ΣΥΓΓΕΝΕΙΣ ΑΝΩΜΑΛΙΕΣ

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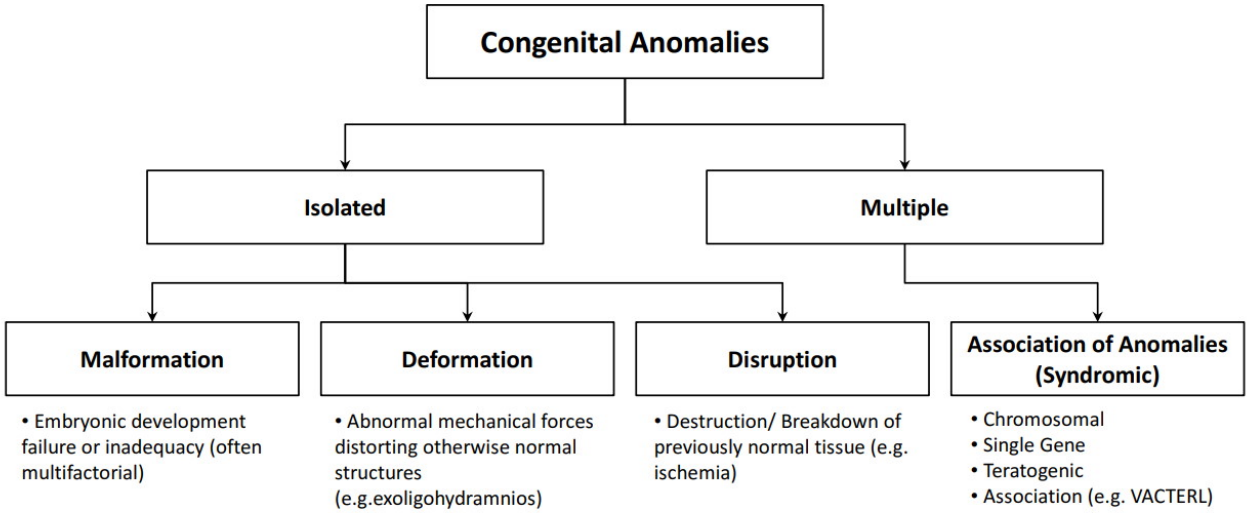
ΜΟΡΙΑΚΟΣ ΒΙΟΛΟΓΟΣ-ΓΕΝΕΤΙΣΤΡΙΑ

Ιστολογία-Εμβρυολογία Ι

Εισαγωγικά…

Congenital Anomalies



Pediatric

313

Things to Consider:
History – Prenatal: maternal health, exposures, screening, ultrasounds; delivery; neonatal
Family History – Three Generations: prior malformations, stillbirths, recurrent miscarriages, consanguinity
Physical Exam – Variants, minor anomalies, major malformation
Diagnostic Procedures – Chromosomes, molecular/DNA, radiology, photography, metabolic
Diagnostic Evaluations – Prognosis, recurrence, prenatal diagnosis, surveillance, treatment

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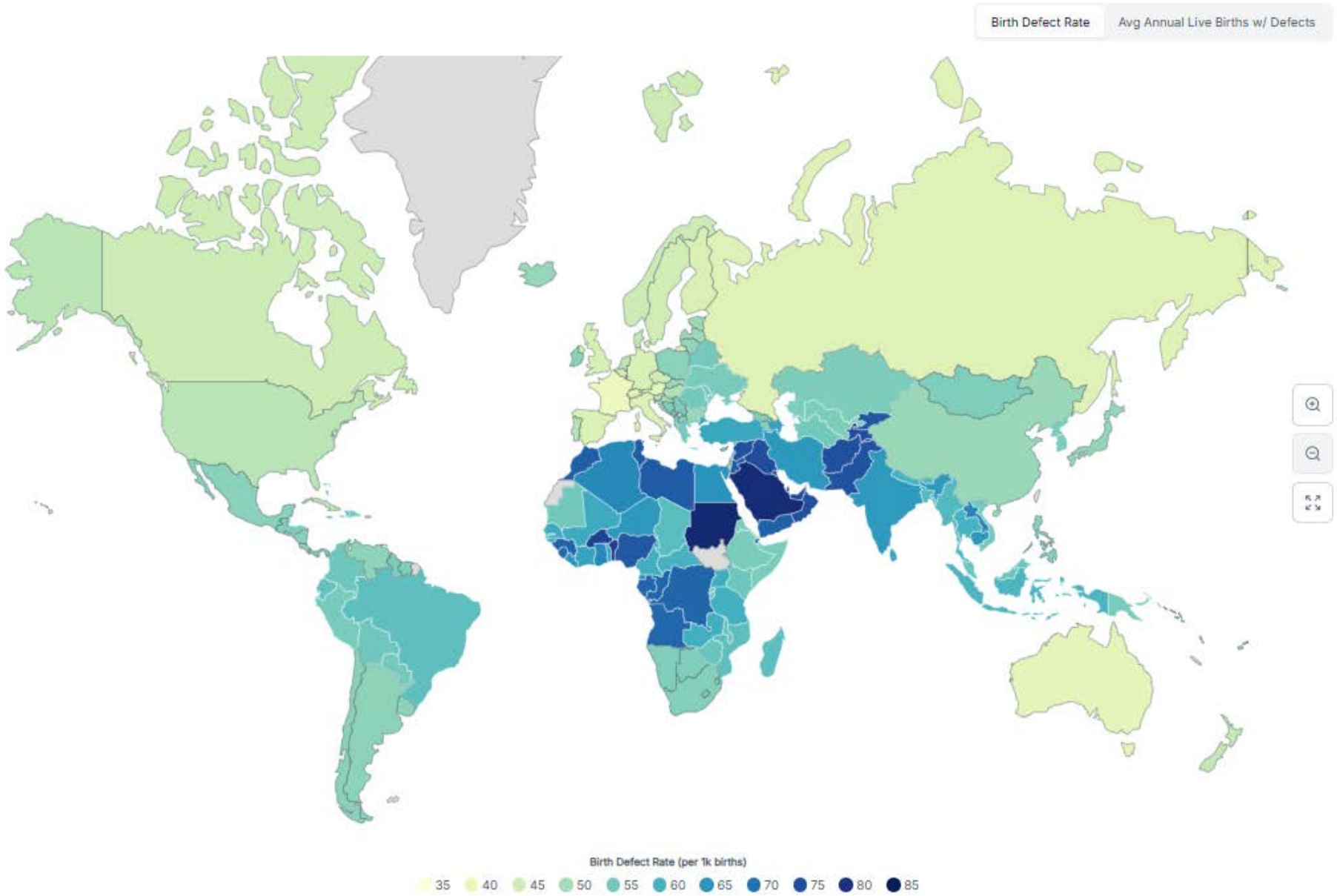
Congenital disorders

27 February 2023

Key facts

- An estimated 240 000 newborns die worldwide within 28 days of birth every year due to congenital disorders. Congenital disorders cause a further 170 000 deaths of children between the ages of 1 month and 5 years.
- Congenital disorders can contribute to long-term disability, which takes a significant toll on individuals, families, health care systems and societies.
- Nine of ten children born with a serious congenital disorder are in low- and middle-income countries.
- As neonatal and under-5 mortality rates decline, congenital disorders become a larger proportion of the cause of neonatal and under-5 deaths.
- The most common severe congenital disorders are heart defects, neural tube defects and Down syndrome.
- Although congenital disorders may be the result of one or more genetic, infectious, nutritional or environmental factors, it is often difficult to identify the exact causes.
- Some congenital disorders can be prevented. Vaccination, adequate intake of folic acid or iodine through fortification of staple foods or supplementation, and adequate care before and during a pregnancy are examples of prevention methods.

