Cranial & Vertebral Anomalies

Last updated: September 13, 2019

SKULL

Head circumference measurement - **occipital-frontal circumference (OFC)** - is routine part of physical assessment of all children ≤ 2 yrs! for norms & charts see p. D5 >>

- *stimulus for head growth* is increase in volume of intracranial contents.

NORMAL DEVELOPMENT

FONTANELLES

Anterior fontanelle: largest fontanelle, diamond shaped, size 4 (AP) x 2.5 (transverse) cm at birth.

- closes by age 2.5 yrs.

Posterior fontanelle: triangular.

- closes by age 2-3 mos.

Sphenoid and mastoid fontanelles: small, irregular.

- sphenoid closes by age ~ 2-3 mos, mastoid by age 1 year.

CRANIAL VAULT

Growth: largely determined by growth of brain;

- 90% of adult head size is achieved by age 1 yr; 95% by age 6 yrs.
- growth essentially ceases at age 7 yrs.

Layers: skull is unilaminar at birth.

- diploe appears by 4 yr and reaches maximum by age 35 yrs (when diploic veins form).

Parietal bossing is formed by motor cortex growth.

Mastoid process: formation commences by age 2 yrs, air cell formation occurs during 6th yr.

SUTURES

- calvarial sutures serve 2 important functions:
  1) head malleability during passage through birth canal.
  2) separation of calvarial bones during intrauterine ÷ early perinatal growth.
- ossification of cranial vault starts in central region of each cranial bone and extends outward toward cranial sutures.
- by end of 2nd yr, bones have interlocked at sutures and further growth occurs by accretion and absorption (sutures serve as site of bone deposition in growing calvarium).

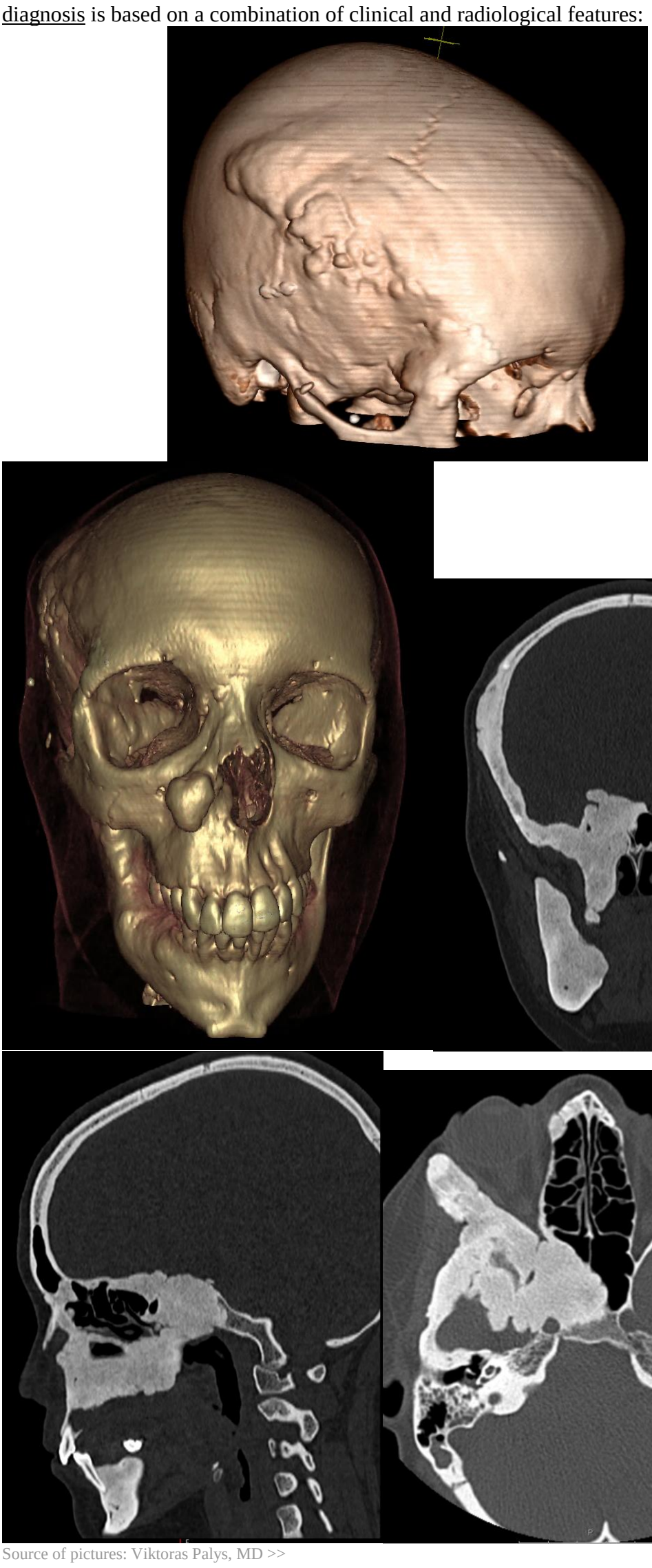
Skull growth occurs perpendicular to suture!

- primary factor that keeps sutures open is ongoing brain growth.
- suture closure occurs by age ≈ 12 years, but completion continues into 3rd decade.

MELORHEOSTOSIS

- rare skeletal abnormality that causes abnormal growth of new bone tissue on top of existing bones.

- around half of cases of isolated melorheostosis are due to acquired, somatic mutations in the MAP2K1 gene; these mutations are not inherited from a parent and occur randomly during a person's lifetime.
- signs and symptoms typically appear by late childhood or adolescence - deformity, contracture, chronic pain, stiffness, and limited range of motion.
  - in some cases, the overlying skin and soft tissue may show thickening, shininess, reddening or darkening, linear scleroderma, and/or swelling.
  - typically affects the long bones, and the legs are affected more often than the arms.
  - sometimes the small bones of the hand or foot are affected, and rarely, bones of the skull or trunk are affected.
  - not life-threatening, but chronic pain can greatly impact quality of life.
- diagnosis is based on a combination of clinical and radiological features:



Source of pictures: Viktoras Palys, MD >>

- management is symptomatic; in skull may need surgical cranial nerve decompression.

CRANIOSYNOSTOSIS

- **premature fusion** of one or more of 6 cranial sutures → abnormal growth of cranium.
  a) primary defect of ossification (PRIMARY CRANIOSYNOSTOSIS)
  b) primary brain growth failure (SECONDARY CRANIOSYNOSTOSIS) - more common (92-98%)!

- overall INCIDENCE – 0.6 / 1000 live births.
  N.B. craniosynostosis is *in utero event*!

ETIOPATHOGENESIS

80-90% are sporadic **ISOLATED** cases;
10-20% cases are **PART OF SYNDROME** (*syndromic craniosynostoses*); > 70 syndromes include craniosynostosis.

1. Many cases are of **unknown etiology**.
2. **Nongenetic** causes:
   1) **metabolic conditions** that can lead to premature fusion of cranial sutures (hyperthyroidism, hypercalcemia, hypophosphatasia).
   2) **hematologic disorders** that cause bone marrow hyperplasia (e.g. sickle cell disease, thalassemia).
   3) severe **constraints in utero** (e.g. amniotic band rupture sequence).
3. **Mutations** (10-20% cases) in family of **FIBROBLAST GROWTH FACTOR RECEPTORS (FGFR)**:
   FGFR1 gene - Pfeiffer's syndrome.
   FGFR2 gene (chromosome 7) - Crouzon's syndrome, Apert's syndrome, Jackson-Weiss syndrome, Pfeiffer's syndrome.
   FGFR3 gene - thanatophoric dysplasia, achondroplasia.
   – mutations in **other genes** are rare (e.g. homeobox gene MSX2 - Boston type of craniosynostosis).
   – gene locus for SINGLE SUTURE craniosynostosis has not been identified.

PATHOPHYSIOLOGY

- prevailing hypothesis suggests that *abnormal development of skull base* creates exaggerated forces on dura that act to disrupt normal cranial suture development.
  N.B. dysfunctional osteoblasts or osteoclasts are not responsible!

CLINICAL FEATURES

- commonly present at birth (but not always noticeable); certainly manifests as clinical deformity in first few months of life.
  N.B. it is PRENATAL abnormality!

1) **abnormal skull growth** → **cosmetic** facial and cranial deformity (often with visible / palpable ridging of closed suture); worsen over time!
   skull growth *restricted* - in plane perpendicular to affected suture ("hand grabs and holds skull at suture");
   skull growth *enhanced* - in plane parallel to affected suture.
   Skull base growth is different in various types of craniosynostosis - important for final skull shape!; morphology of cranial base has been shown to be normalized following cranial expansion surgery in some synostoses!

2) **ICP↑** - only when > 1* suture is affected (cause and mechanism is not well understood** – may be present even in cases where absolute intracranial volume is increased) → adverse effects on development! (sun-setting eyes, headaches, vomiting, school performance↓, gradual visual failure).
   *esp. in syndromic cases (some experts say – up to 11% of single suture cases cause ICP↑)
   **abnormalities of cerebral venous drainage due to maldevelopment of foramina at skull base
   N.B. papilledema is rarely seen even in presence of intracranial hypertension!
   N. B. hydrocephalus is rare! (assertion that CSO may follow CSF shunting for HCP is unproven)

3) **airway problems** in syndromic cases (hypoplastic maxilla → dental malocclusion, difficulty breathing through nose; sleep apnea).

4) **vision loss** (coronal synostosis can cause amblyopia).

DIAGNOSIS

1. **Skull XR**
   - initially – 4-view + Towne.
   - to visualize all sutures, special Waters views must be taken.
     N.B. make sure you see suture of interest on XR before patient leaves radiology facility (then request radiology rapport on that suture)
   - **sutures** - straight with heaped-up sclerotic margins or completely absent (invisible).
   - indentations of inner table (evidence of ICP↑).
   - any suture is functionally closed even if it has closed only over short distance.
   - several **indices** have been devised and used for comparisons (most popular - *cranial index described by Cronqvist*).

2. **CT with 3D reconstruction** (method of choice!, esp. before surgery) - fused sutures are clearly identified; abnormal contour of skull is better appreciated; skull base is clear; 3D-CT is especially indicated in multiple-suture synostosis – to assist surgery.

3. Direct **mutation analysis** of FGFR genes.

4. PRENATAL detection with **3D ultrasonography**.

SINGLE-SUTURE SYNOSTOSES

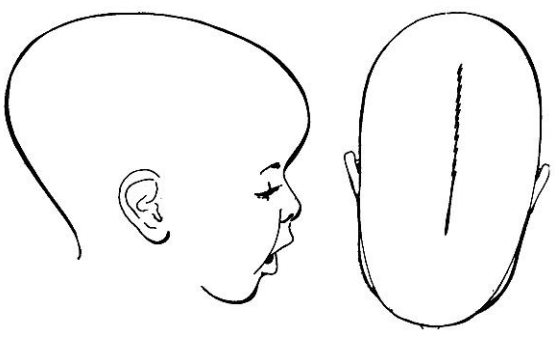
- can be very mild phenotypically; majority are sporadic; only rarely causes neurologic deficit.

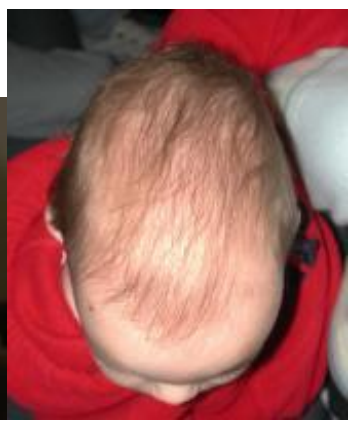
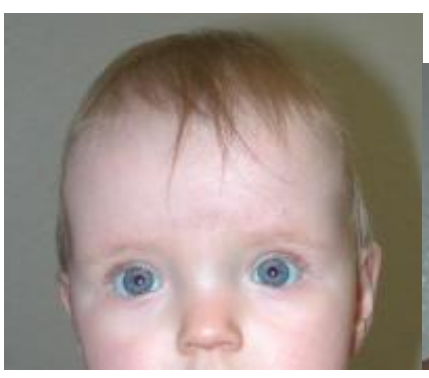
Sagittal 50-60%, coronal 20-30%, metopic 4-10%, lambdoid 2-4%.

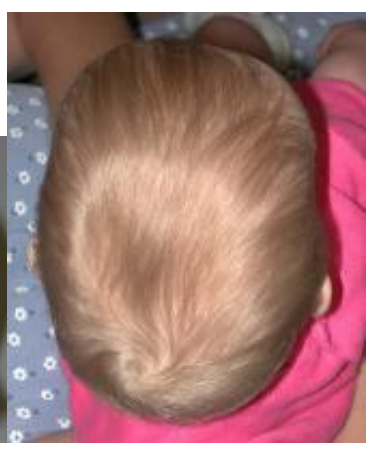
Sagittal synostosis → scaphocephaly

(most often affected suture! – seen in ≈ 55% of all cases!)
- elongated skull with compensatory frontal bossing and exaggerated occiput (occipital bathrocephaly), absent anterior fontanelle.
N.B. **DOLICHOCEPHALY** term is reserved for normal anatomic variant!

