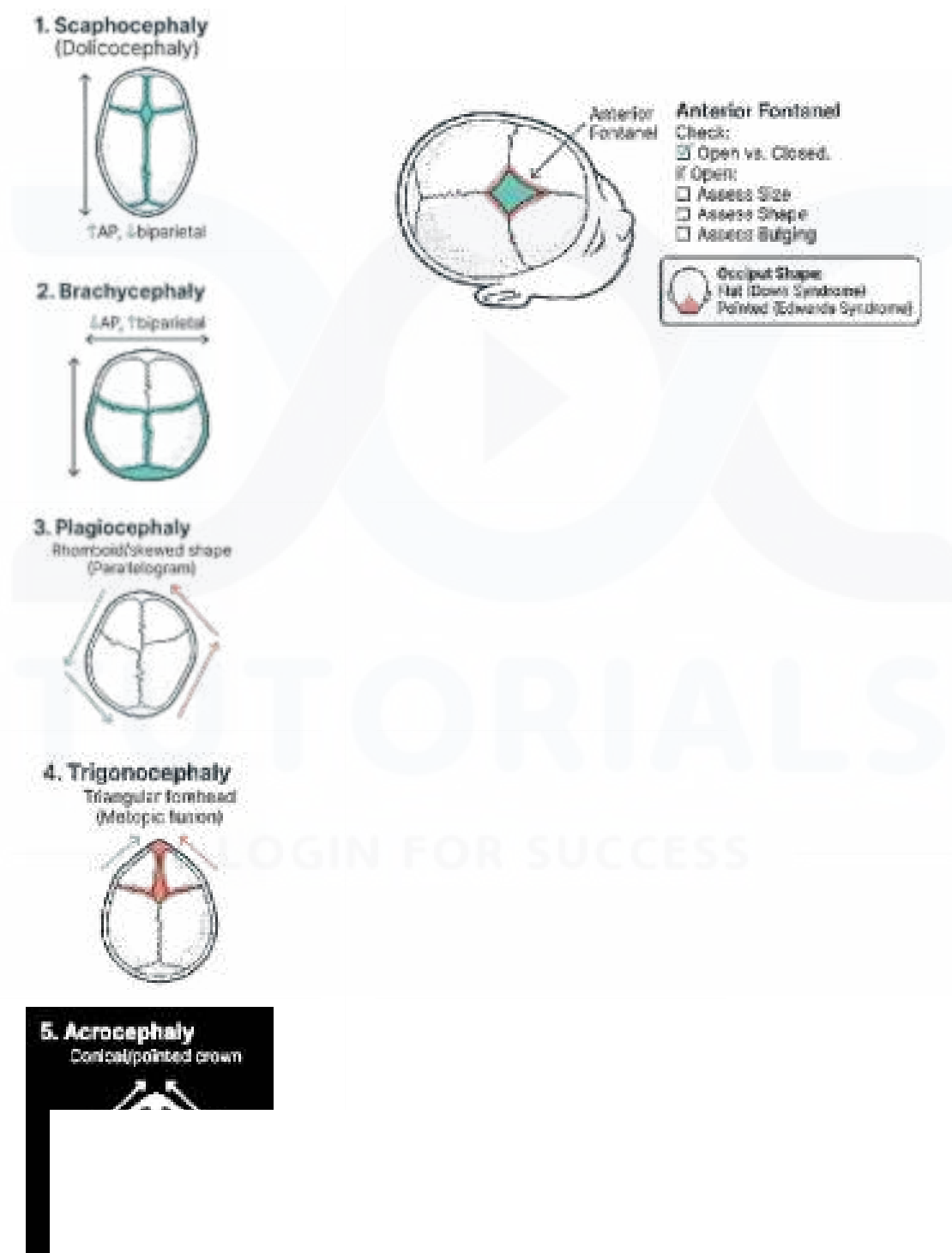
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Head & Skull Examination

Differentiating Normal Variants from Craniosynostosis

Occipitofrontal Circumference (OFC): Measure and plot on growth chart. Interpret as Normal, Microcephaly, or Macrocephaly.