Birth Defect Rates by Country 2025



Sudan	Saudi Arabia	Benin	Burkina Faso	Palestine
82	81.3	77.9	77	76.6

Εισαγωγικά…

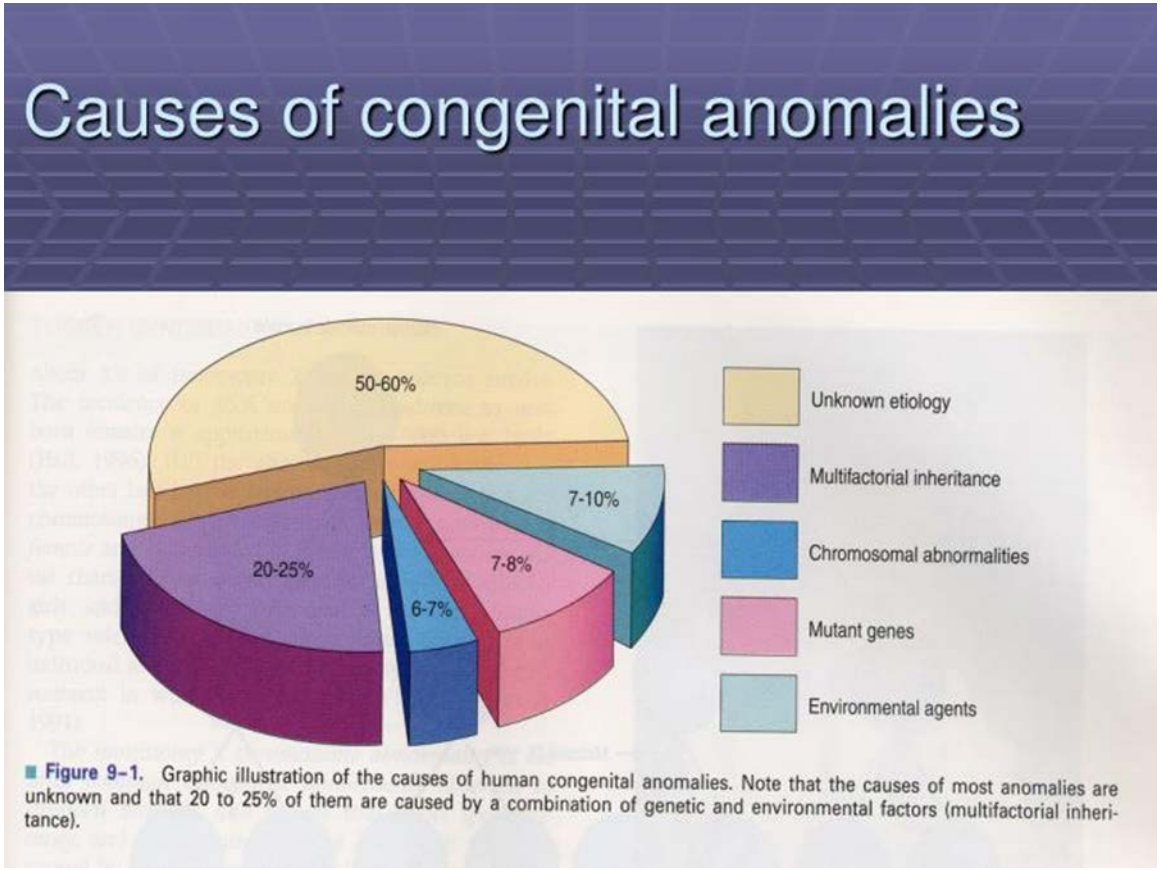


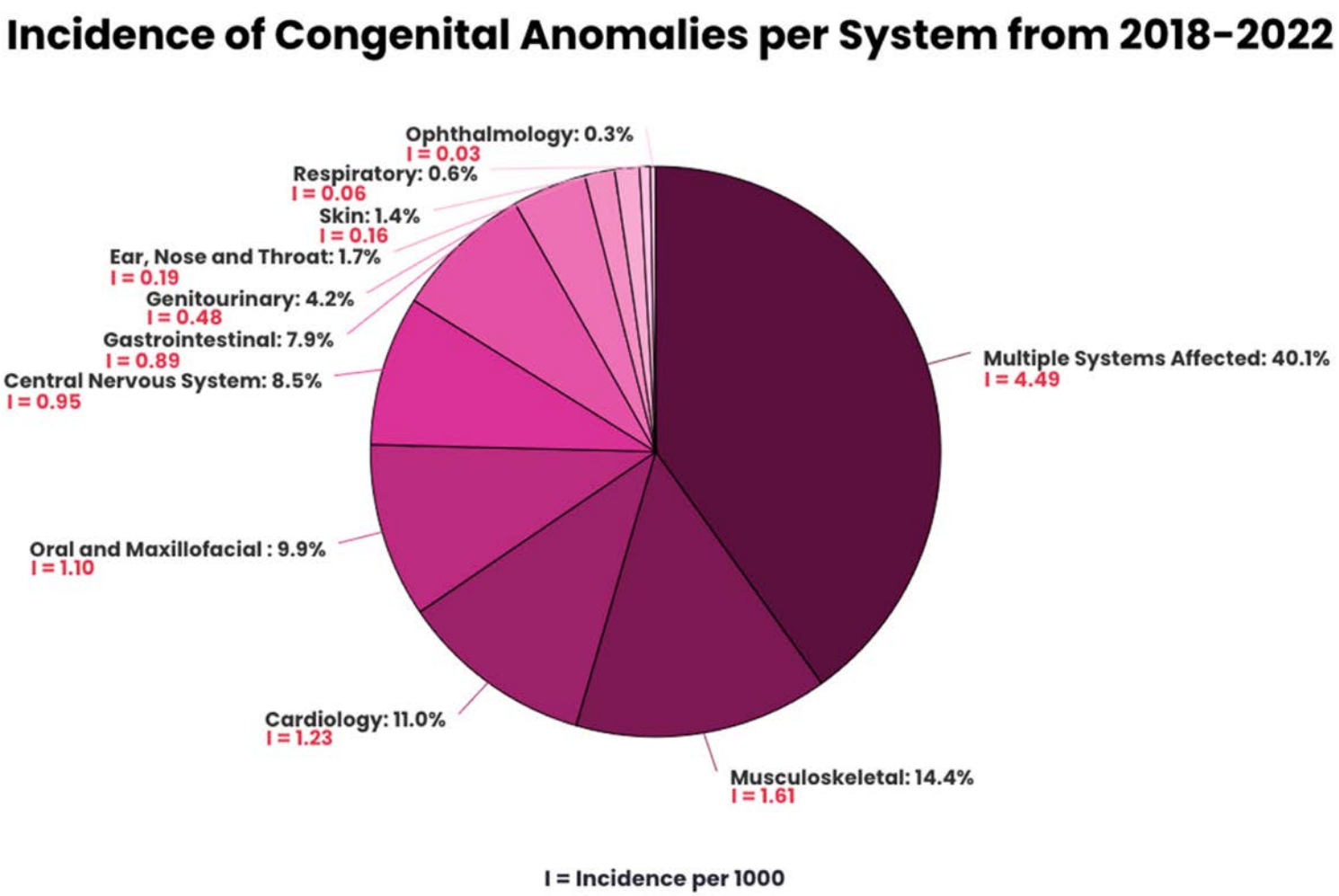
Figure 9–1. Graphic illustration of the causes of human congenital anomalies. Note that the causes of most anomalies are unknown and that 20 to 25% of them are caused by a combination of genetic and environmental factors (multifactorial inheritance).