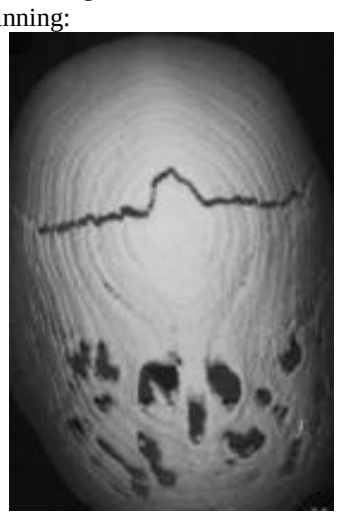
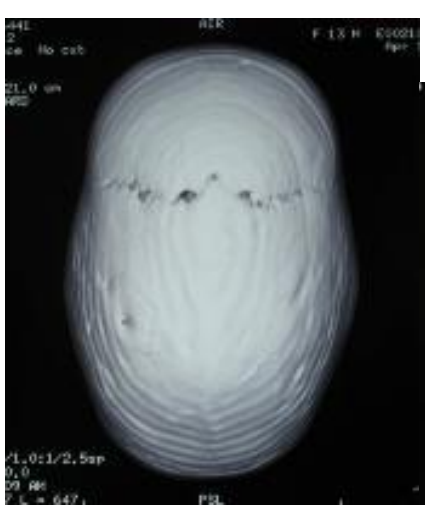
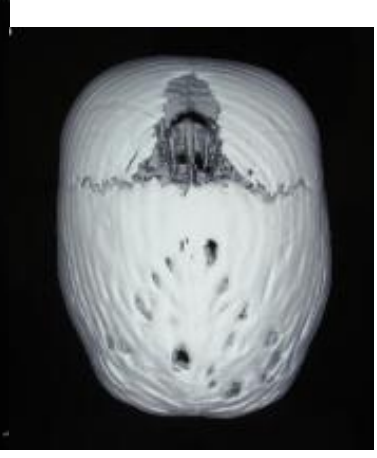
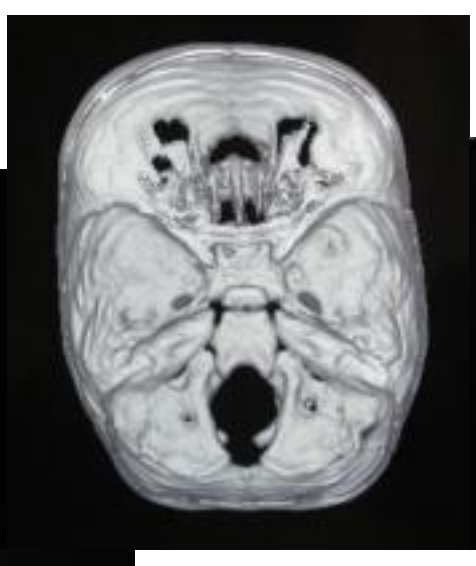
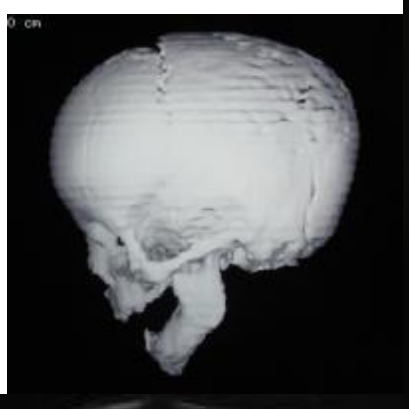
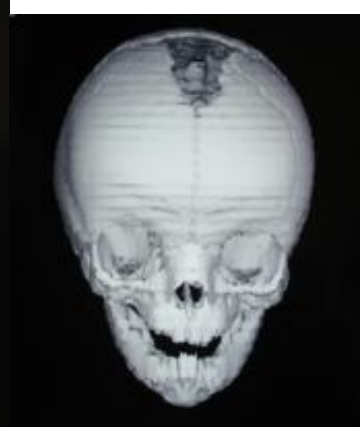
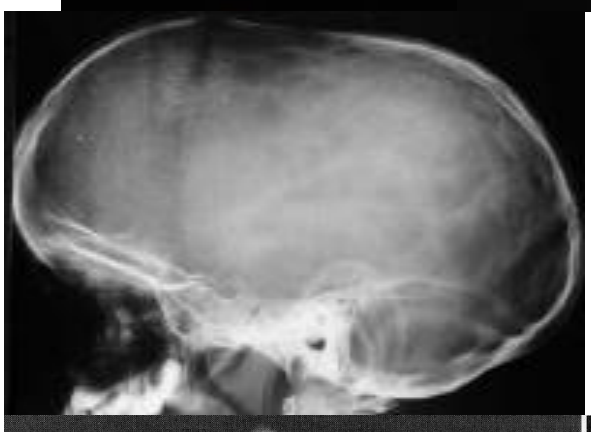
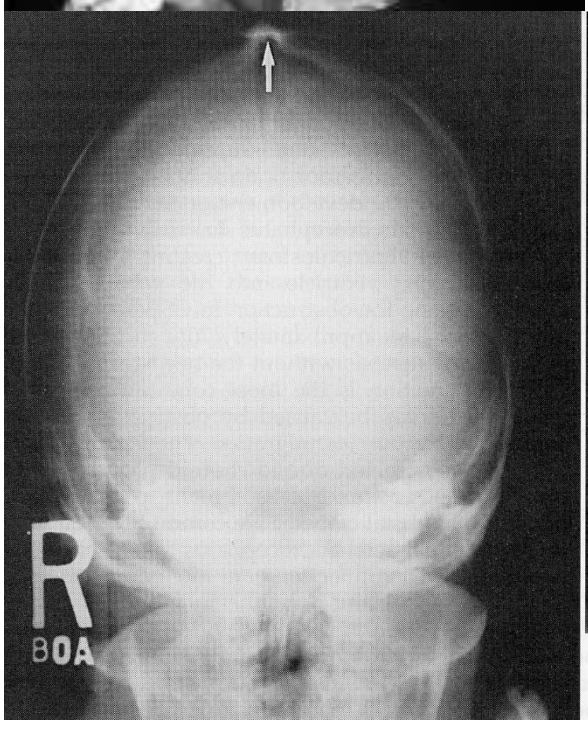
- **head circumference** is above 95th percentile (although biparietal diameter is low) - actual ***intracranial volume*** is normal or even increased - brain growth impairment does not occur (although ICP may be elevated in some cases) - **no neurological deficits**!
- normal face.
- often causes *labor difficulties* (cephalopelvic disproportion).
- frequent in premature infants.
- 80% are males.

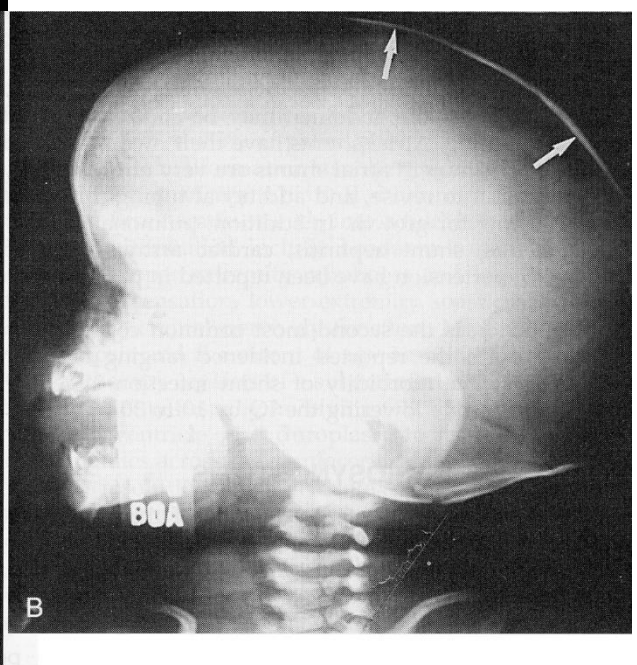




3D-CT scan - complete fusion of sagittal suture, with patent coronal suture and elongated cranial contour; apparent holes in posterior parietal regions are due to normal thinning:

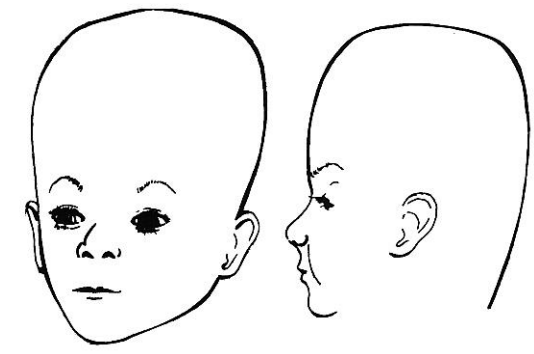


Coronal synostosis → brachycephaly

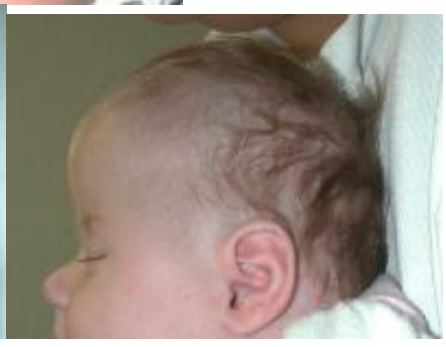
(18-30% of all cases)

- <u>foreshortened skull</u> and corresponding enlargement of bitemporal and biparietal diameter:
  - variable degree of exophthalmos (shallow orbits)!!!
  - orbits may be elliptical (i.e. **HARLEQUIN features**).
  - fronto-orbital bar is recessed; consequently, supraorbital rim is more posterior to corneal plane (normally, rim is 2 mm ventral to corneal plane).
  - higher incidence of *neurological complications*:
    1) optic atrophy (traction of chiasm and optic nerves due to upward displacement of chiasm + ICP↑)
    2) mental retardation.

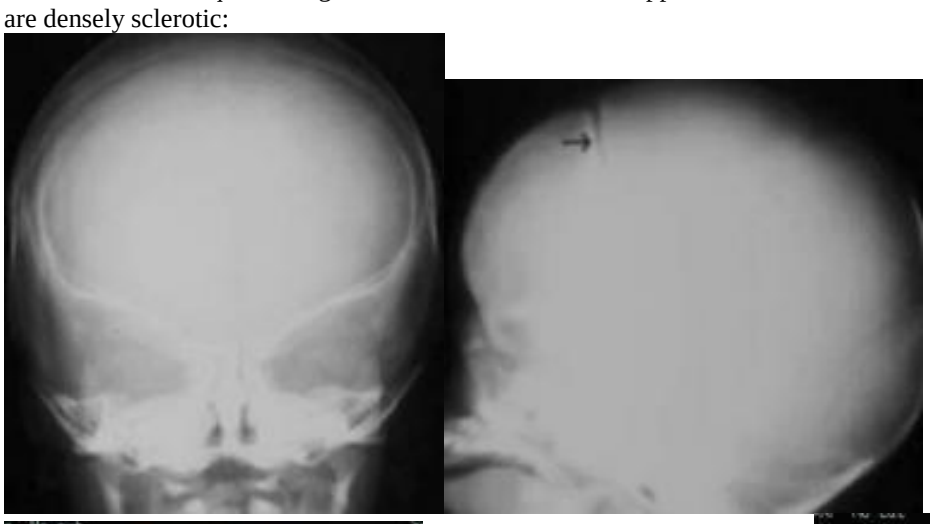
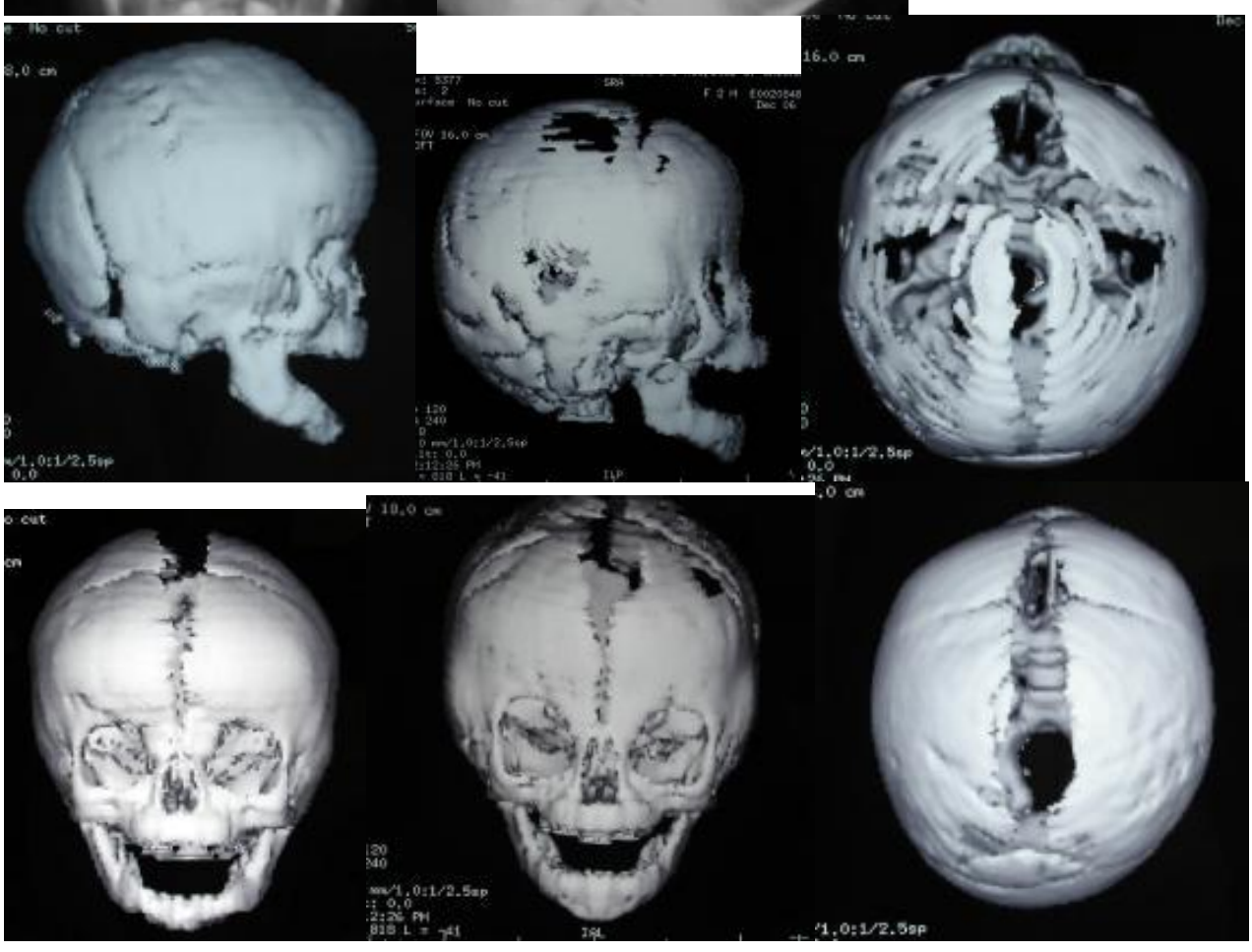
- often syndromic (e.g. Apert's syndrome).
- more common in females.