SKULL SHAPE VARIANTS



Facial Features & Eye Assessment

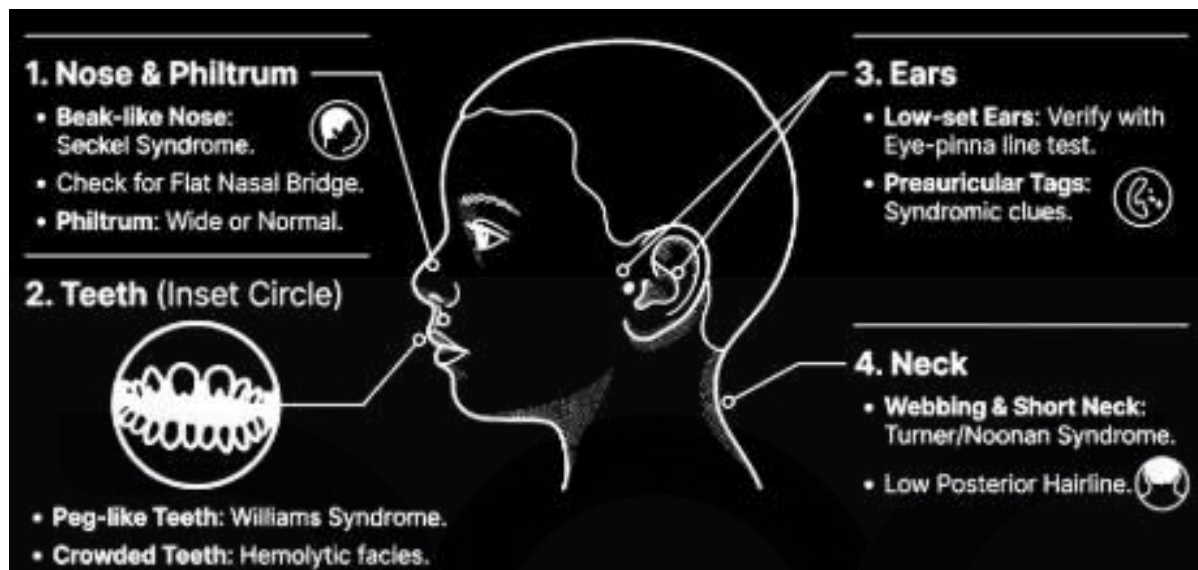
Eye Slant & Assessment



Common Facies & Associated Syndromes

FACIES TYPE	KEY FEATURES	ASSOCIATED CONDITIONS
 Coarse Facies	Thick lips, flat bridge, large tongue	MPS, Cretinism
 Elfin Facies	Wide mouth, smooth philtrum, pointed teeth	Williams Syndrome
 Hemolytic Facies	Frontal bossing, crowded teeth	Thalassemia
 Other Facies	Moon, Hepatic, Mask-like	Cushing's, CLD, Moebius

Midline Structures: Nose, Mouth, Ears, Neck



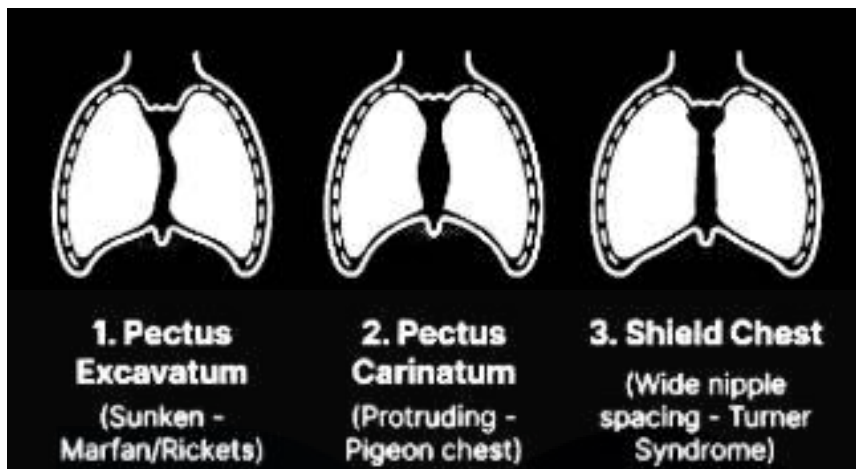
Extremities: Hands, Feet & Chest

The Syndromic Hand (Down Syndrome Focus)