Risk Factors

While congenital disorders have many causes, here are broad percentages:

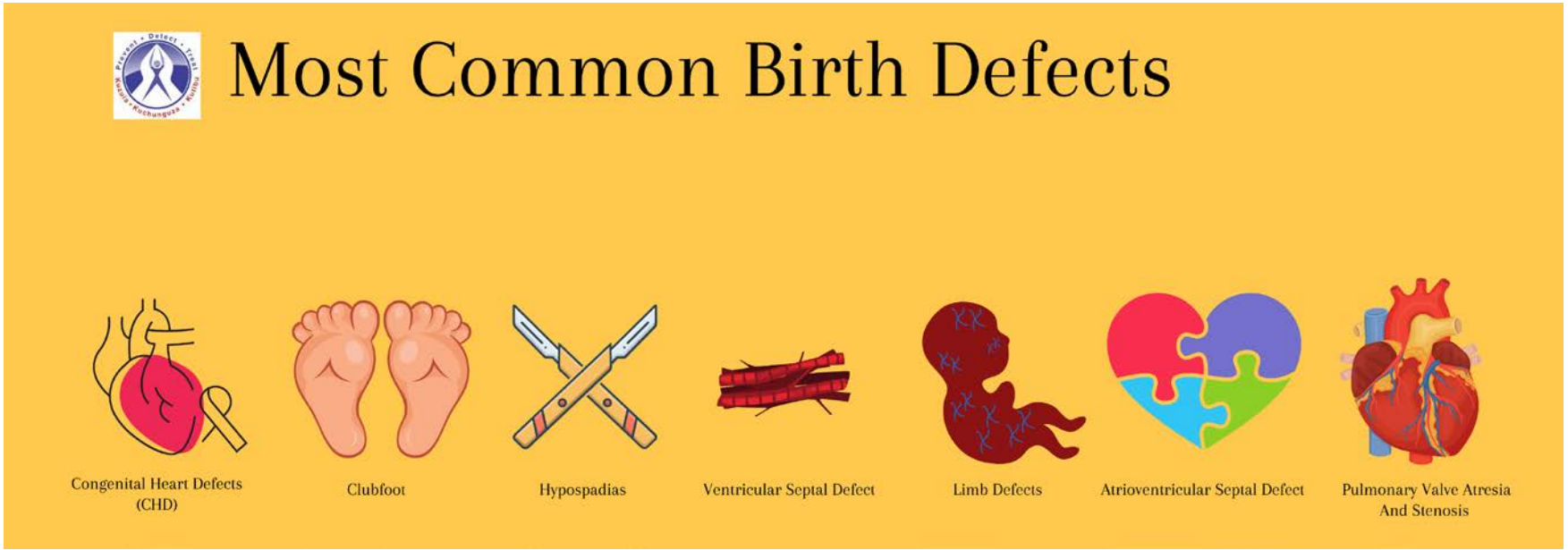
- **Genetic or chromosomal causes:** ~**20–30%**
- **Environmental/teratogenic factors** (e.g., infections, drugs, alcohol, radiation): ~**10%**
- **Multifactorial (gene–environment interactions):** ~**40–60%**
- **Unknown causes:** A substantial proportion remains unexplained.

Εισαγωγικά…



Buhasan, A.; Al Zayer, L.; Al-Jabery, L.; Mohamed, A.; Almadhoob, A.; Zaman, E.; Jeddy, R. Prevalence of Congenital Anomalies and Predictive Factors in Pregnant Women in Bahrain: A Retrospective Analysis. Life 2025, 15, 650. https://doi.org/10.3390/life15040650

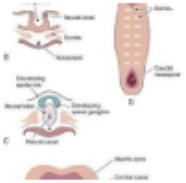
Εισαγωγικά…



Most Common Types

1. **Congenital heart defects** – the most common category, affecting about **1% of live births**.
2. **Neural tube defects** (e.g., spina bifida, anencephaly) – about **0.1–0.2% of births**, but rates vary by folic-acid intake.
3. **Chromosomal disorders** –
   - **Down syndrome (Trisomy 21)**: roughly **1 in 700 births** (varies by maternal age).
   - Other trisomies (13, 18) are less common.
4. **Orofacial clefts** – **~1 in 700 births**.
5. **Limb reduction defects**, **genitourinary defects**, and **gastrointestinal anomalies** occur at lower but significant frequencies.

ΚΝΣ



The most common congenital disorders of the Central Nervous System (CNS) are **Neural Tube Defects (NTDs)**, primarily **Spina Bifida** (including myelomeningocele, meningocele, and occulta) and **Anencephaly**, resulting from the neural tube failing to close early in development. Other frequent CNS anomalies include **Congenital Hydrocephalus** (fluid buildup in the brain), **Arnold-Chiari malformations**, and **Encephalocele**, often related to NTDs or structural issues.

Top Congenital CNS Disorders

1. **Neural Tube Defects (NTDs)**: The broadest and most common category, occurring when the neural tube (which forms the brain and spinal cord) doesn't close properly.
   - **Spina Bifida**: The most common NTD, ranging from mild (occulta, often asymptomatic) to severe (myelomeningocele, where spinal cord protrudes).
   - **Anencephaly**: Severe NTD where major parts of the brain and skull don't develop, often leading to stillbirth or early death.
2. **Hydrocephalus**: A buildup of cerebrospinal fluid (CSF) in the brain, causing increased pressure and head enlargement, often linked with NTDs or blockage.
3. **Arnold-Chiari Malformations**: Brain tissue extends into the spinal canal, often seen with spina bifida or hydrocephalus, causing symptoms like headaches or balance issues.
4. **Encephalocele**: A sac-like protrusion of the brain and membranes through an opening in the skull.

Other Related Conditions

- **Dandy-Walker Malformation**: Cyst in the back of the brain, affecting cerebellum and fluid flow.
- **Holoprosencephaly**: Failure of the forebrain to divide properly.
- **Agenesis of the Corpus Callosum**: Absence of the nerve fibers connecting the brain's hemispheres.