Note bilateral harlequin configuration of orbits and slitlike appearance of coronal suture (*arrow*); margins of coronal suture are densely sclerotic:

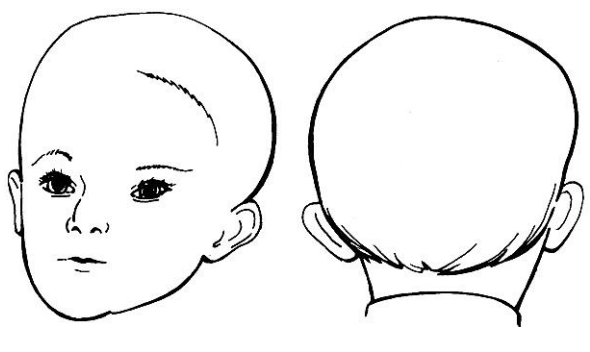
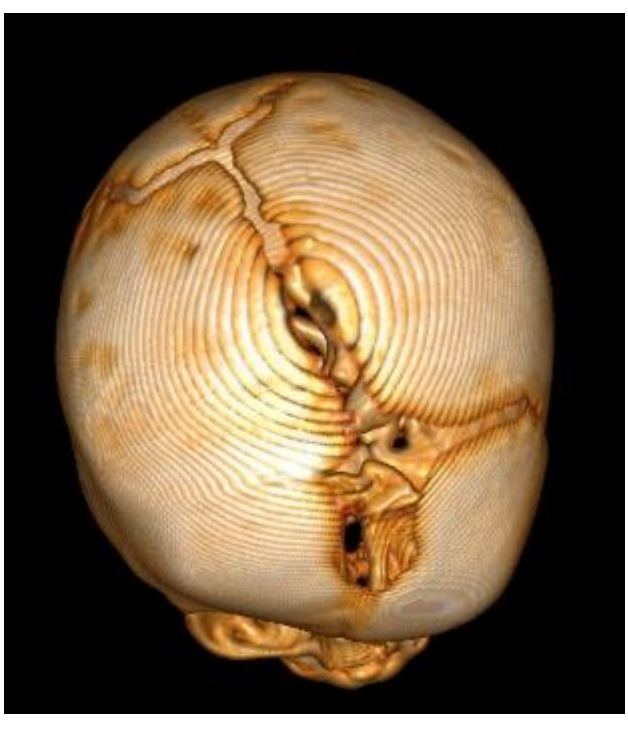



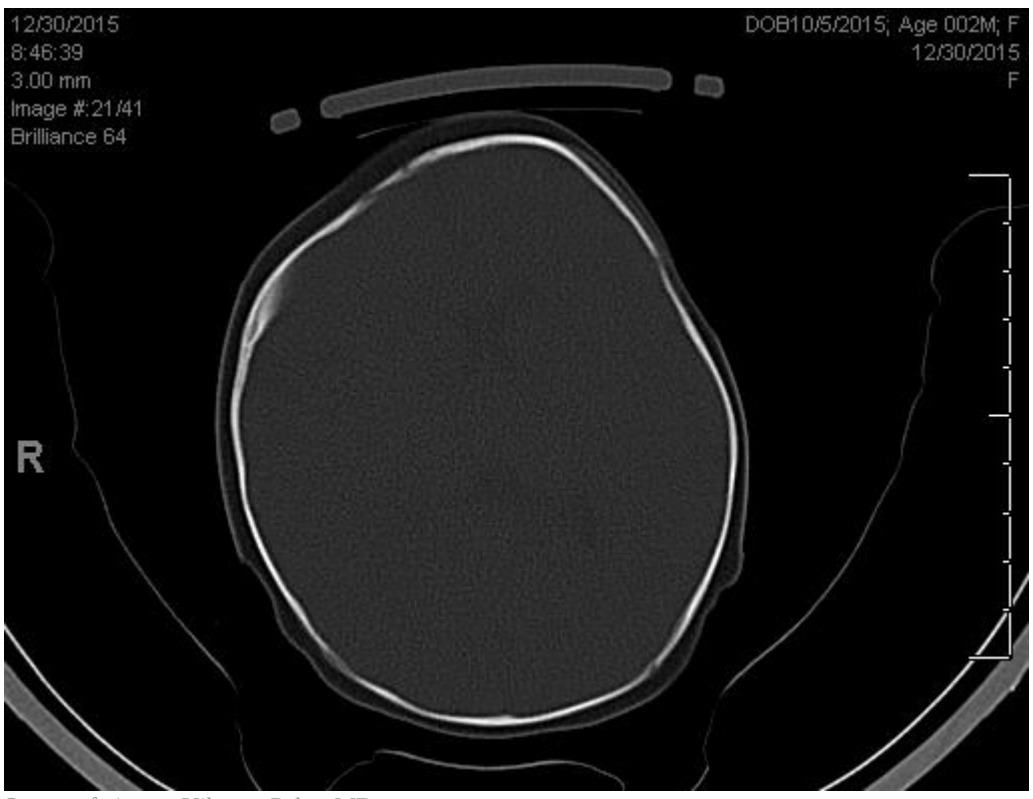
Unicoronal synostosis → anterior plagiocephaly

- flattening of ipsilateral frontal and parietal bones, bulging of contralateral frontal region, and bulging of ipsilateral temporal bone; *displacement of eyebrow downwards* on that side, asymmetric orbits, nose curvature (*nasal root deviated* toward fused suture).

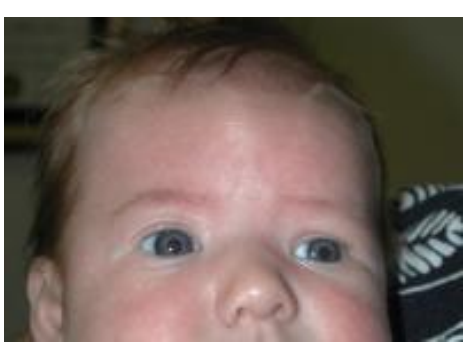
Unilateral cases outnumber bilateral forms by 2:1 !

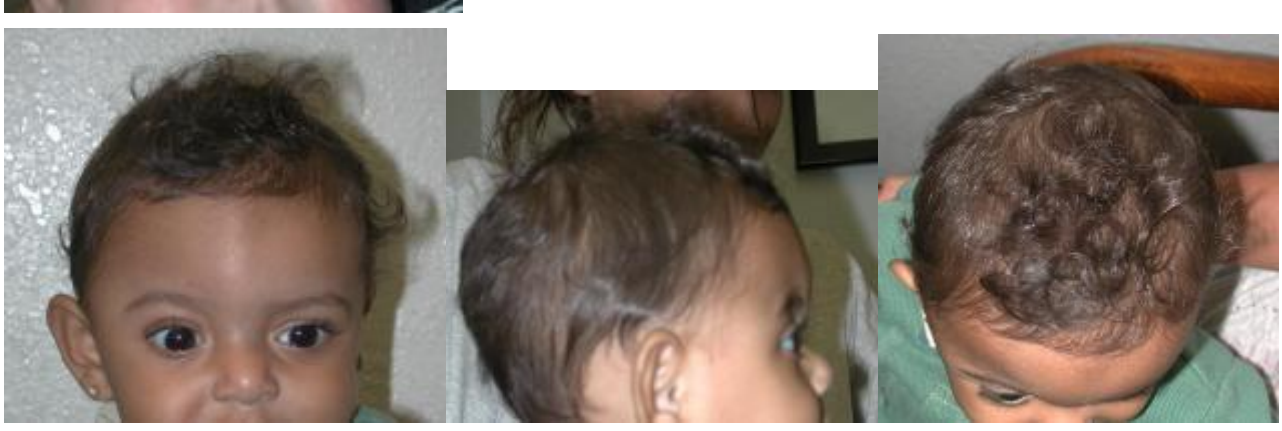
- parents often like affected **HARLEQUIN** eye (bigger) more than normal eye.

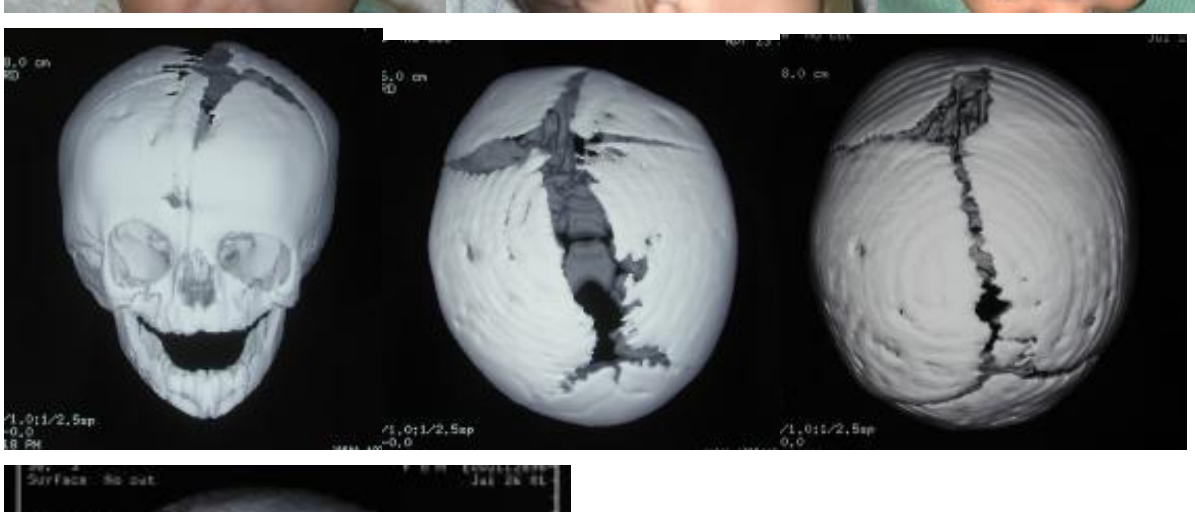



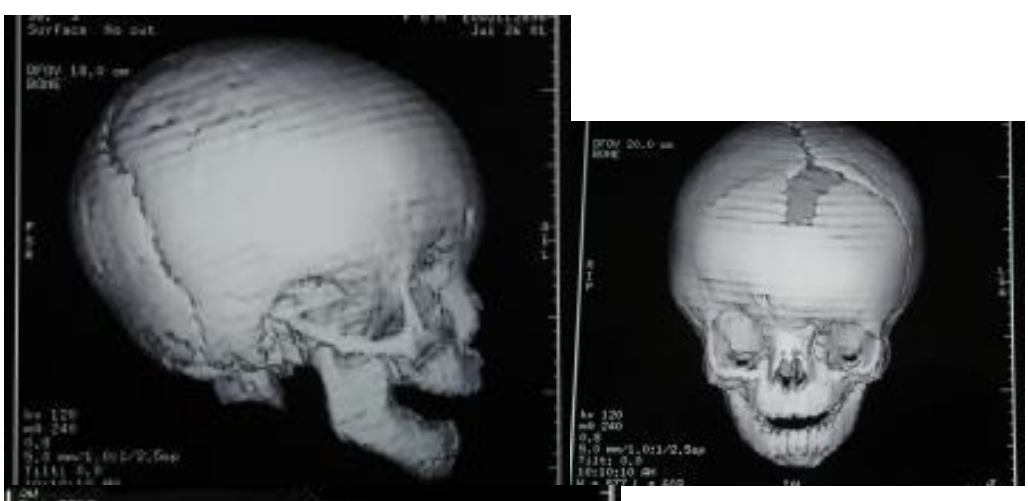


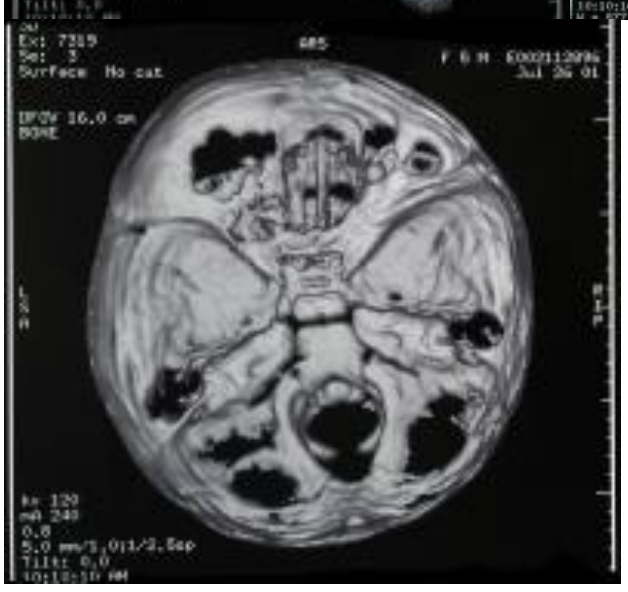
Source of picture: Viktoras Palys, MD >>



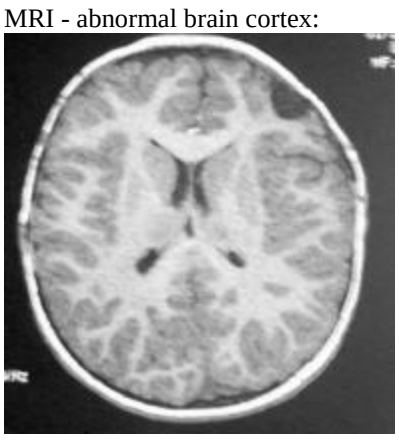








MRI - abnormal brain cortex:

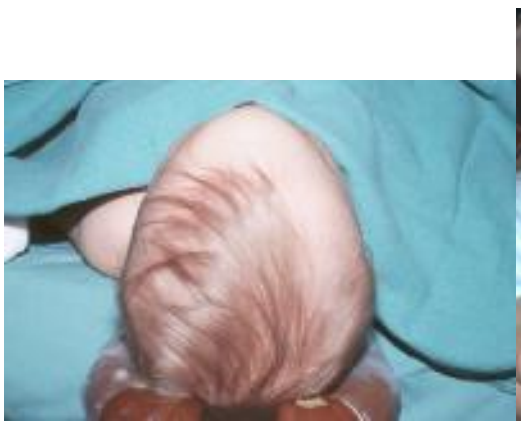
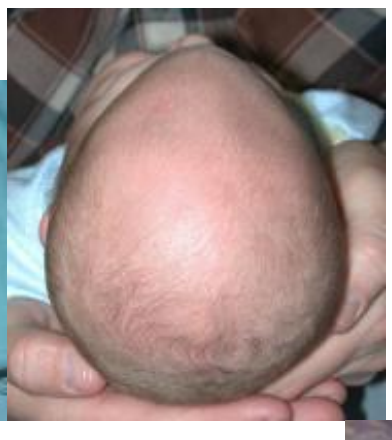
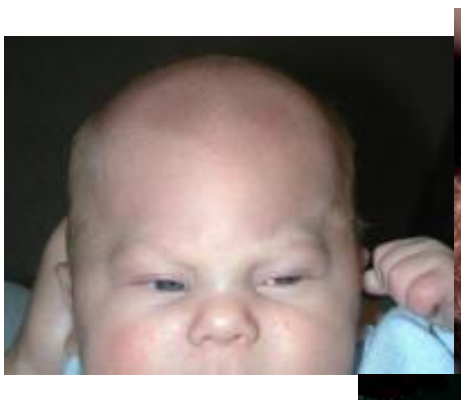
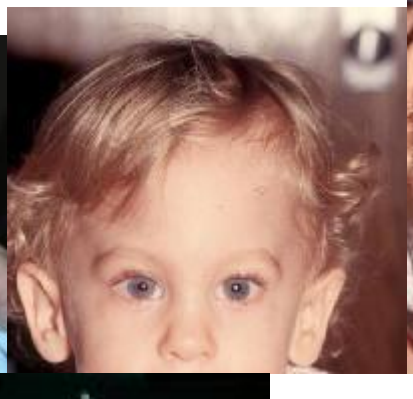
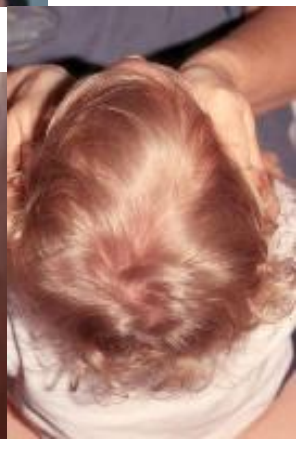
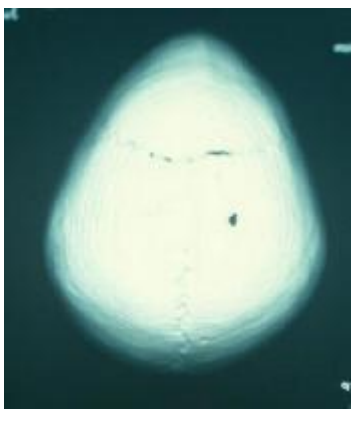
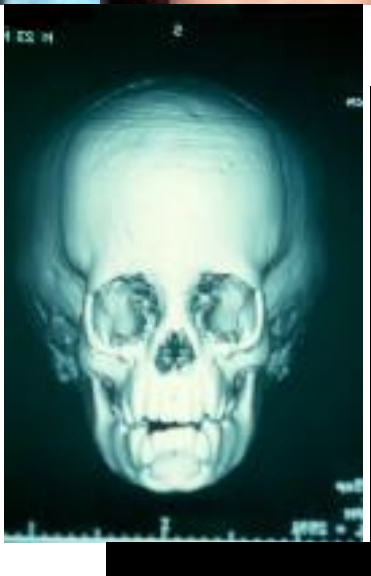
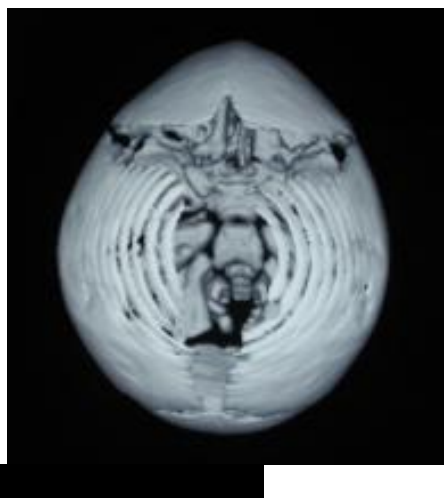
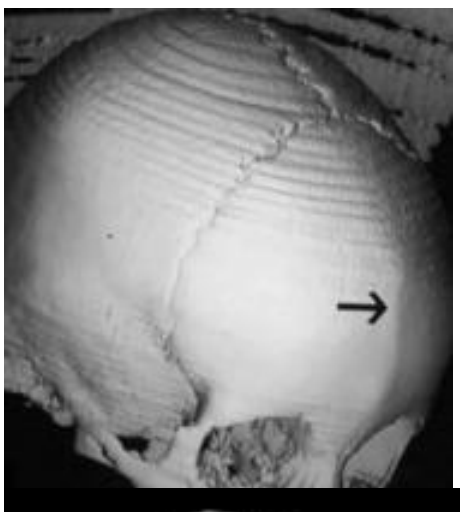
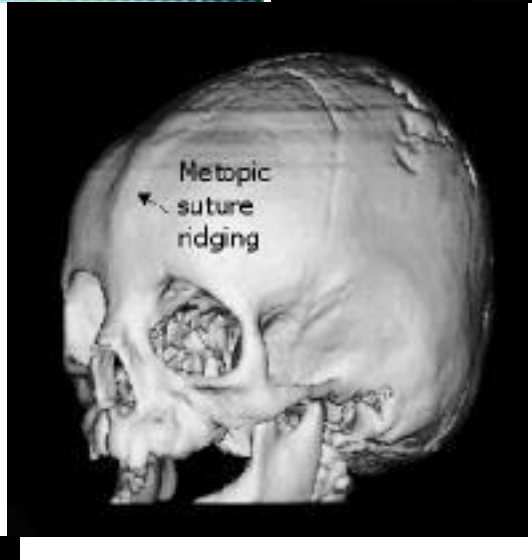


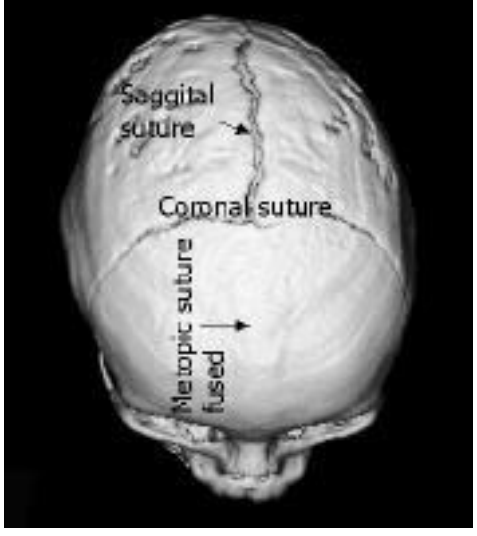
Metopic synostosis
→ trigonocephaly

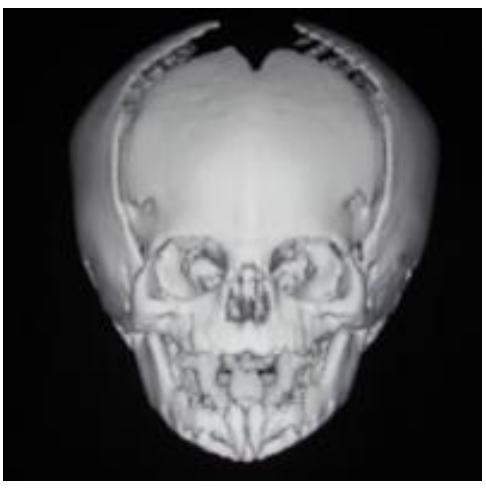
(4-10 % of all cases) - <u>prominent midline frontal ridge</u> (keel-shaped forehead), recessed orbital rims, hypotelorism (indicates early fusion of metopic suture*).

*physiologically, metopic suture is first suture to close (as early as 3rd postnatal month)

- often occurs in syndromic context (e.g. 19 chromosome, Opitz trigonocephaly syndrome) or in conjunction with holoprosencephaly.
- abnormality is usually mild and requires no surgical intervention.

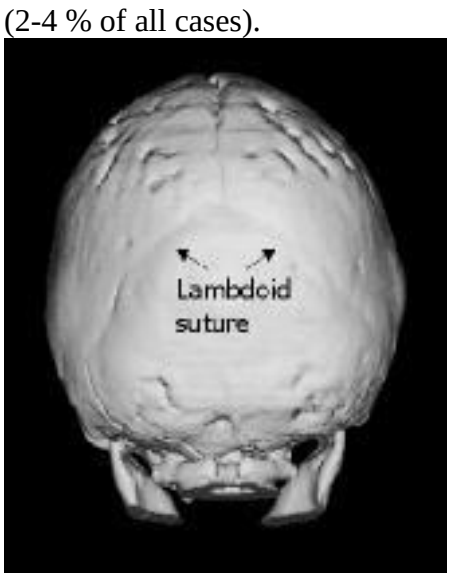















Lambdoid synostosis

(2-4 % of all cases).



Unilambdoid synostosis → posterior plagiocephaly

1) ipsilateral occipital flattening and *enlargement of ipsilateral* MASTOID PROCESS - pathognomonic for lambdoid synostosis!!!
2) compensatory bulge at contralateral parietal eminence
3) in most serious cases, *ear on affected side is displaced* forward and out.

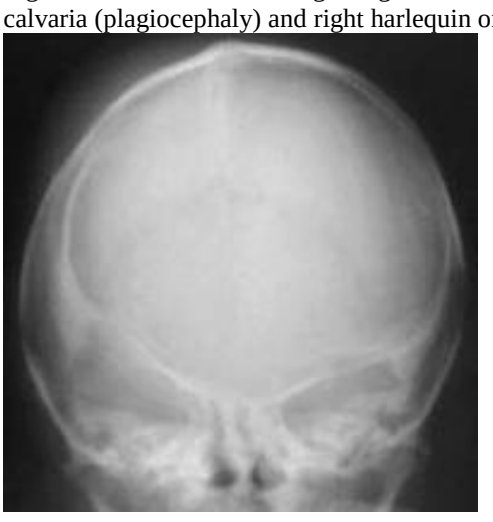
COMBINED-SUTURE SYNOSTOSES

- strongly suggests craniofacial syndrome!

Coronal + sagittal synostosis → oxycephaly

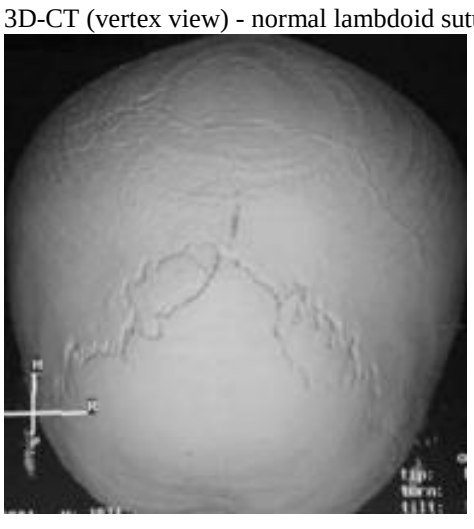
- high, conical head with sharp bossing in region of anterior fontanelle (Gr. *oxys* - sharp).
  - microcephaly with crowding of intracranial contents – elevated ICP!

Sclerotic margins and heaped-up bone of fusing sagittal suture. Note flattening of right side of calvaria (plagiocephaly) and right harlequin orbit:



The same patient: right coronal suture is abnormally straight (*large arrow*) and narrow in appearance, whereas left is normal (*small arrow*):

3D-CT (vertex view) - normal lambdoid suture with complete fusion of sagittal and coronal sutures:



Coronal + sagittal + lambdoid synostosis → triphyllocephaly, s. kleeblattschädel

(cloverleaf, or trilobed, skull) - calvarial bone between sutures is expanded by developing brain but held at area of sutures - bilateral constrictions at sylvian fissures, very prominent temporal bones, bulging forehead, ocular proptosis (resulting from shallow orbits).

N.B. **metopic** and **squamosal** sutures are normal!

- severe neurological impairment:

Most severe craniosynostosis! – urgent surgical repair!

  - shallow orbits, traction & compression in optic canals → proptosis → optic atrophy (22%), legal *blindness* (7%).
  - conductive *hearing loss* (55%).
  - headaches, seizures.
  - mental deficiency is rare.
- occurs in Crouzon's syndrome, thanatophoric dysplasia.

SYNDROMIC SYNOSTOSES

= craniosynostosis + other body deformities.

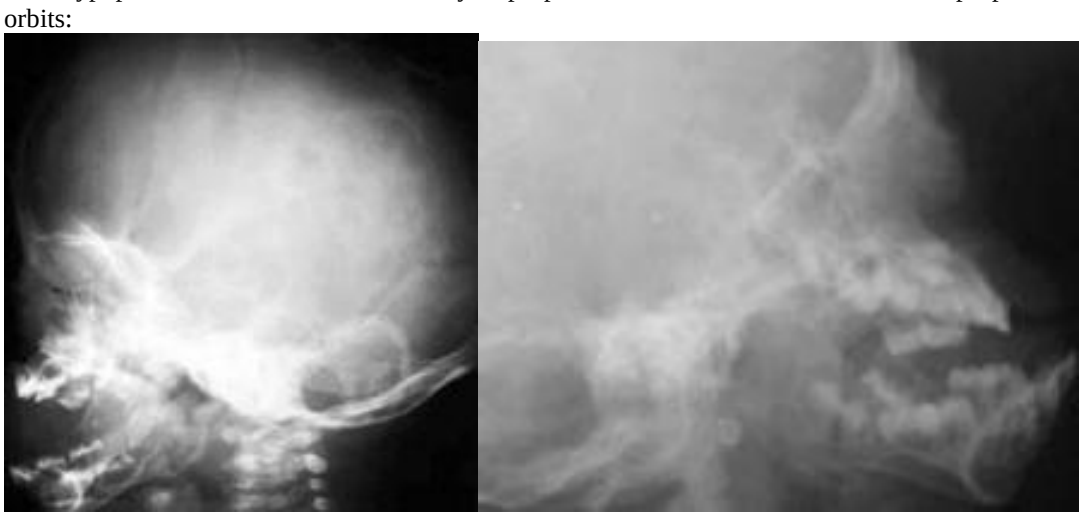
- Crouzon's and Apert's syndromes account for 2/3 of syndromic craniosynostoses cases.
- **midface retrusion** is associated with Apert or Crouzon syndrome.
- all are AUTOSOMAL DOMINANT!

CROUZON'S syndrome (S. CRANIOFACIAL DYSOSTOSIS)

- autosomal dominant **KLEEBLATTSCHÄDEL**

- INCIDENCE - 1 case in 60,000 live births.
- 25 mutations in *FGFR2 gene*; 25% cases sporadic.
- 60% patients have intracranial hypertension.
- normal limbs.

Note hypoplastic maxilla, which is severely disproportionate to normal mandible, severe proptosis due to underdeveloped orbits:



APERT'S syndrome (S. ACROCEPHALOSYNDACTYLY)

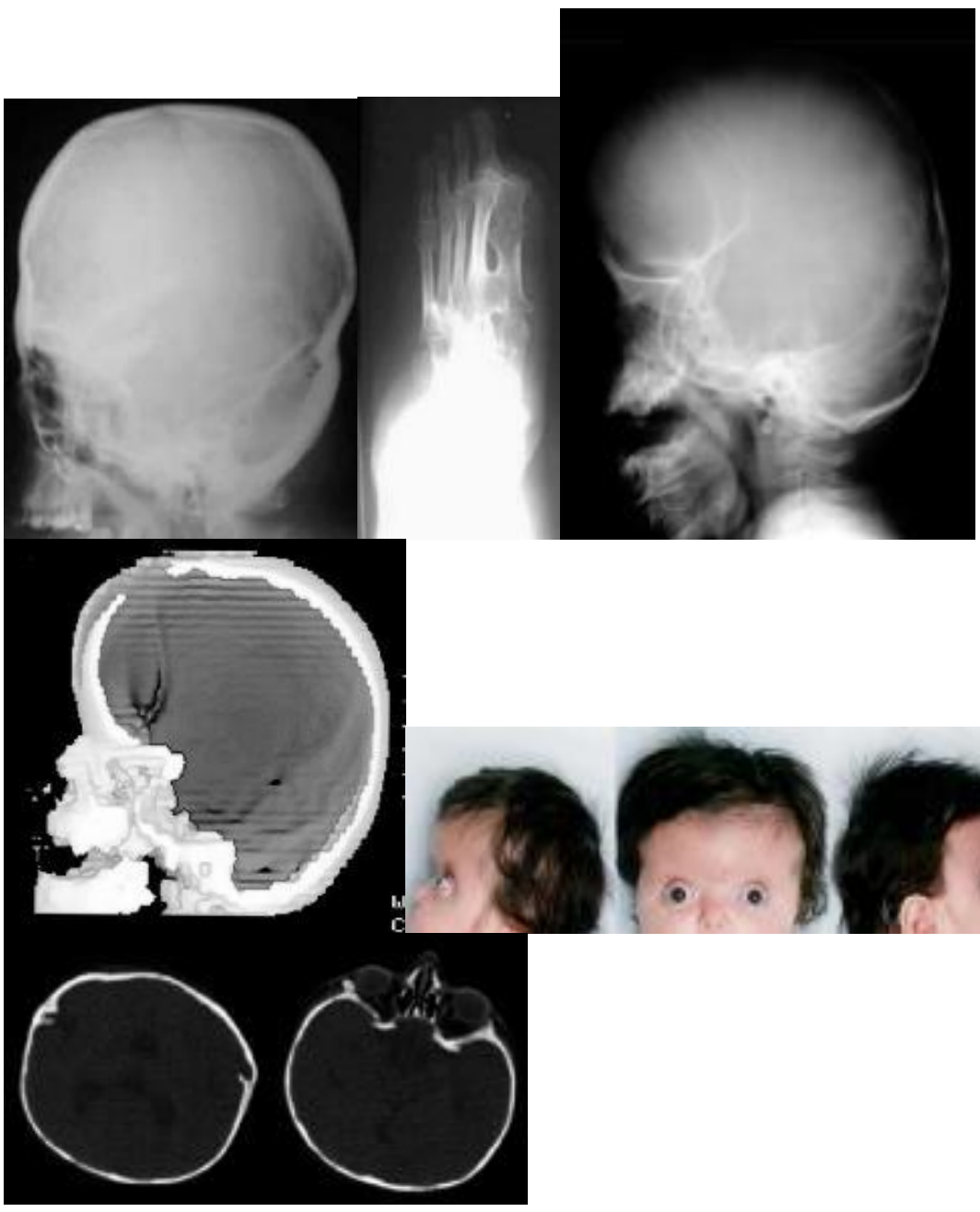
- autosomal dominant or sporadic BRACHYTURRICEPHALY = **BRACHYCEPHALY** + **TURRICEPHALY, S. TURMSCHÄDEL*** = short and high skull; prominent forehead and flat occiput.