- Simian crease
- Clinodactyly
- Short stubby fingers

Chest Morphology



Note: Wide carrying angle (Cubitus valgus).

Foot & Digit Anomalies

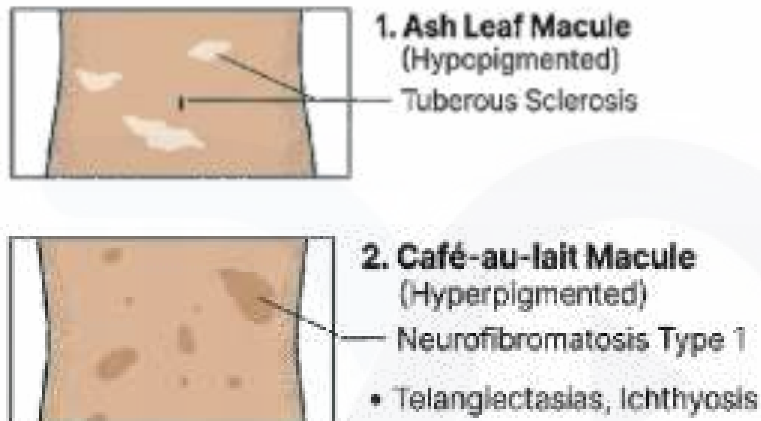


- Polydactyly
- Syndactyly (Craniosynostosis)
- Triphalangeal thumb (Holt-Oram)
- Broad thumb/toes (Rubinstein-Taybi)

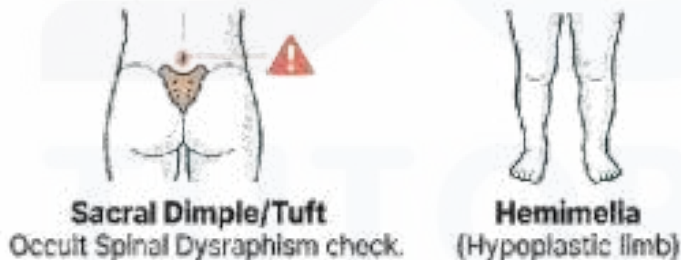
Extremities: Hands, Feet & Chest

The Hidden Signs

Dermatological Markers



Spine & Limbs



Genital & Systemic

Male Checklist

- Testicular descent
- Micropenis
- Hypospadias

Female Checklist

- Clitoromegaly (Virilization/CAH)