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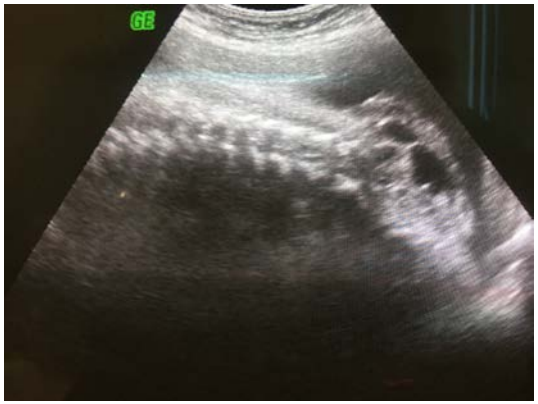

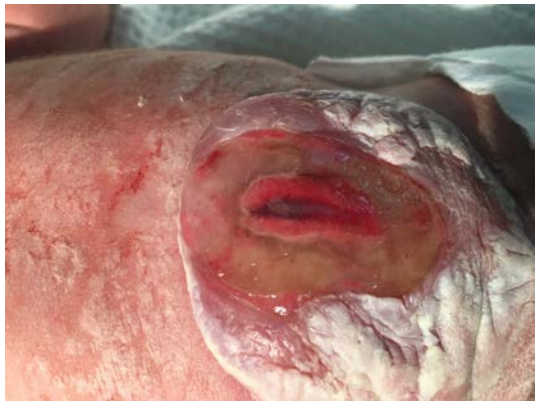

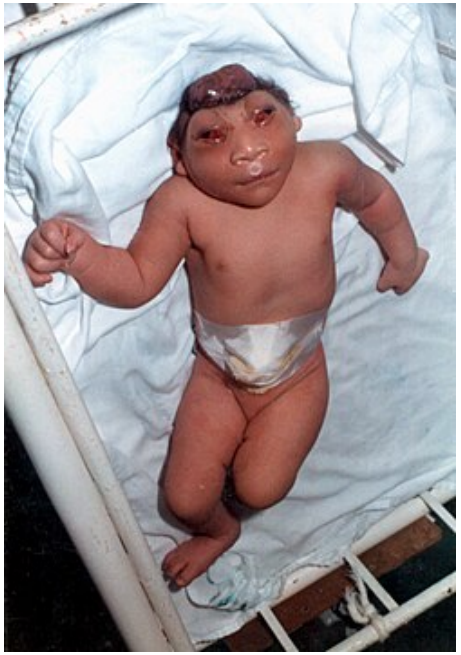

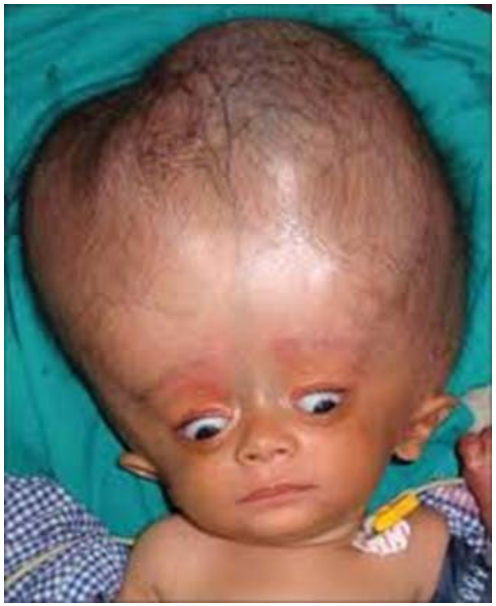

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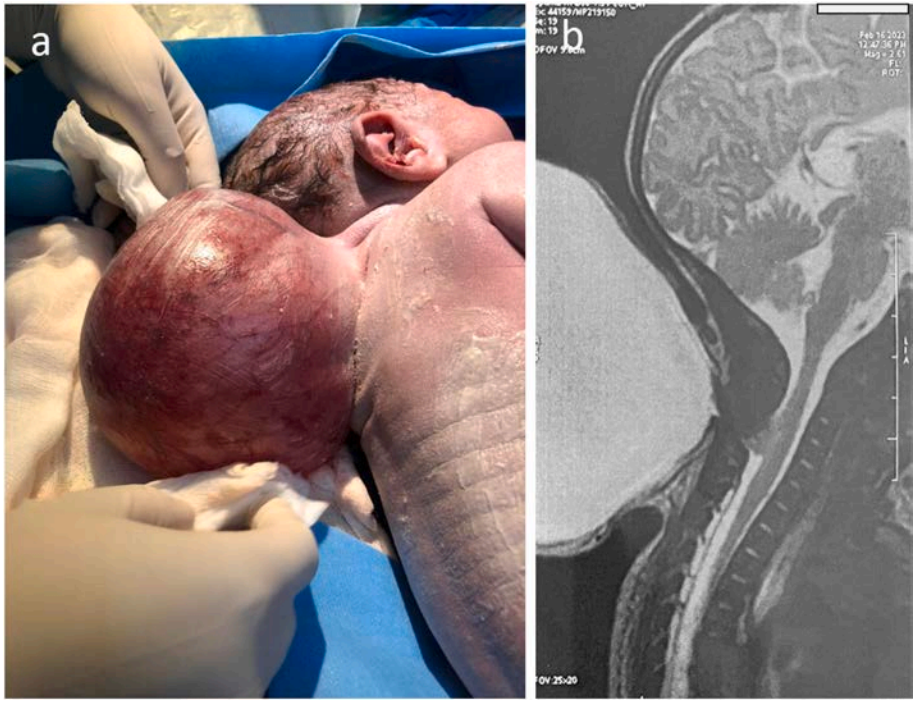
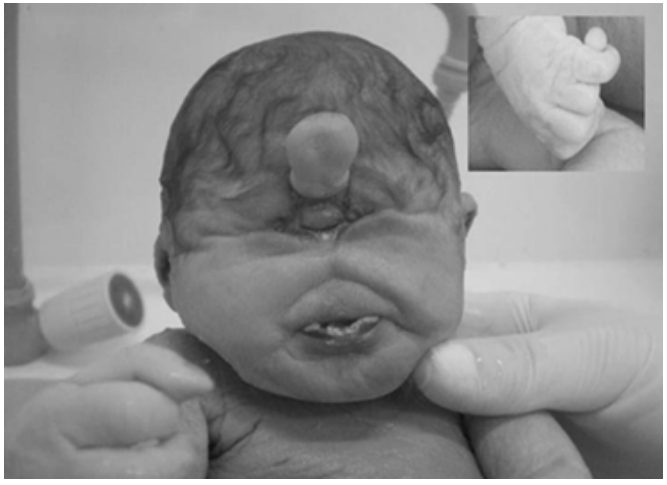




Μυοσκελετικό Σύστημα

The most common congenital musculoskeletal issues involve **limb/digit anomalies** (clubfoot, polydactyly, syndactyly, hip dysplasia, bowlegs/knock-knees), **spinal issues** (scoliosis, Klippel-Feil, spina bifida occulta, torticollis), and **muscle disorders** (arthrogryposis, congenital myopathies, muscular dystrophies like Myotonic Dystrophy). These often present as isolated problems or part of broader genetic syndromes, affecting feet, hips, spine, limbs, and muscles, requiring early diagnosis for proper management.