*tall round (conical) skull due to malformed short cranial base - premature fusion of **sphenofrontal and frontoethmoidal sutures**; as well as **coronal suture**.

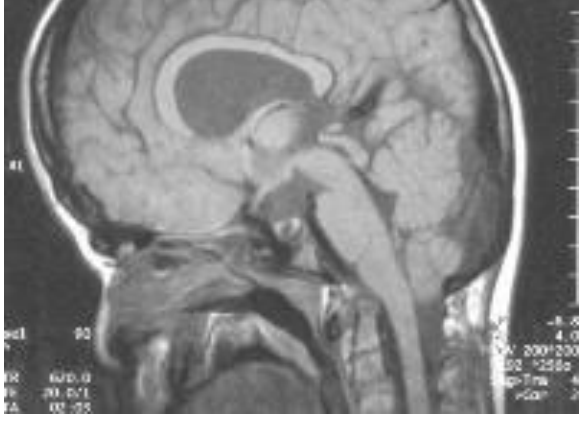
Skull ≈ as in Crouzon; main differences – common hydrocephalus, shortened arms with II-IV digit syndactyly

- INCIDENCE - 1 case in 10,000 live births.
- almost all cases are due to 1 of 2 described mutations of *FGFR2 gene* (chromosome 7); sporadic in 95% cases.
- intracranial volumes tend to be higher than normal! (but 45% patients have intracranial hypertension)
- in infancy, wide and gaping sagittal and metopic sutures.
- frequent platybasia, maxillary hypoplasia, C5-6 vertebrae fusion (68% patients).

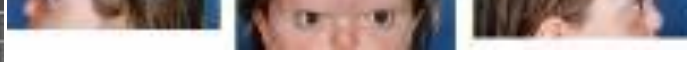
- proptosis of eyes, hypertelorism, downward slanting palpebral fissures, small nose, low-set ears.
- optic atrophy and conductive hearing loss can occur.
- 50% patients have IQ < 70; but normal (or above average) intelligence is not exception.
- CNS can be affected in number of ways (esp. agenesis of corpus callosum, migrational anomalies).
- **acrosyndactyly** - osseous* syndactyly of hands & feet (mitten hands and sock feet) is prominent feature. *progressive calcification and fusion of bones.



abnormal configuration of brain parenchyma; distortion of corpus callosum and ventricular system; posterior fossa is shallow and hindbrain herniation is present:



postoperative photograph of 4-year-old girl (she had fronto-orbital advancement when aged 12 months): orbits are well covered, but ears remain low-set and turricephaly has not changed significantly:



PFEIFFER syndrome

- autosomal dominant or sporadic **coronal** and perhaps **sagittal** suture closure (perhaps TURRICEPHALY).
  - mutations of both *FGFR1* and *FGFR2 genes*.
  - **down slanting of palpebral fissures** is characteristic.
  - hypertelorism, narrow maxilla.
  - broad distal phalanges (esp. of thumb and great toe), **polydactyly**.
  - 3 types:
    - type I (most common) – moderate÷severe hearing loss (auditory canal stenosis or atresia, hypoplasia or enlargement of middle ear cavity).
    - type II - severe proptosis, ankylosis of elbows.
    - type III - ocular proptosis, hydrocephalus, hearing defects, short stature, cervical fusion, cone-shaped epiphysis and hypoplastic bones about elbow.

SAETHRE-CHOTZEN syndrome

- autosomal dominant asymmetric **coronal** suture closure (→ PLAGIOCEPHALY).
  - **bilateral ptosis** is common (usually requires surgical treatment).
  - facial asymmetry, maxillary hypoplasia, shallow orbits, hypertelorism, small ears.
  - shortened fingers + cutaneous syndactyly, short stature.
  - cervical fusion is often seen at C2-3.

CARPENTER syndrome

- probably autosomal recessive synostosis of **coronal** and often **sagittal** and **lambdoid** sutures (→ KLEEBLATTSCHÄDEL).
  - shallow supraorbital ridges, laterally displaced inner canthi.
  - neurosensory and conductive hearing loss; mental retardation is common.
  - brachydactyly & soft-tissue **syndactyly** of hands and feet.
  - hypogenitalism, obesity.

JACKSON-WEISS syndrome

- **coronal** and ***basal skull*** synostosis.
  - mapped to same gene as Crouzon disease.
  - enlarged great toes and craniofacial abnormalities similar to Pfeiffer syndrome but in absence of thumb abnormalities.

AUTLEY-BIXLER syndrome

- probable autosomal recessive **multiple suture** closure.
  - brachycephaly with midfacial hypoplasia, proptosis, choanal stenosis, dysplastic ears.
  - arachnodactyly, joint contractures.

BALLER-GEROLD syndrome

- autosomal recessive one or more suture synostosis (usually **metopic**)
  - mental deficiency.
  - radial hypoplasia, and other preaxial limb anomalies.
  - anal malformation.

OTHER

9p monosomy (deletion of distal portion of short arm chromosome 9) - **metopic** suture closure.

- midfacial hypoplasia, poorly formed ears.
- long middle phalanges of fingers with extra flexion creases, short distal phalanges with short nails.
- cardiac and genitourinary defects.

BEARE-STEVENSON syndrome

MUENKE syndrome

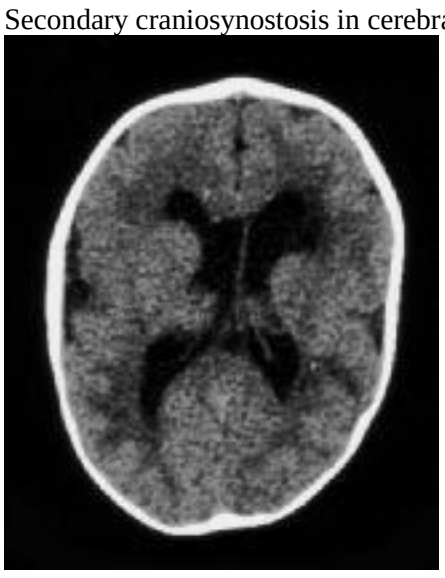
JACKSON-WEISS syndrome

DIFFERENTIAL DIAGNOSIS

SECONDARY CRANIOSYNOSTOSIS

- retarded brain growth / atrophy is primary abnormality, i.e. *secondary craniosynostosis* is frequent with microcephaly (e.g. unilateral destructive brain lesions, microencephaly, shunt placement in hydrocephalus?) - ICP is normal, and surgery seldom is needed.

Secondary craniosynostosis in cerebral atrophy:



POSITIONAL POSTERIOR PLAGIOCEPHALY, "LAZY LAMBDOID", OCCIPITAL PLAGIOCEPHALY

- not progressive flattened posterior part of head; due to *position head takes during sleep**; normal lambdoid sutures; frequently associated with torticollis (may be the cause of specific head position in bed!)

*since American Academy of Pediatrics recommended that infants sleep on their backs to reduce SIDS

N.B. **OCCIPITAL** (not LAMBDOID) to stress that suture is normal!

N.B. *true lambdoid synostosis is rare* (≈ 2% posterior plagiocephaly cases)!
View from above ("bird's-eye view):

POSITIONAL MOLDING:

1) head shape is parallelogram (rhomboid) - skull is pushed ventrally on one side.
2) ear position is more anterior on side of flattening.
3) frontal bossing is ipsilateral.

TRUE CRANIOSYNOSTOSIS:

1) head shape is trapezoid - growth is restricted on side of fused suture.
2) ear position is more posterior on side of flattening.
3) frontal bossing is contralateral (if any).

Treatment (only for severe cases):

1) ***plastic caps*** (***molding helmets***) fitted externally on head and worn 23 h/d until age 1 year (can gradually manipulate shape of skull)
2) ***frequent posture change*** (tummy time↑ esp. when apnea monitors are now available)
3) no surgical treatment!

Note anterior displacement of right occiput and of right frontal region on same side:

TREATMENT

INDICATIONS

1. **Cosmetic** problems (the only consideration in single-suture nonsyndromic synostosis cases!).
2. **Elevated ICP** (in some units, routine measurement of ICP is performed in all syndromic cases).
   N.B. if any restriction of brain growth by skull occurs, it is only in first 6 months of life; after infant is > 6 months, effect of craniosynostosis becomes exhausted (burnt out); i.e. *maximum constrictive effect of craniosynostosis occurs at birth* when difference in intracranial volume between healthy neonates and neonates with craniosynostosis is maximal!
3. **Progressive exophthalmos** threatening eyes.

CONTRAINDICATIONS

- only absolute contraindication is **microcephaly**.

SURGERY TIMING

a) **EARLY SURGERY** - soon after birth (minimized risk of mental impairment due to restricted brain growth; bones grow rapidly and easily cover surgical defects - best cosmetic results, but high risk of recurrent deformity).
b) **LATE SURGERY** - at age 12 months.

> surgery is recommended as soon as child can safely tolerate physiological stress of surgery, usually best is – **6 weeks÷6 months** of age.

Do not operate in patients without raised ICP until considering following:

- infants have large head relative to body size - deformity appears more prominent in young infant and may be less obvious with age.
  N.B. do not operate on mild metopic synostosis (just ridge) – sometimes disappears with time
- as child grows and more hair appears, visible abnormality may decrease.
- if head shape does not improve by age 2-4 months, then abnormality is unlikely to resolve with age.

SURGICAL PROCEDURE

- see p. Dev15 >>

A. **Open surgery** – at age 4-10 months (Dr. Ritter prefers 10 mos – better withstands anesthesia stress, has more Hb, no harm with waiting so long); bone fragments are replaced back and secured with plates.
B. **Minimally invasive surgery** (mostly for single suture synostosis) – at age ≤ 3 months: **(endoscopic) linear craniectomy** (wide excision of fused suture – SUTURECTOMY) → optional separation of bony margins by implanted matrix, optional – barrel stave osteotomies → custom-made **molding helmet** for 6-18 months

Main principle – **OVERCORRECT** (as head grows back to original shape)!!!

Complex forms of craniosynostosis - more complex **cranial expansion & remodeling procedures** (linear craniectomies have been abandoned!)

- *in earlier years*, tendency was for **monoblock facial advancement** (forehead and midface in one osseous block) - now waned in popularity (extensive surgery with considerable morbidity, less than superior results).
- most *modern procedures* constitute variations of **fronto-orbital advancement** (cranial vault remodeling) - ***mobilization*** of ***supraorbital bar*** with series of facial osteotomies → ***advancement and stabilization*** of ***supraorbital bar*** in new more anterior position (results in expansion of floor of anterior fossa and roof of orbits). see p. Dev15 >>
  - with this technique, connection of coronal suture complex with skull base is disrupted.
  - problem often encountered after any type of fronto-orbital advancement is *persistent narrowing in temporal regions* (difficult to correct).

PLAGIOCEPHALY - although only one suture is prematurely fused, in fact, deformity is bilateral because normal side is attempting to compensate - ***bilateral correction*** is usually necessary (i.e. both ipsilateral and contralateral suture lines must be surgically corrected to allow for smooth and symmetric correction).

KLEEBLATTSCHÄDEL - **early subtotal craniectomy** is only reasonable attempt at correction:
a) all sutures are resected, **skull is morcellized → bone fragments replaced** and sutured loosely to dura.
b) alternative surgical approach - **remove all bone**!

Progressive maxillary hypoplasia (midface hypoplasia) → **midface advancement** at 10-15 yrs:
a) **Le Fort III osteotomy and advancement** in one operation.
b) **midface distraction** (patient wears external frame for several weeks) - gaining popularity - better, long-lasting result.

POSTOPERATIVE

- PEDIATRIC INTENSIVE CARE unit for 24 hours.
- ***considerable edema*** may be encountered, but it quickly resolves in following days.
- some restriction in activity to avoid head injury.
- optional CT on 4th postoperative day → discharge.
- routine postoperative follow-ups: 3 weeks, 6 weeks, 3 months, 6 months, and 1 year, with annual visits until age 6 years → every 2-3 years.
- MRI at yearly intervals (in syndromic cases) - to exclude development of hindbrain hernia.
- continue head circumference measurements, watch for signs of raised ICP.
- minor asymmetries are encountered; H: HYDROXYAPATITE paste.
- *brain is slow to expand* - new space is mostly occupied by CSF (± extradural collections).
- children (aged 5-10 years) may develop **recurrent craniostenosis** → repeat operations (≈ 7%)
  - features of recurrent craniostenosis: copper-beaten appearance (localized or generalized), sclerotic hyperdense bands of bone in calvarium.
  - some loss of advancement is normally expected in first few years after operation.

MACROCEPHALY

Head circumference (related to age, sex, and body size):
a) ≥ **2 standard deviations above mean** for age.
b) above **98th percentile** for age.