System

- (
- (



Pediatrics

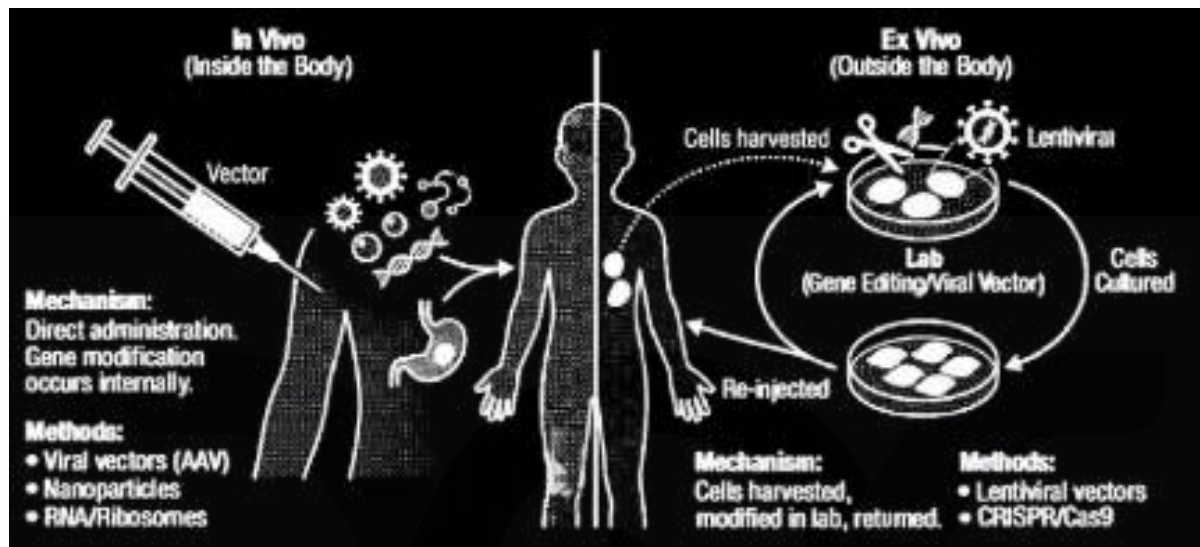
Clinical Case

Structured Clinical Case Videos



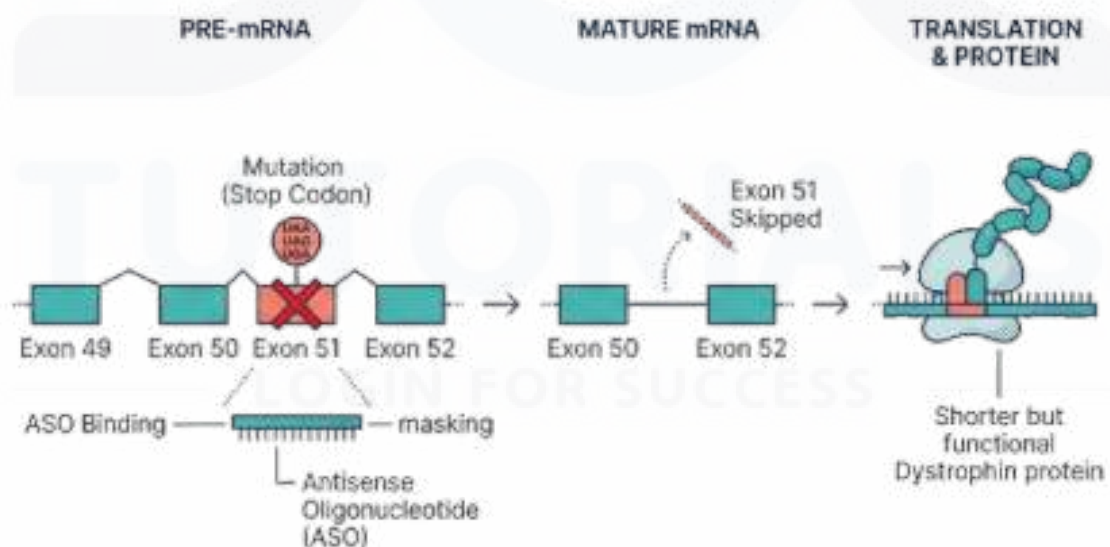
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Gene Therapy Strategies: The Framework



In Vivo: Antisense Oligonucleotides (ASO)

Mechanism of Exon Skipping in Duchenne Muscular Dystrophy



Key Concepts & Therapeutics

Duchenne Muscular Dystrophy (DMD): X-linked recessive, deficient dystrophin.

- **Eteplirsen** (Exondys 51): Intrathecal. Skips Exon 51.
- **Golodirsen**: Subcutaneous. Skips Exon 45.
- **Viltolarsen**: Subcutaneous. Skips Exon 53.