Common Congenital Musculoskeletal Conditions

Limb & Foot Anomalies

- **Clubfoot (Talipes Equinovarus):** Foot turns inward and downward.
- **Developmental Dysplasia of the Hip (DDH):** Hip socket doesn't fully cover the ball joint.
- **Bowlegs (Genu Varum) & Knock-knees (Genu Valgum):** Knees angle outward or inward.
- **Polydactyly/Syndactyly:** Extra fingers/toes or webbed digits.
- **Limb Deficiencies:** Underdevelopment or absence of limbs (e.g., fibular deficiency).

Spinal & Neck Issues

- **Scoliosis:** Sideways curvature of the spine, often with chest wall issues.
- **Klippel-Feil Syndrome:** Fused cervical (neck) vertebrae.
- **Congenital Torticollis:** Head tilt due to neck muscle issues.
- **Spina Bifida Occulta:** Incomplete closure of the spine.

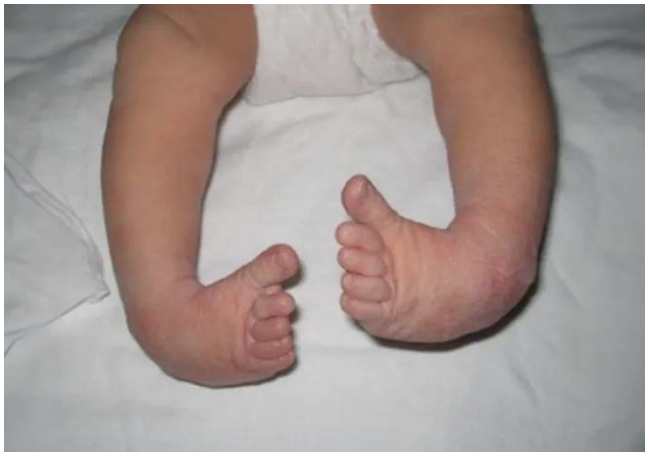
Muscle & Joint Disorders

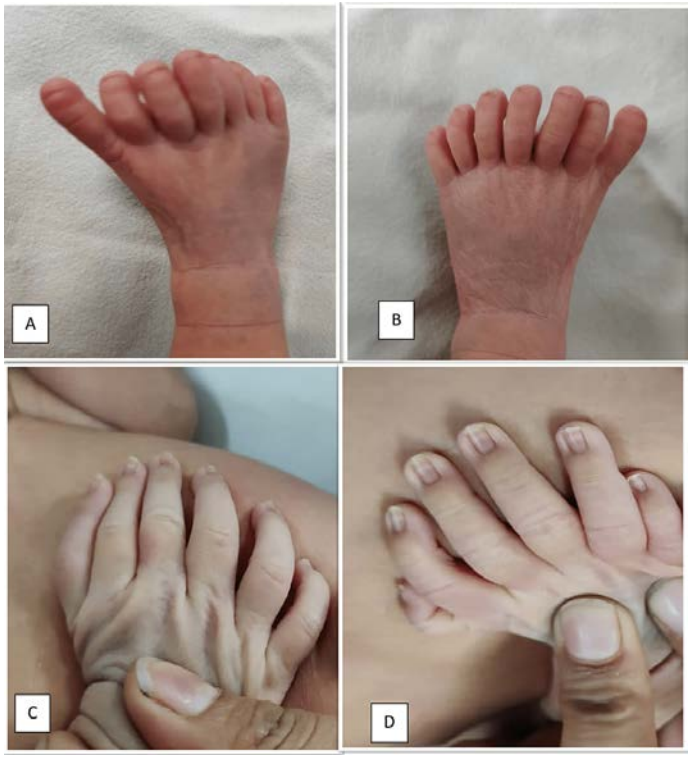
- **Arthrogryposis Multiplex Congenita:** Multiple joint contractures present at birth.
- **Congenital Muscular Dystrophies:** Muscle weakness present from birth.
- **Congenital Myopathies:** Primary muscle disorders causing weakness and hypotonia.

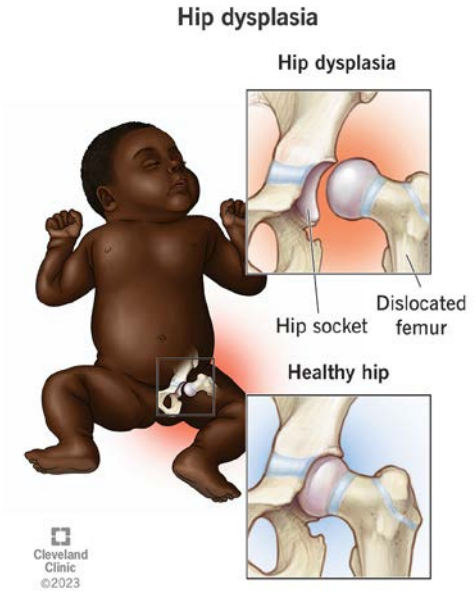
Syndromes & Other

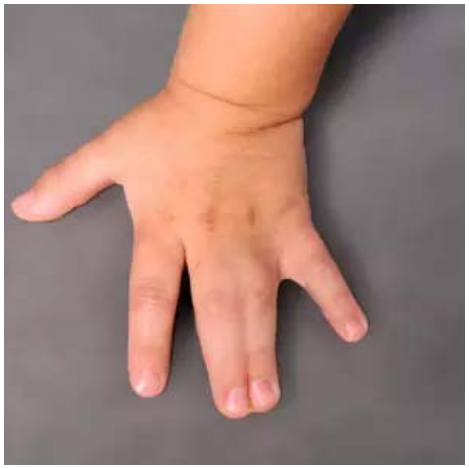
- **Osteogenesis Imperfecta:** Brittle bone disease.
- **Craniosynostosis:** Premature fusion of skull bones (e.g., Crouzon syndrome).