ETIOLOGY

- disorders in infant ÷ young child (closed sutures in pubertal child prevent skull enlargement!):

1. **Pressure-inducing** disorders (ICP↑, rapidly increasing head circumference):
   1) **progressive hydrocephalus**
   2) **mass lesions** (e.g. chronic subdural collections, tumour, expanding arachnoid cyst)
2. **Syndromes** (normal ICP, head grows at ≈ normal rate; commonly, child is macrosomic):
   1) **mucopolysaccharidoses**, **osteopetrosis**, **achondroplasia**
   2) **syndromes with MEGALENCEPHALY** see p. Dev7 >>
   3) **thickened cranium** (e.g. chronic anemia, rickets, osteogenesis imperfecta, epiphyseal dysplasia).
   4) **fragile X syndrome** (all patients with macrocephaly should be evaluated for mental retardation!)
   5) **trisomy 9p syndrome** - macrocephaly with somatic and genital growth delay; facial, hand & feet deformities, periscapular muscle hypoplasia, delayed bone maturation; severe mental retardation.
   6) **ROBINOW syndrome** - macrocephaly with macroglossia and other facial deformities; hemivertebrae and limb defects; genital hypoplasia; ± seizures, variable degree of mental deficiency.
   7) **GREIG cephalopolysyndactyly** - autosomal dominant macrocephaly, frontal bossing and hypertelorism, broad thumbs.

8) **BESS syndrome (benign enlargement of subarachnoidal space)** - no signs of raised ICP; infants develop normally clinically.
   – head shows initial rapid growth followed by normal rate (larger than normal head growing at normal pace).
   – positive history in one or both parents.
   – **imaging** (ultrasound / CT / MRI) - wider than normal ventricular system, wider than normal subarachnoid spaces (particularly over frontal lobes); brain is otherwise normal - findings compatible either with communicating hydrocephalus (*hydrocephalus ex vacuo*), megalencephaly or atrophy
      N.B. imaging is impossible to interpret without knowledge of previous head growth and circumference measurements, i.e. radiologist is unable to make correct interpretation without knowledge of clinical history.
   – larger than normal intracranial fluid-containing spaces will eventually reduce and become normal in size.

DIAGNOSIS

- **CT / MRI** - mildly dilated lateral ventricles and increase in subarachnoid fluid*.
  *CSF shunts are reserved for progressive enlargement of CSF spaces and evidence of neurologic dysfunction.

MICROCEPHALY

Head circumference (related to age, sex, and body size):
a) ≥ **2-3 standard deviations below mean**.
b) below **5th percentile**.

- very small head circumference implies process that began ***early in embryonic or fetal development***.

ETIOLOGY

Commonest cause is abnormal brain development with subsequent **reduction in brain volume (microencephaly)**!

A. **Secondary (nongenetic) microcephaly** - noxious agents that affect fetus or infant during first 2 yrs of life:
   1. Conditions that ***restrict brain growth***:
      1) craniosynostosis
      2) skeletal dysplasias
      3) external restriction of skull growth in utero
   2. Conditions that ***destroy brain substance*** before completion of brain growth:
      1) hypoxic-ischemic insults
      2) congenital infections (esp. CMV, rubella, toxoplasmosis)
      3) meningitis/encephalitis
      4) drugs & toxins (alcohol, hydantoin)
      5) radiation
      6) endocrinopathies (maternal diabetes mellitus, maternal hyperphenylalaninemia)
      7) hyperthermia
      8) malnutrition (?)

B. **Primary (genetic) microcephaly** - conditions that ***intrinsically impair brain growth***; manifest at birth;
   1. **Microcephalia vera** - autosomal recessive significant microcephaly (up to 5 standard deviations below mean);
      – not associated with other malformations.
      – brain is < 300 g (normal 1200-1500 g).
      – primitive gyral pattern, cortex thickened and disorganized without clear lamination.
      – severe mental retardation (no recognizable speech but relatively preserved personality).
      – may also be less severe autosomal dominant.
   2. Chromosomal syndromes (Down, Edwards, cri-du-chat).
   3. **CORNELIA de LANGE syndrome** - prenatal & postnatal growth delay, synophrys, thin down-turning upper lip, proximally placed thumb.
   4. **RUBINSTEIN-TAYBI syndrome** - beaked nose, downward slanting of palpebral fissures, epicanthic folds, short stature with broad thumbs and toes.
   5. **SMITH-LEMLI-OPITZ syndrome** - ptosis, scaphocephaly, inner-epicanthic folds, anteverted nostrils, low birthweight, marked feeding problems.

- microcephaly is very common in syndromes that have mental retardation and cortical migration abnormalities as component!
- microcephaly is common among mentally retarded population.

DIAGNOSIS

1) **head circumference**; obtain ***at birth***; ***serial measurements*** are more meaningful than single determination; head circumference of *each parent and sibling*.
2) **skull films** (to exclude primary craniosynostosis) → **CT** / **MRI**
3) **karyotype**
4) fasting plasma & urine **amino acid** analysis; serum **ammonium**.
5) **TORCH titers** of mother and child.
6) urine **culture of CMV**.
7) mother's serum **[phenylalanine]**; high serum phenylalanine in asymptomatic mother can produce marked brain damage in otherwise normal nonphenylketonuric infant.

TREATMENT

Most intracranial conditions causing microcephaly are untreatable!
The only treatable cause is craniosynostosis!

CRANIOCERVICAL JUNCTION (SKULL BASE & CERVICAL VERTEBRAE)

Mechanical compression of neuraxis (lower brain stem & cervical cord):
– **nuchal pain & vertigo** may be early nonspecific complaints.
– primary position ***downbeating nystagmus*** (fast component downward) of craniocervical junction origin may give diagnostic lead!
– most important syndrome – **cervical myelopathy**.
– Lhermitte's sign.

N.B. symptoms can be intermittent; symptoms worsen with head movement and Valsalva maneuvers.

BASILAR IMPRESSION, BASILAR INVAGINATION, PLATYBASIA, CONVEXOBASIA

- floor of posterior fossa bulges upward in region about foramen magnum (i.e. skull base flattened on cervical spine) → narrowing of foramen magnum.

1. **PLATYBASIA, S. BASILAR INVAGINATION** (angle* between planes of anterior cranial fossa and clivus > 135-140° on lateral skull X-ray) - generally **asymptomatic**!
   *i.e. angle formed by line connecting anterior margin of foramen magnum, tuberculum sellae, and nasion.
2. **CONVEXOBASIA** (more extreme form).

BASILAR IMPRESSION - upward displacement of occipital bone and cervical spine with *protrusion of odontoid process into foramen magnum* (i.e. odontoid process is above *Chamberlain's line* [hard palate to base of skull]).

ETIOLOGY

A. **Congenital** maldevelopment or hypoplasia of basiocciput:
   1) Chiari types I and II
   2) osteogenesis imperfecta
   3) Hajdu-Cheney syndrome

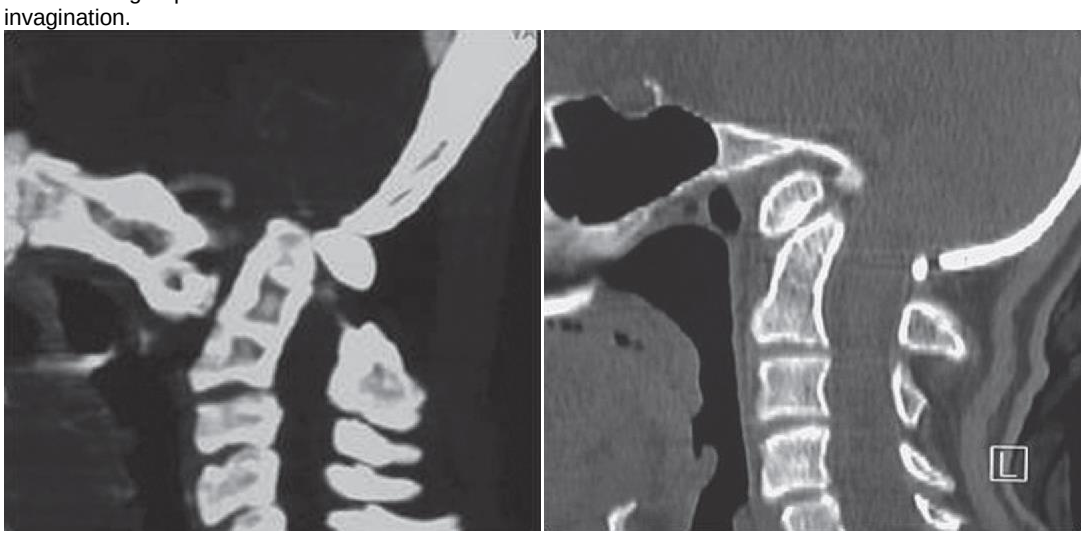
B. **Acquired** softening of skull bones (Paget disease, osteomalacia, RA*). *see also p. 1167 (7a) >>

CLASSIFICATION

- based on a single criterion of the absence or presence of Chiari malformation:
Group I - invagination of the odontoid process into the foramen magnum and indented into the brainstem. The tip of the odontoid process distanced itself from the anterior arch of the atlas or the inferior aspect of the clivus. The distancing of the odontoid process from the anterior arch suggested the presence of instability in the region and atlantoaxial dislocation. The angle of the clivus and the posterior cranial fossa volume were essentially unaffected in these cases.
Group II - the assembly of the odontoid process, anterior arch of the atlas, and the clivus migrated superiorly in unison, resulting in ***reduction of the posterior cranial fossa volume*** → Chiari malformation or herniation of the cerebellar tonsil (a result of reduction in the posterior cranial fossa volume).

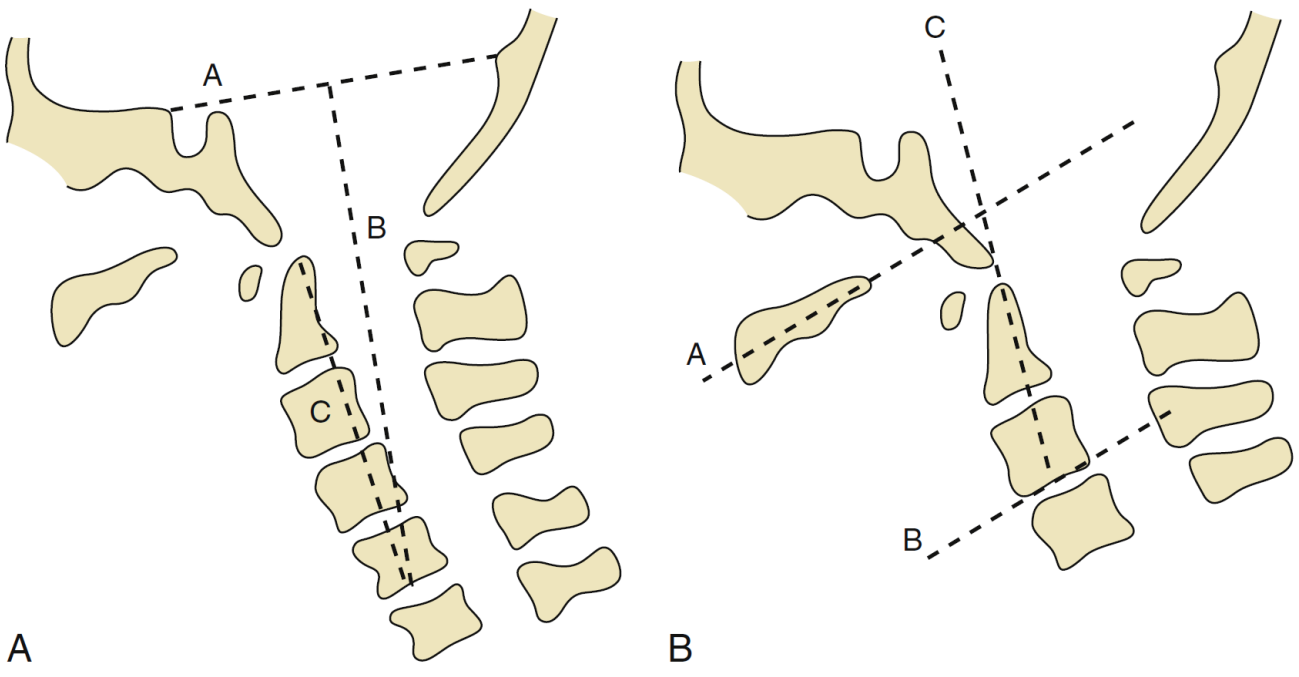
Newer Classification into Groups A and B
In our 2004 study, we identified a subgroup of patients in whom there was clear radiological evidence of instability of the region that was manifested by distancing of the odontoid process away from the anterior arch of the atlas, and the radiological features matching those of group I cases. Considering this current evaluation we have proposed a new classification for basilar invagination into two groups based on parameters that determined an alternative treatment strategy.23,24 In group A basilar invagination there was a "fixed" atlantoaxial dislocation and the tip of the odontoid process "invaginated" into the foramen magnum and was above the Chamberlain line,25 McRae line of the foramen magnum,26 and Wackenheim's clival line.27 The definition of basilar invagination of prolapse of the cervical spine into the base of the skull, as suggested by von Torklus,18 was suitable for this group of patients (Fig. 29.7A). In group B basilar invagination the odontoid process and clivus remained anatomically aligned despite the presence of basilar invagination and other associated anomalies. In this group, the tip of the odontoid process was above the Chamberlain line but below the McRae and Wackenheim's lines (Fig. 29.7B). The radiological findings suggested that the odontoid process in group A patients resulted in direct compression of the brainstem. Essentially, in group A basilar invagination, the pathogenesis appeared to be mechanical instability of the region that was manifested by the tip of the odontoid process distancing itself from the anterior arch of the atlas or the lower end of the clivus. In some group A patients there was Chiari malformation, and this feature differentiates the present classification from the earlier classification. In this group, the atlantoaxial joints were "active" and their orientation was oblique, as shown in Figure 29.8A, instead of the normally found horizontal orientation. Similarities of such a position of the C1-C2 facets with spondylolisthesis in the subaxial spine can be seen. It appears that the atlantoaxial joint in such cases is in an abnormal position and progressive worsening of the dislocation is probably secondary to increasing "slippage" of the facets of the atlas over the facets of the axis. In group B, the pathogenesis appeared to be congenital dysgenesis, and atlantoaxial joints were normally aligned or were entirely fused. The pathogenesis of basilar invagination appears to be different in the two groups. Understanding of these two types of basilar invaginations is crucial in understanding the various management issues involved.
Vertical mobile and reducible atlantoaxial dislocation occurs when there was basilar invagination when the neck was flexed, but the alignment was normal when the head was in an extended position.28 Although such mobility is only rarely identified, it does indicate the need for dynamic flexionextension studies to preoperatively assess the craniovertebral instability. Vertical dislocation is due to incompetence of the atlantoaxial joint and lateral masses.

FIGURE 29.7 A, Computed tomography (CT) scan shows group A basilar invagination. **B,** CT scan shows group B basilar invagination.



(Ellenbogen 2012)

FIGURE 29.8 A, Drawing of the Goel modified omega angle. A line (*A*) is drawn along the hard palate. Another line is drawn that is parallel to line *A* that travels through the midpoint of the base of the C3 vertebral body. The angle of the odontoid process to this line is the omega angle. **B,** Drawing of the parameters used to measure the occipitocervical length (*B*) and the neck length (*C*). Line *A* is the tuberculum sella–torcula line.