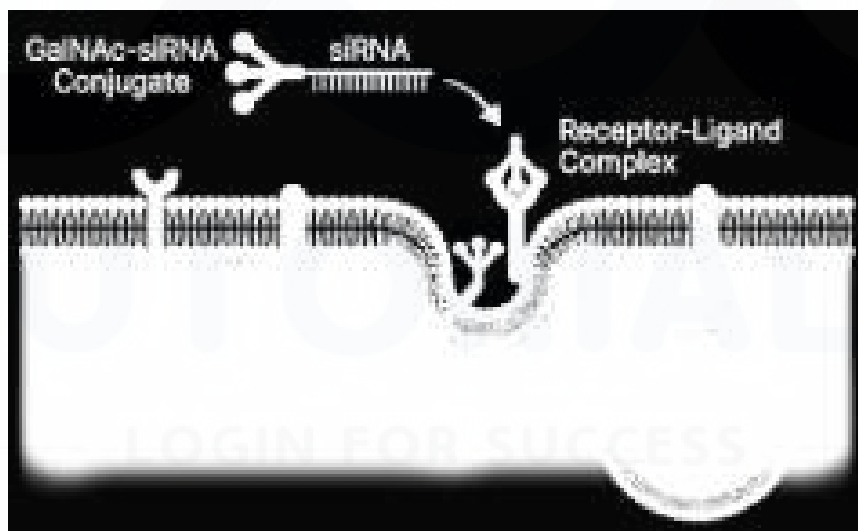
In Vivo: ASO Therapeutics Overview

Disease	Drug Name		Gene Target	Route
Spinal Muscular Atrophy (SMA)	Nusinersen (Spinraza)	Activates inactive SMN2 to compensate for missing SMN1.	SMN2 Gene	Intrathecal
TTR-Amyloidosis •	Inotersen		Transthyretin (TTR)	Subcutaneous
Hyperlipidaemia •	Mipomersen		Apolipoprotein B100	Subcutaneous
Familial Chylomicronaemia •	Volanesorsen		Apolipoprotein C3	Subcutaneous

In Vivo: Delivery Systems

GalNAc Conjugates vs. Lipid Nanoparticles

GalNAc Conjugates

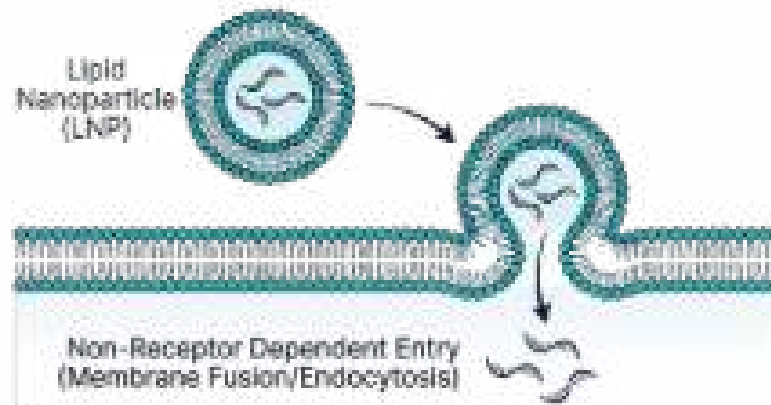


Receptor-Dependent (Liver Specific)

- Givosiran (Porphyria)
- Inclisiran (Cholesterol)
- Lumasiran (Hyperoxaluria)
- Fitusiran (Hemophilia)

Route: All Subcutaneous.

Lipid Nanoparticles (LNP)



Non-Receptor Dependent.