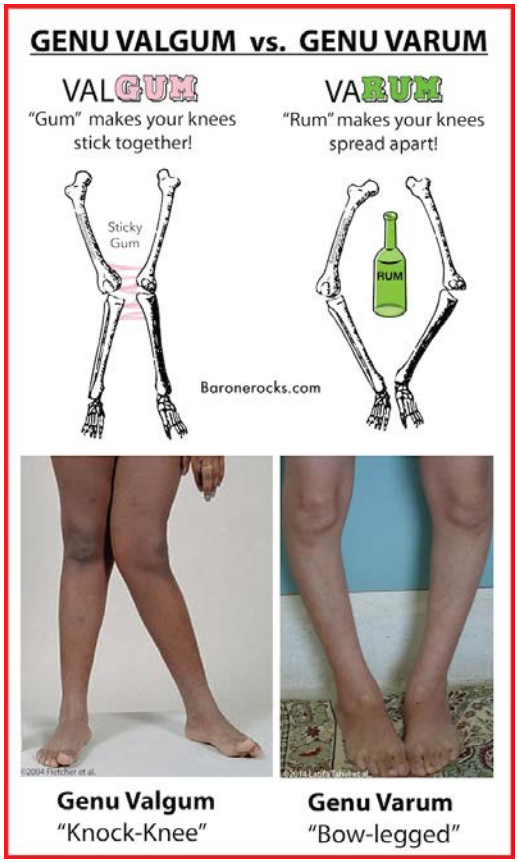
Μυοσκελετικό Σύστημα











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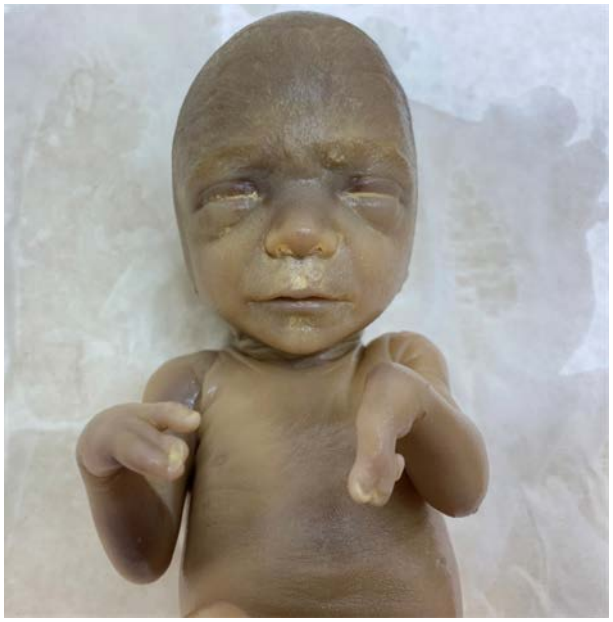
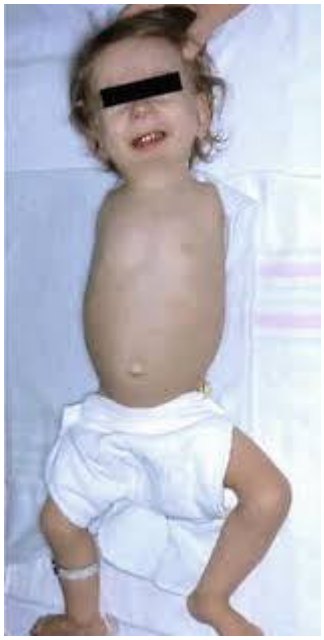
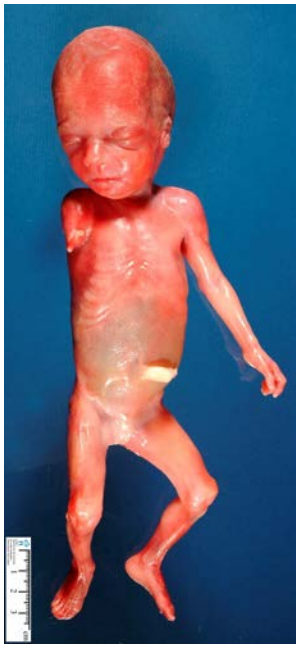
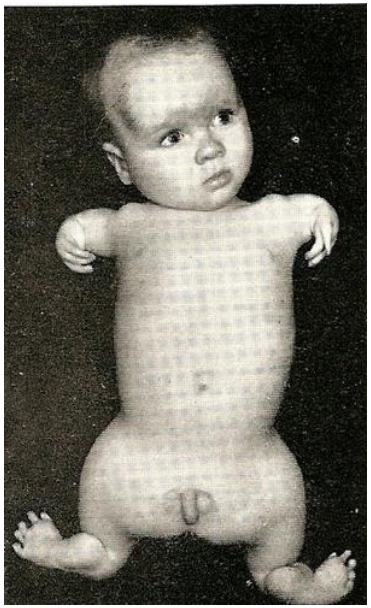
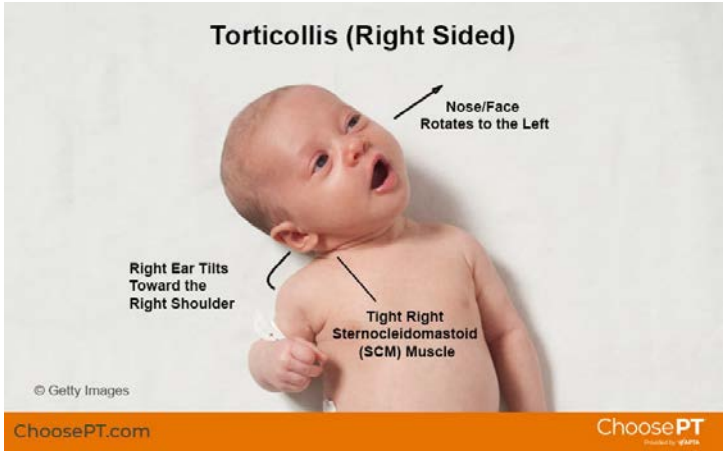
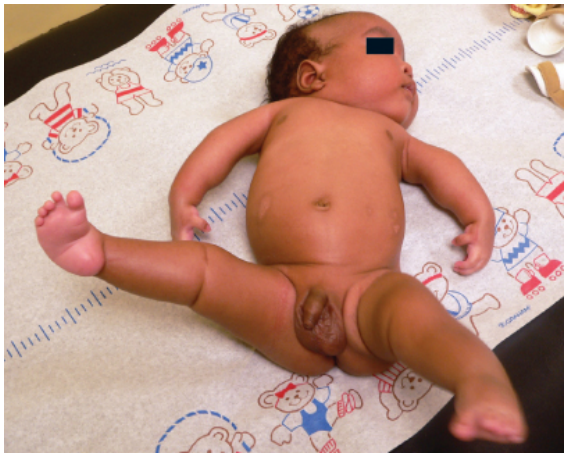




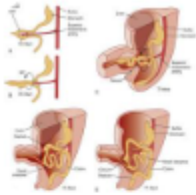








Γαστρεντερικό Σύστημα

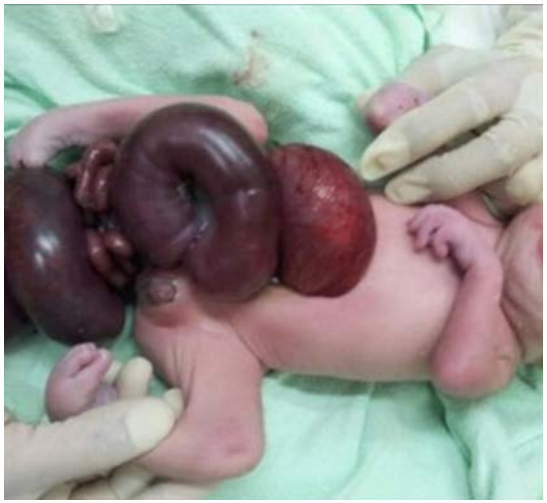


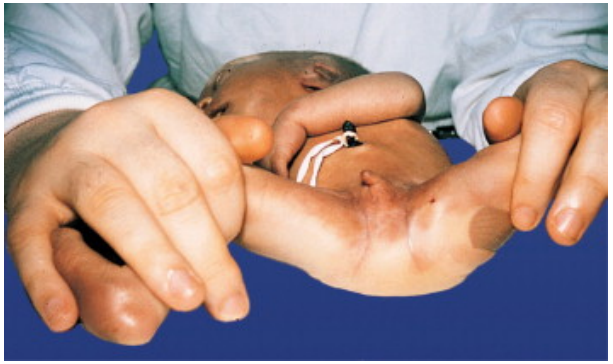
The most common congenital GI disorders include **Meckel's Diverticulum** (a small pouch in the small intestine), various types of **Atresia & Stenosis** (blockages in the esophagus, intestines), **Hirschsprung's Disease** (nerve absence in the large intestine), and **Malrotation** (improper organ rotation), alongside **Anorectal Malformations**, all impacting digestion and requiring potential surgical correction, with some associated with syndromes like VACTERL.