(Ellenbogen 2012)

CLINICAL FEATURES

- short neck, low hairline, web-shaped neck muscles, torticollis, reduction in the range of neck movements, vertical head diameter↓
- degeneration of the atlantoaxial joint and primary or secondary destruction of the facets of atlas or axis can lead to superior and posterior migration of the odontoid process and result in basilar invagination and fixed or irreducible atlantoaxial dislocation; such involvement of the lateral masses can result in laxity of the posterior atlantoaxial or retro-odontoid ligaments that result in ***retro-odontoid pannus*** or "tumor-like" or "osteophyte-like" formation.
- progressive slippage of the atlas over the axis, a process similar to spondylolisthesis in the lumbosacral spine, results in invagination of the odontoid process into the craniocervical cord → compression of pons / medulla / cerebellum / cervical cord, stretching of cranial nerves and blood supply - symptoms begin insidiously in childhood or early adult life:
    1) nuchal pain, neck stiffness, torticollis.
    2) spasticity in lower extremities
    3) proprioception loss in upper extremities.
    4) ataxic gait, downbeating nystagmus
    5) vertebral artery obstruction may be significant.
    6) brainstem & lower CN dysfunction late in course (incl. sleep apnea, dysarthria and dysphagia*).

    *in RA patients, it also can be caused by laryngeal arthritis
- if deformity interferes with CSF circulation → ICP↑; subarachnoid block (partial or complete) is present at lumbar puncture in most cases.

From Ellenbogen 2012:
Diagnosis and Clinical Features The majority of patients with group A basilar invagination (58%) had a history of minor to major head injury prior to the onset of the symptoms. The pyramidal symptoms formed a dominant component. Kinesthetic sensations were affected in 55% of these patients. Spinothalamic dysfunction was less frequent (36%). Neck pain was a major presenting symptom in 77% of cases. Torticollis was present in 41% of cases.23 The analysis of radiological and clinical features suggests that the symptoms and signs were a result of brainstem compression by the odontoid process. The presentation was relatively acute in group A cases but it was long-standing and slowly progressive in group B cases. In group B cases, the onset of symptoms and their evolution were insidious. Precipitating Factors Trauma of varying severity was a noteworthy precipitating factor in group A cases.29,30 Trauma seldom plays any major role in precipitating the symptoms in group B cases. The fact that trauma influenced the acute development of symptoms pointed toward an element of instability of the craniovertebral region in group A patients. Associated Clinical Features Mere inspection of the patients with basilar invagination was of diagnostic value in the majority of cases in both the groups. However, short neck and torticollis were more frequently encountered in cases with group A basilar invagination. The symptoms and signs in group B basilar invagination appeared to be directly related to the "crowding" of neural structures at the foramen magnum. Although the dimensions of the foramen magnum were large and sometimes larger than even in a normal state, the volume of its contents and probably the "pulsatile" compression of the structures at the foramen magnum resulted in neurological symptoms.31 The markedly reduced girth of the brainstem in group A cases clearly showed that direct compression of the brainstem by the odontoid process caused the neurological symptoms. Central cord symptoms and related signs were noted in cases associated with syringomyelia.

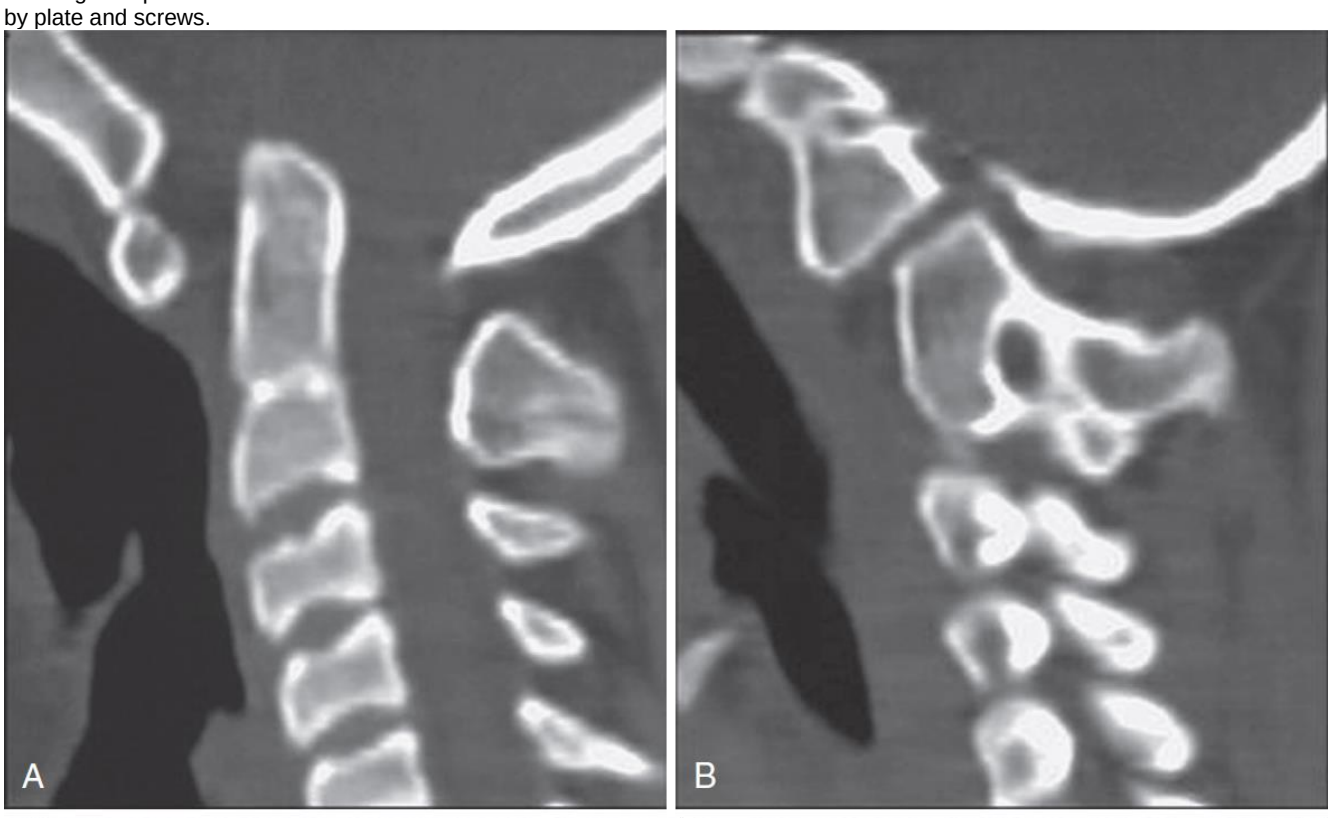
DIAGNOSIS

- **skull X-ray + sagittal CT + sagittal MRI**.

From Ellenbogen 2012:
Radiological Criteria The Chamberlain Line Basilar invagination was diagnosed when the tip of the odontoid process was at least 2 mm above the Chamberlain line.25 Measurement of the Chamberlain line on lateral sagittal reconstruction pictures of CT scan and sagittal MRI were seen to be reliable and accurate. The analysis of basilar invagination in the two groups on the basis of the Chamberlain line suggested that the basilar invagination is much more severe in group B than in group A. Distance Between the Odontoid Tip to the Pontomedullary Junction The distance of the tip of the odontoid from the pontomedullary junction, as observed on MRI, is a useful index to define the reduction of the posterior cranial fossa bone size.25 The distance was markedly reduced in group B cases but it was relatively large in group A cases. Atlantodental or Clivodental Interval In group A cases, it was seen that the odontoid process migrated superiorly and posteriorly into the foramen magnum and distanced itself from the anterior arch of the atlas and the inferior end of the clivus. As judged from the atlantodental or clivodental interval, there was an element of "fixed" atlantoaxial dislocation in these cases. Actual mobility of the atlantoaxial joint on flexion and extension of the neck can be demonstrated only rarely. In group B the alignment of the odontoid process with the anterior arch of the atlas and with the inferior aspect of the clivus remains normal and there is no instability. Wackenheim's Clival Line Wackenheim's clival line is a line drawn along the clivus. The tip of the odontoid process was significantly superior to Wackenheim's clival line in group A cases. In group B cases, the relationship of the tip of the odontoid process and the lower end of the clivus and the atlantodental and clivodental interval remained relatively normal. In a majority of these cases, the tip of the odontoid process remained below Wackenheim's clival line4,27 and McRae's line of the foramen magnum. 26 The basilar invagination thus resulted from the rostral positioning of the plane of the foramen magnum in relation to the brainstem. Platybasia A line is drawn along the anterior skull base. The angle of this line to the clivus is referred to as the "basal angle." Reduction of the basal angle is referred to as "platybasia." Platybasia does not directly result in any neurological symptoms, but it participates with basilar invagination in critically reducing the posterior cranial fossa volume. Posterior Cranial Fossa Volume The Klaus' height index,32 measured on MRI, was seen to be much more accurate than the conventional measurements based on plain radiographs. The tentorium could be clearly identified on MRI and the distance of the tip of the odontoid from the line of the tentorium indicated the height of the posterior cranial fossa. On the basis of Klaus' index, the posterior fossa height was found to be markedly reduced in group B cases but it was only moderately affected in group A cases. Omega Angle Although not frequently used, the omega angle, or the angulation of the odontoid process from the vertical as described by Klaus, was found to be a useful guide.32 Goel described a modified omega angle as the measurement of the angle from the vertical and noted it was affected by the flexion and extension of the neck.22 A line was drawn traversing through the center of

the base of the axis parallel to the line of the hard palate. The line of the hard palate was unaffected by the relative movement of the head and the cervical spine during the movement of the neck in these "fixed" craniovertebral anomalies. Facial hypoplasia or hard palate abnormality was not seen in any case in this series and did not affect the measurements. The omega angle depicted the direction of displacement of the odontoid process. The omega angle was severely reduced in group A cases but it was much larger in group B cases. The reduction in the omega angle in group A cases depicted that the odontoid process had tilted toward the horizontal and was posteriorly angulated in group A cases, but it was near vertical and superiorly migrated in group B cases (Fig. 29.9). Brainstem Girth The effective brainstem girth measured on MRI is a useful additional parameter.22 Although the brainstem girth is markedly reduced in group A cases, the girth is only marginally affected or unaffected in group B cases, indicating thereby that there is no direct brainstem compression as a result of the odontoid process in the latter group. Occipitalization of the Atlas Occipitalization of the atlas associated with basilar invagination was noted first by Rakitansky (cited by Grawitz 1880)19 and has since been referred to frequently.19,25,26,33,34 Many authors have regarded assimilation as a characteristic feature of basilar invagination. The assimilation of the atlas can be partial or incomplete. Neck Size Measurement of craniovertebral height can be done using a modification of Klaus' posterior fossa height index.13,22,32 The cervical height was measured from the tip of the odontoid process to the midpoint of the base of the C7 vertebral body (see Fig. 29.9). Direct physical measurement of the neck length can be a useful parameter. The parameter of direct physical measurement of the neck length from the inion to the tip of the C7 spinous process can be useful.13,35 Cervical lordosis is evaluated with a modification of the Klaus omega angle13,32 and a modified omega angle.

FIGURE 29.9 **A,** Computed tomography (CT) showing group A basilar invagination. **B,** Sagittal cut through the atlantoaxial facet joints. The angulation of the joint and the listhesis of C1 over C2 can be appreciated. **C,** Postoperative CT scan showing reduction of basilar invagination. **D,** Sagittal cut through the atlantoaxial joint, showing the spacer and fixation by plate and screws.



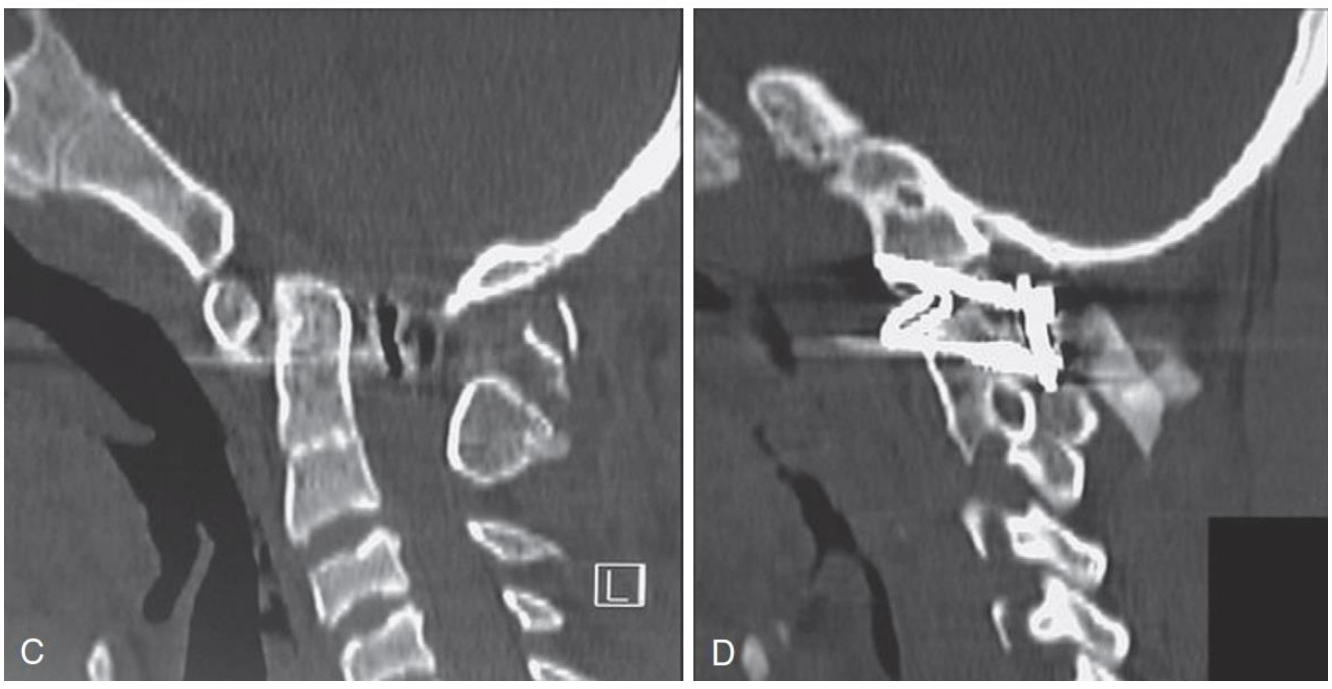
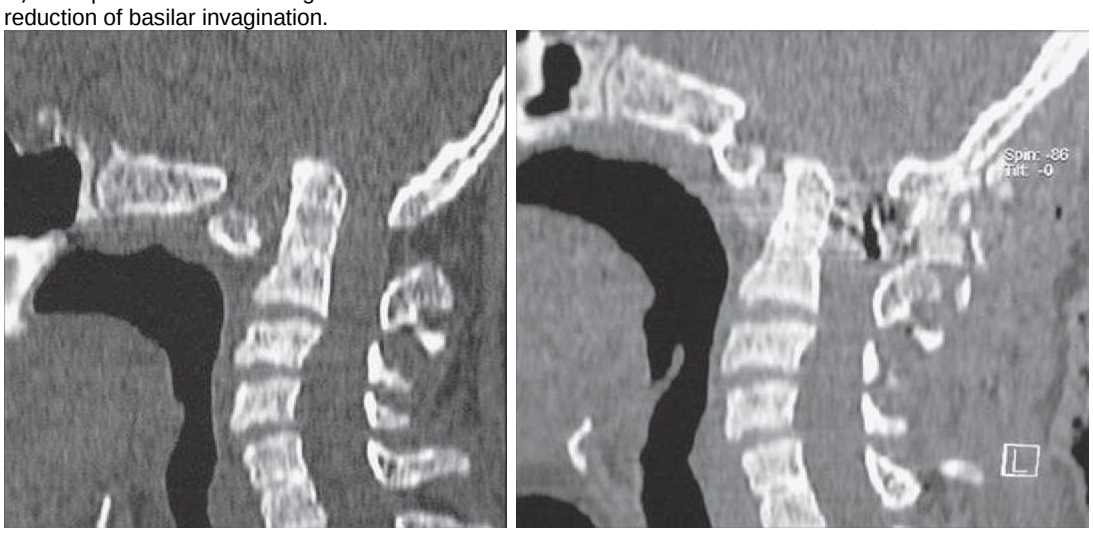


FIGURE 29.10 **A,** Computed tomography (CT) scan showing group A basilar invagination. **B,** Postoperative scan showing reduction of basilar invagination.