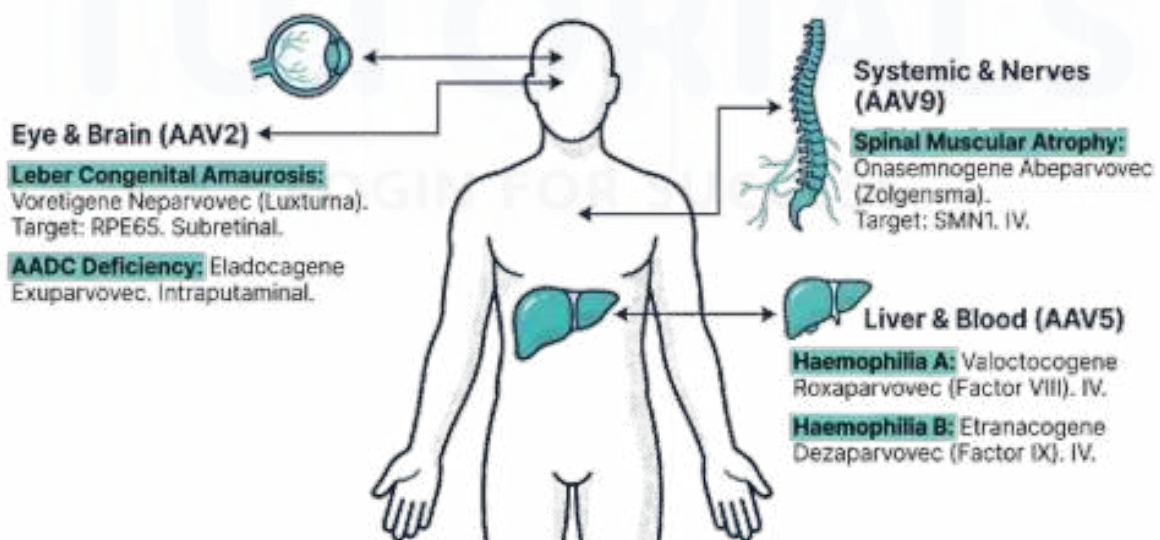
Approved Drugs:

- Patisiran (TTR Amyloidosis)

Route: IV Route.

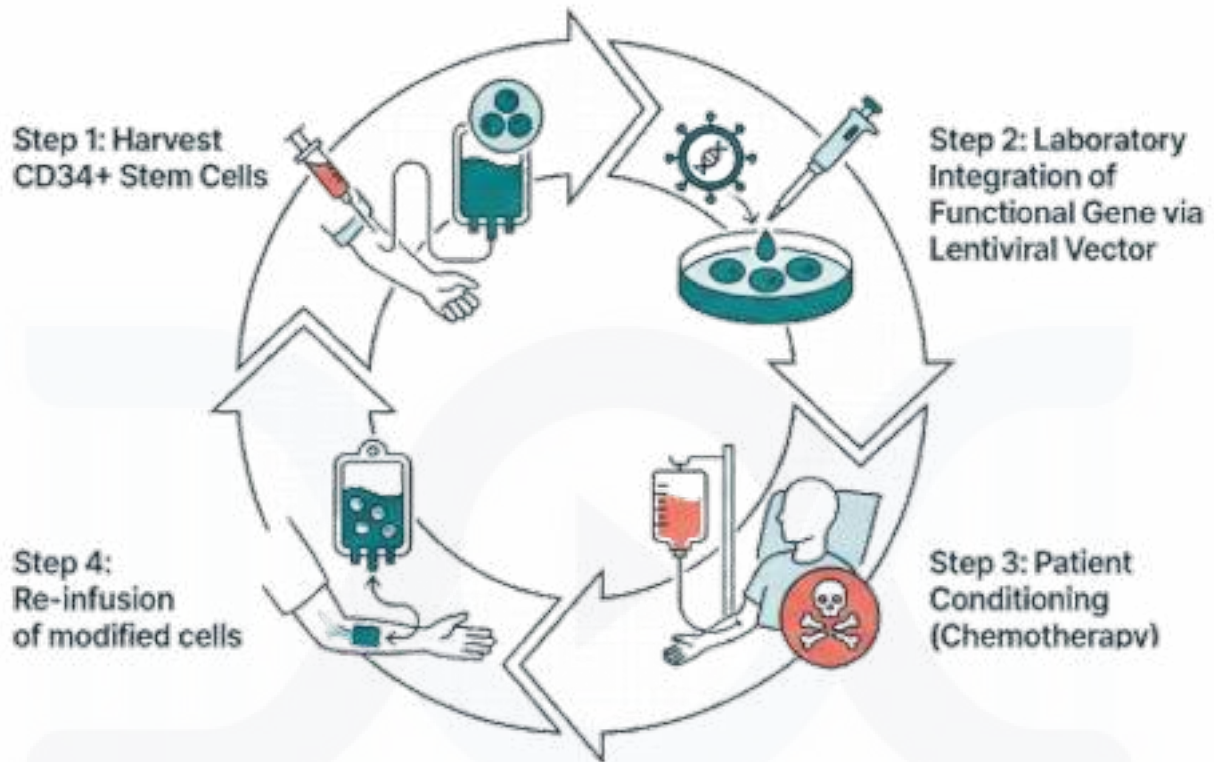
In Vivo: Adeno-Associated Virus (AAV) Vectors