Common Congenital GI Disorders:

- **Meckel's Diverticulum:** A small pouch in the small intestine, considered the most frequent birth defect of the GI tract, affecting about 1 in 50 babies.
- **Atresia & Stenosis:** Blockages (atresia) or narrowing (stenosis) in the digestive tract, with esophageal atresia (often with fistula) and intestinal atresia (duodenal, jejunoileal) being frequent.
- **Hirschsprung's Disease:** Absence of nerve cells (ganglia) in the large intestine, preventing normal muscle contractions and bowel movements.
- **Malrotation & Volvulus:** Incorrect rotation of the intestines during fetal development, potentially leading to twisting (volvulus) and obstruction.
- **Anorectal Malformations (Imperforate Anus):** Malformations of the anus and rectum, often requiring surgical repair.
- **Abdominal Wall Defects:** Like gastroschisis or omphalocele, where organs form outside the body.

Γαστρεντερικό Σύστημα





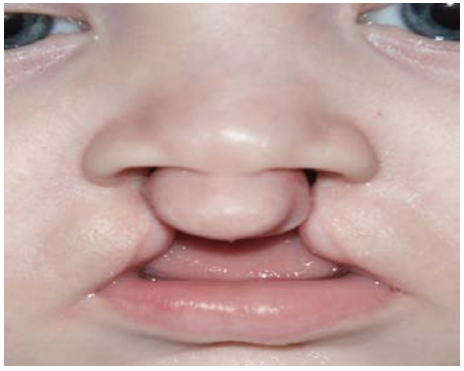
Συγγενείς Διαμαρτίες Κεφαλής & Προσώπου

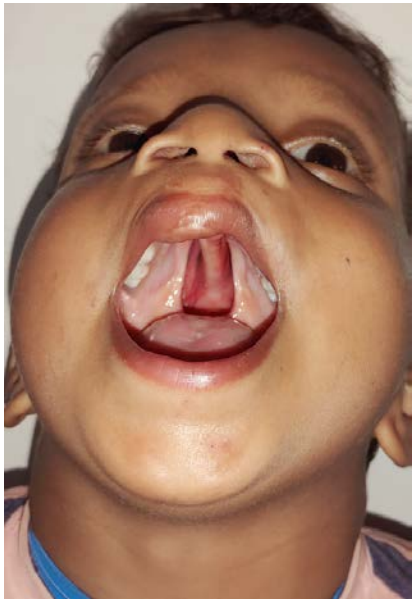
The most common congenital facial disorders are **Cleft Lip and Palate**, affecting the lip, roof of the mouth, or both, making them the leading craniofacial anomaly, followed by conditions like **Craniosynostosis** (premature skull bone fusion) and **Hemifacial Microsomia** (underdeveloped one side of the face). Other frequent issues include facial paralysis, ear/jaw deformities, and vascular malformations like hemangiomas or port-wine stains.

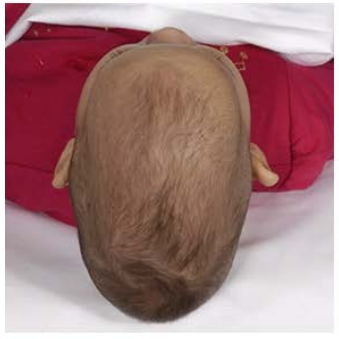
Top Common Conditions:

- **Cleft Lip and Palate:** A separation in the upper lip or roof of the mouth (palate), or both, occurring in about 1 in 1000 births.
- **Craniosynostosis:** Early fusion of the skull's soft spots (sutures), hindering normal brain and skull growth.
- **Hemifacial Microsomia (Goldenhar Syndrome):** Underdevelopment of one side of the face, affecting the ear, mouth, and jaw.
- **Vascular Malformations & Hemangiomas:** Birthmarks made of blood vessels (like port-wine stains) that can affect appearance and function.

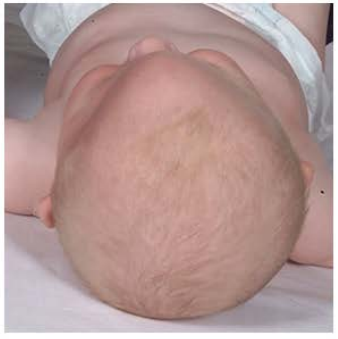
Συγγενείς Διαμαρτίες Κεφαλής & Προσώπου





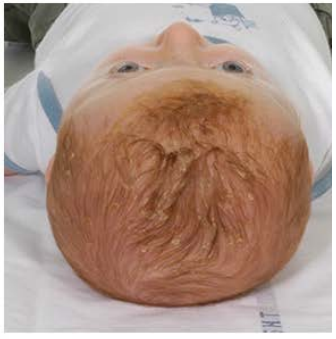


Scaphocephaly



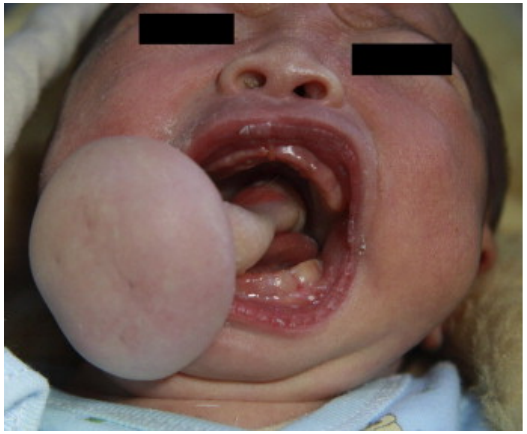
Trigonocephaly

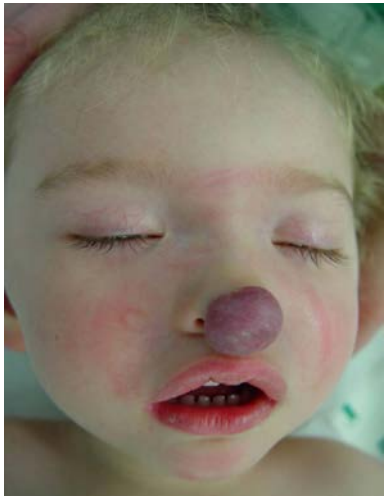
Plagiocephaly

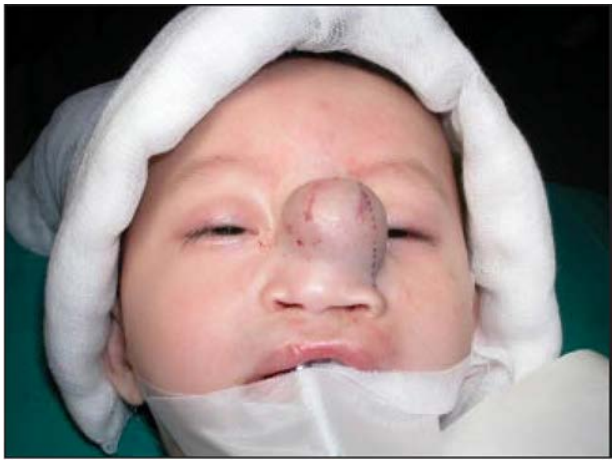


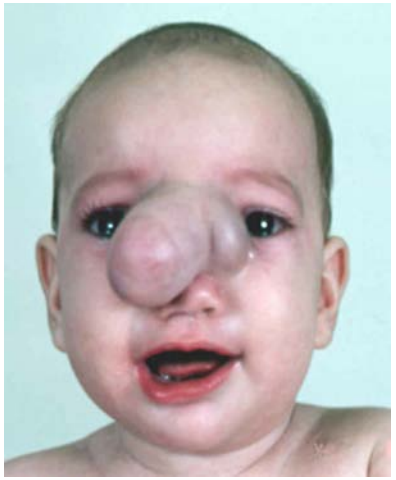
Brachycephaly

Συγγενείς Διαμαρτίες Κεφαλής & Προσώπου

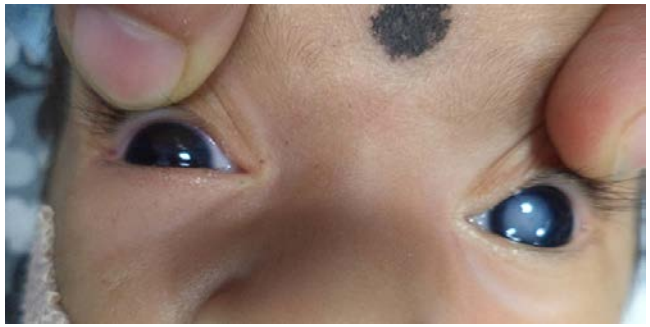


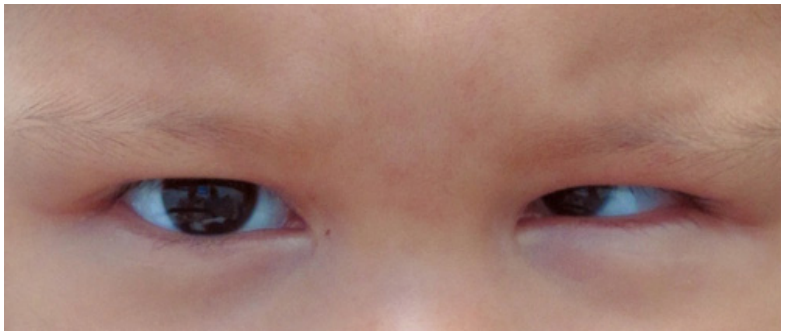


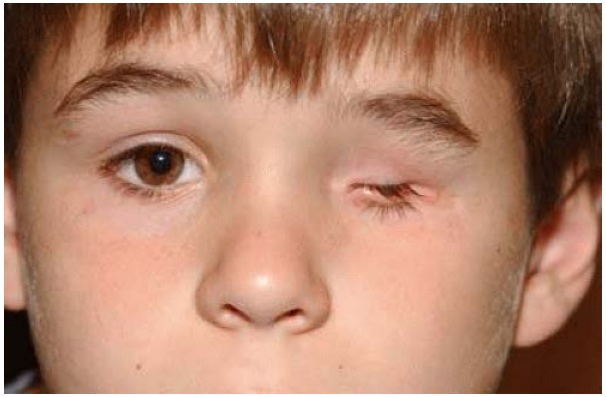




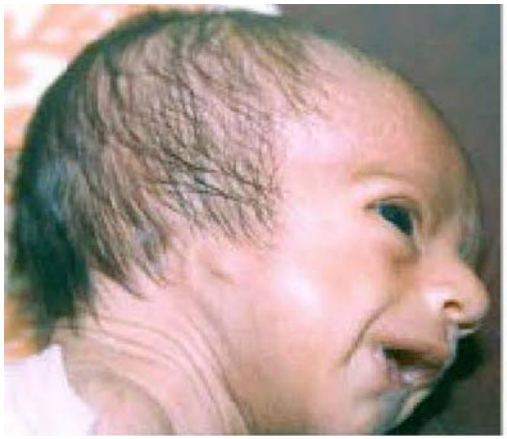
Συγγενείς Διαμαρτίες Κεφαλής & Προσώπου

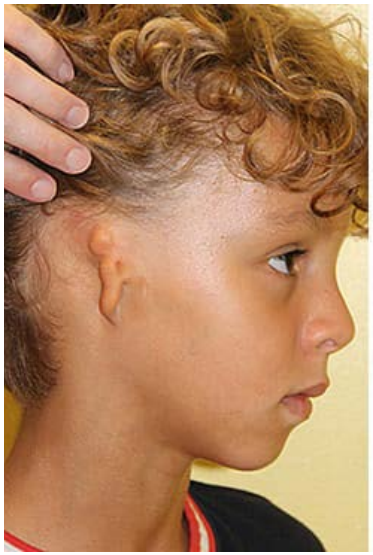


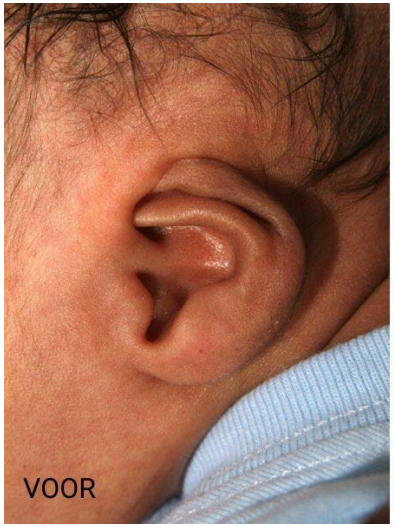


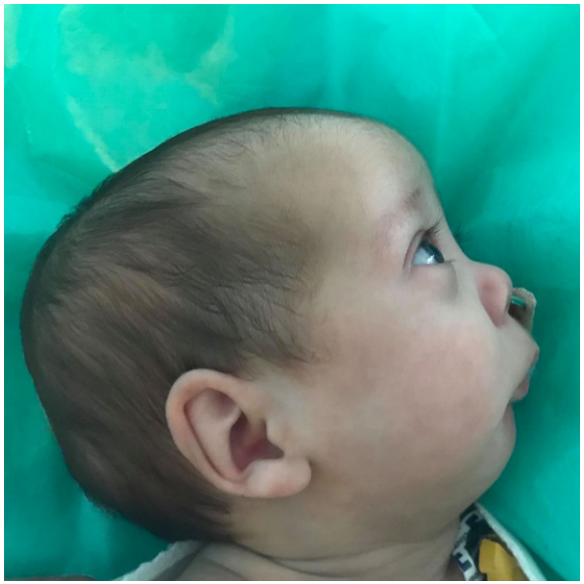


Συγγενείς Διαμαρτίες Κεφαλής & Προσώπου










The End!
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