The 'One-Shot' Gene Replacement Strategy



Ex Vivo: Lentiviral Gene Addition

Hematopoietic Stem Cell Modification



Key Therapies

1. Beta-Thalassaemia Major

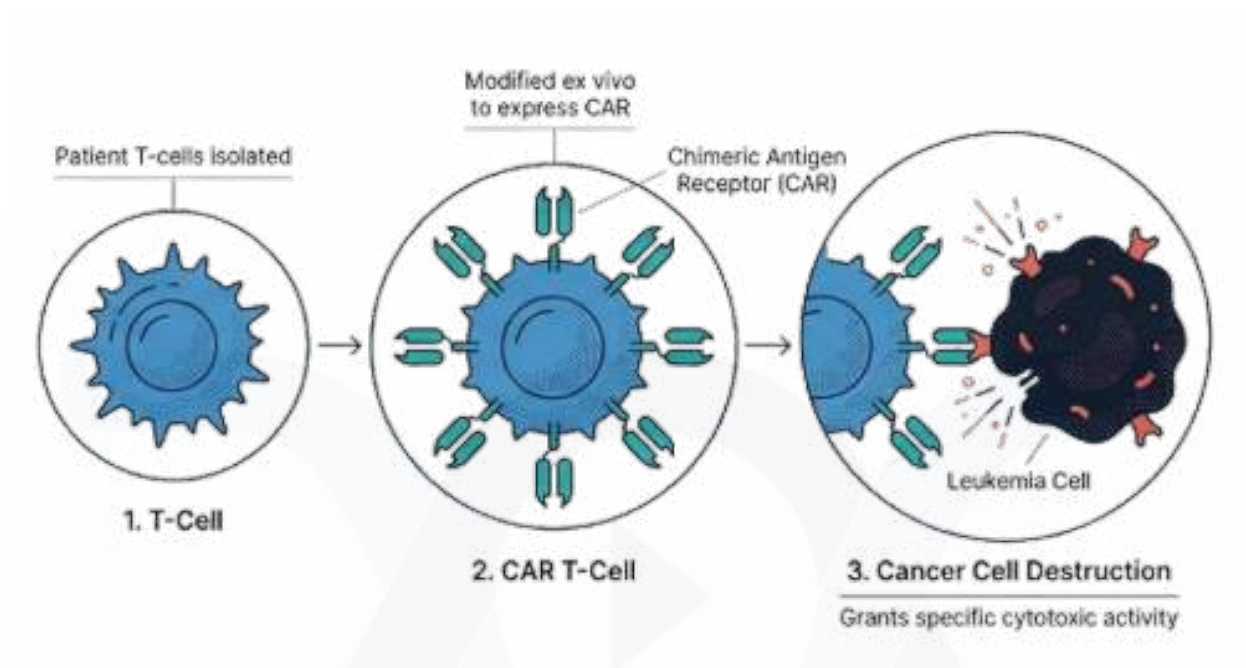
- Betibeglogene Autotemcel (Zynteglo)
- Target: Adds functional beta-globin gene.
- Outcome: 90% transfusion independence.

2. Sickle Cell Anaemia

- Lentiglobine Autotemcel (Lyfgenia)
- Target: Adds functional beta gene -> HbA production.
- Outcome: Reduced vaso-occlusive crises.

Ex Vivo: CAR T-Cell Therapy

Immunotherapy for Acute Lymphoblastic Leukaemia (ALL)



Mechanism:

- Patient T-cells isolated ☐ Modified ex vivo to express CAR ☐ Expanded ☐ Re-infused.

Action:

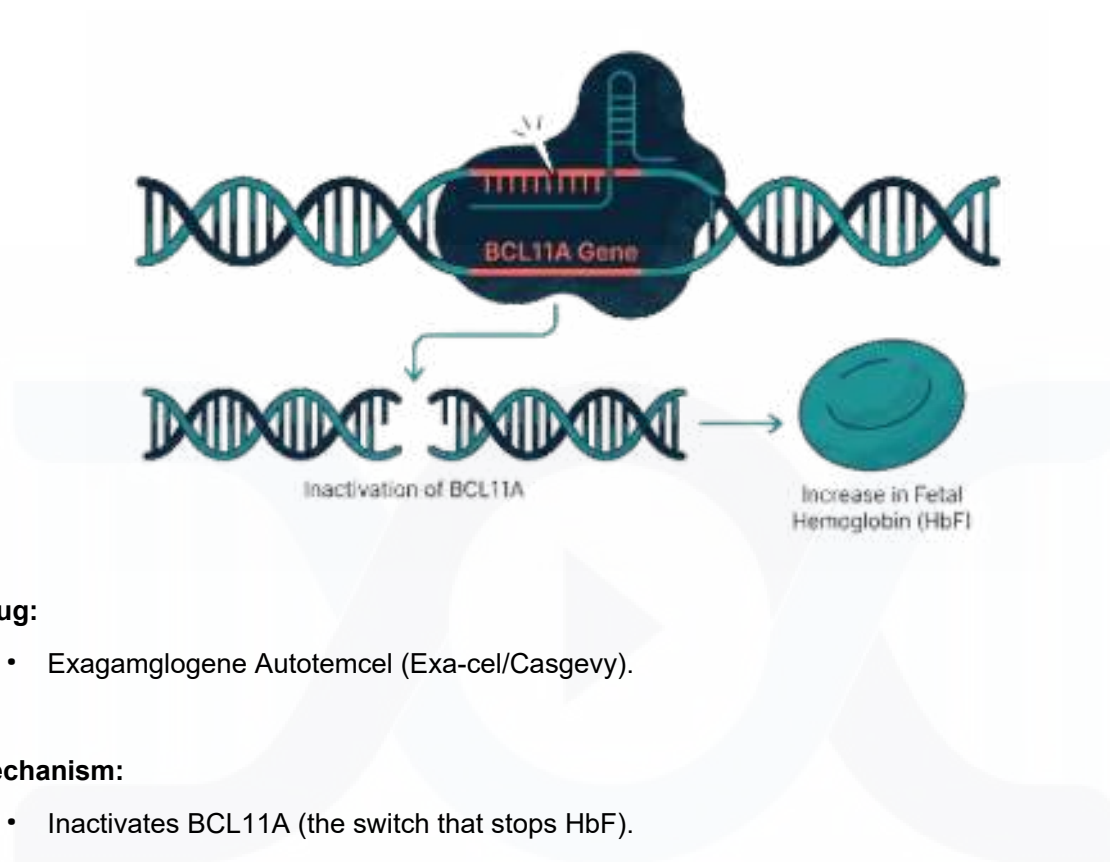
- Grants specific cytotoxic activity against target cancer cells.

Primary Pediatric Use:

- Acute Lymphoblastic Leukaemia (ALL).

Ex Vivo: CRISPR/Cas9 Gene Editing

Molecular Scissors for Sickle Cell Anaemia



Drug:

- Exagamglogene Autotemcel (Exa-cel/Casgevy).

Mechanism:

- Inactivates BCL11A (the switch that stops HbF).

Result:

- Increased HbF prevents sickling and crises.

Safety Note:

- Monitor for **off-target effects/malignant transformation** (permanent genetic change vs. Hydroxyurea).

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