TREATMENT

Preoperative* **halo traction** (to reduce vertical instability + C2 compression neuralgia) for 2-5 days with traction force 5 → 25 lbs. *awake patient reports if something is going wrong

↓ then

Surgical decompression at foramen magnum ± **C2-3 laminectomies** with **cervico-occipital fusion** (s. occipitocervical fixation).

Indications for surgery – **compression of neural structures** (myelopathy, neuropathies)

- **suboccipital neuralgia** (C2 entrapment between occiput and posterior arch) as sole indication is controversial (but justified if traction eliminates pain – confirms C2 entrapment)

From Ellenbogen 2012:

Surgical Management Craniovertebral Realignment for Group A Basilar Invagination The conventional form of treatment of group A basilar invagination is a transoral decompression22,23,36 that is followed by posterior occipitocervical fixation. However, the long-term clinical outcome following the twin operation of transoral decompression followed by posterior stabilization was seen

to be inferior to the clinical outcome following surgery that involves craniovertebral realignment without any bone, dural, or neural decompression. An attempt can be made to reduce basilar invagination by performing occipitocervical fixation following institution of cervical traction.22,36 However, all our cases treated in this manner subsequently needed transoral decompression as the reduction of the basilar invagination and of atlantoaxial dislocation could not be sustained by the implant. The technique of craniovertebral realignment by wide removal of the atlantoaxial joint capsule and articular cartilage by drilling and subsequent distraction of the joint by manual manipulation provides a unique opportunity to obtain reduction of the basilar invagination and of atlantoaxial dislocation. Technique of Craniovertebral Realignment This operation is suitable for patients with group A basilar invagination. The exposure of the atlantoaxial joint in cases with basilar invagination is significantly more difficult and technically challenging when compared to a normally aligned atlantoaxial joint encountered during the treatment of posttraumatic instability. The joint is rostral in location and the microscope needs to be appropriately angled. The atlantoaxial facet joints are widely exposed on both sides after sectioning of the large C2 ganglion. The joint capsule is excised and the articular cartilage is widely removed using a microdrill. The joints on both sides are distracted using an osteotome. The flat FIGURE 29.9 A, Computed tomography (CT) showing group A basilar invagination. B, Sagittal cut through the atlantoaxial facet joints. The angulation of the joint and the listhesis of C1 over C2 can be appreciated. C, Postoperative CT scan showing reduction of basilar invagination. D, Sagittal cut through the atlantoaxial joint, showing the spacer and fixation by plate and screws.
edge of the osteotome is introduced into the joint and it is then turned vertical to effect distraction. The status of the dislocation and of basilar invagination is evaluated by intraoperative radiographic control. Corticocancellous bone graft harvested from the iliac crest is stuffed into the joint in small pieces. Specially designed titanium spacers are used in selected cases as strut graft and impacted into the joints to provide additional distraction and stability. Subsequent fixation of the joint with the help of interarticular screws and a metal plate provides a biomechanically firm fixation and sustained distraction. Holes in the titanium metal spacer provide space for bone fusion. The fixation is seen to be strong enough to sustain the vertical, transverse, and rotatory strains of the most mobile region of the spine. Postoperatively the traction is discontinued and the patient is placed in a four-post hard cervical collar for 3 months during which all physical activities involving the neck are restrained (Figs. 29.8 to 29.10). Reversibility of Musculoskeletal Changes Following Surgery13 A number of bone and soft tissue anomalies are associated with basilar invagination. These include short neck, torticollis, platybasia, cervical vertebral body fusion (Klippel-Feil abnormality) including assimilation of atlas, spondylotic spinal changes, and restriction of neck movements. A number of these abnormalities were seen to be reversible following decompression and stabilization of the region. Considering that several physical features associated with this group of basilar invagination are reversible, it appears that the pathogenesis in such cases may be more due to mechanical factors rather than to congenital causes or embryological dysgenesis. The common teaching on the subject is that the short neck and torticollis are a result of embryological dysgenesis and effectively result in indentation of the odontoid process into the cervicomedullary cord. However, it appears that it is the cord compression due to indentation by the odontoid process that is the primary event and all the physical alterations and bony abnormalities, including the short neck and torticollis, are secondary natural protective responses that aim to reduce the stretch of the cord over the indenting odontoid process. Pain, restriction of neck movements, and hyperlordosis of the neck indicate the presence of instability of the craniovertebral junction. All of these natural responses probably allow the cord a relatively stretch-free traversal over the indenting odontoid process. Reduction of the disk spaces, osteophyte formation, incomplete and complete cervical fusions, and alterations in the craniospinal and cervical angulations appear to be directly related to the reduction in neck length. The reduction in the disk-space height and fusions are more prominently seen in the upper cervical vertebrae. It appears that cervical fusions and assimilation of the atlas may be related to long-standing and progressive reduction in the disk-space height. Foramen Magnum Decompression for Group B Patients Foramen magnum bony decompression appears to be ideal for group B basilar invagination. The procedure resulted in amelioration of symptoms and at least an arrest in the progression of the disability. Driesen (1960) reported that during operations for craniovertebral anomalies, he often had to remove noticeably thickened pieces of bone from the posterior edge of the foramen magnum.30 In our cases, the suboccipital bone and posterior rim of the foramen magnum and the dura overlying the herniated cerebellar tissue were thin in a significant number of cases. This probably was related to the chronic pressure changes secondary to the reduced posterior cranial fossa volume. The bulbous lipping of the posterior rim of the foramen magnum represents the rudiments of the posterior arch of the atlas assimilated into the occipital bone.16 Various authors have recommended that to achieve maximal decompression, it is necessary to open the dura mater and to cut all constrictive dural and arachnoidal bands. Some authors have recommended leaving the dura open while others have recommended the placement of a graft. Current papers do not recommend resection of the herniating tonsils37 or even sectioning of adhesions around them. The fact that dural opening was not necessary while performing posterior fossa or foramen magnum decompression was first described by Goel and co-workers in 1997.22 This finding was based on the understanding that the dura is an expansile structure and cannot be a compressive factor. 22,38 Opening of the dura is not only unnecessary but also subjects the patient to an increased risk of cerebrospinal fluid fistula. It makes an otherwise simple surgery into a relatively complex and dangerous surgical maneuver.

ATLANTOAXIAL INSTABILITY

- anterior arch of atlas - dens interval > 3 mm.

- chronic subluxation → remodelling of lateral atlanto-axial joints - enhances instability and prevents reduction.
- 70% cases are associated with **os odontoideum** >>, most of remainder with **cranial assimilation of atlas** >>

ETIOLOGY

- weakness or absence of structures maintaining stability (e.g. ligament laxity):

1. **Rheumatoid arthritis**!!! see also p. 1167 (7a) >>
2. **Vertebral metastases, trauma**
3. **Mucopolysaccharidoses** (e.g. type IV - Morquio-Brailsford disease), osteogenesis imperfecta, metatrophic dwarfism, spondyloepiphyseal dysplasia, multiple epiphyseal dysplasia, pseudo-achondroplasia, chondrodysplasia calcificans.
4. **Genetic conditions** (e.g. 30% Down's syndrome patients*).
   *screen all trisomy 21 patients at 5-8, 10-12, and 18 years for atlantoaxial instability!

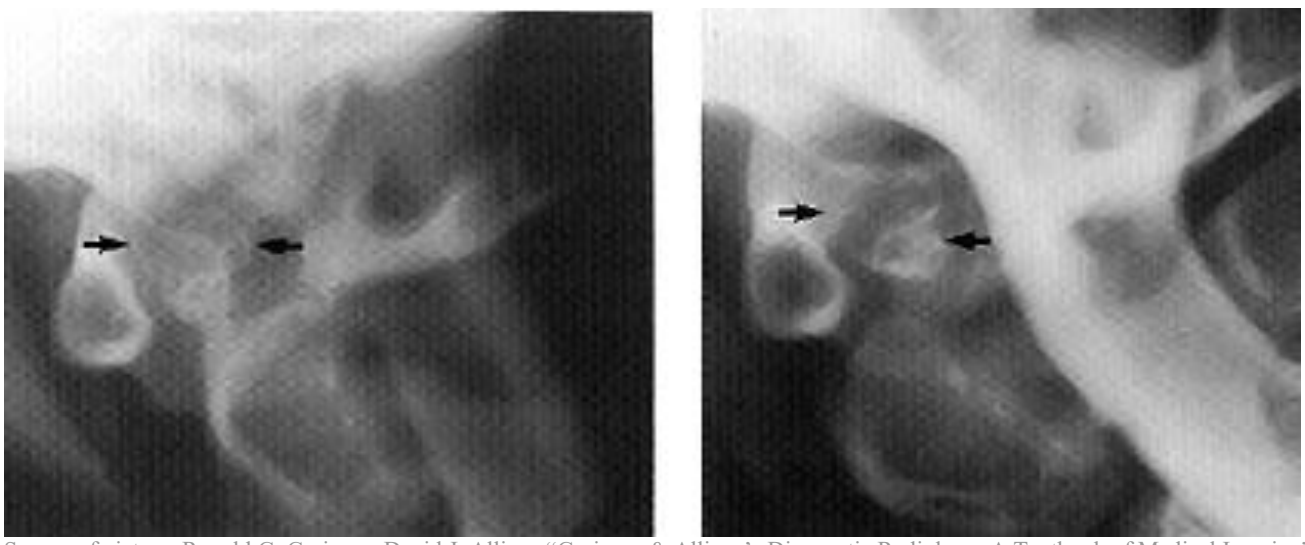
CLINICAL FEATURES

- **atlantoaxial subluxation** (displacement of atlas anteriorly in relation to axis) → acute or chronic **spinal cord compression** between dens (mainly thickened ligaments) and posterior rim of foramen magnum.
  - head movement causes neck pain.
  - children may show head tilt.
  - dislocation can cause immediate death from respiratory failure!

DIAGNOSIS

- **lateral neck roentgenograms** in neutral, flexion, and extension positions - anterior dislocation of atlas.

Os odontoideum (*arrows*) and **atlanto-axial instability** (cervical myelogram, lateral projections): posterior atlanto-axial subluxation in extension (B) is reduced in flexion (A).

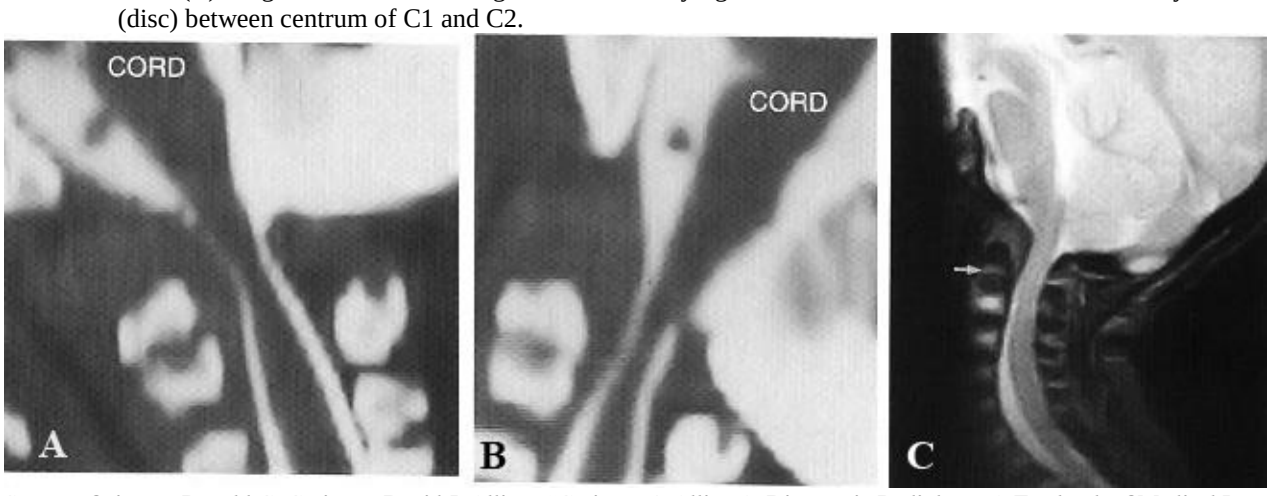


Source of picture: Ronald G. Grainger, David J. Allison "Grainger & Allison's Diagnostic Radiology: A Textbook of Medical Imaging", 4th ed. (2001); Churchill Livingstone, Inc.; ISBN-13: 978-0443064326 >>

Morquio-Brailsford disease (type IV mucopolysaccharidosis):

Case 1 - sagittal CT myelography with neck flexed (A) and extended (B); cephalic two-thirds of dens are not ossified; hypermobility in extension (spinal cord is compressed, mainly by thickened ligaments).

Case 2 (C) - sagittal T2-MRI: cartilaginous dens overlying ossified centrum of C1; arrow indicates synchondrosis (disc) between centrum of C1 and C2.



Source of picture: Ronald G. Grainger, David J. Allison "Grainger & Allison's Diagnostic Radiology: A Textbook of Medical Imaging", 4th ed. (2001); Churchill Livingstone, Inc.; ISBN-13: 978-0443064326 >>

TREATMENT

- restrict potentially harmful physical activities (tumbling, diving, football).
- indications for surgery: instability, myelopathy.
- **external immobilization** alone is unlikely to achieve permanent reduction → **surgical arthrodesis** (posterior atlanto-axial internal-fixation and fusion).
  - all unstable levels must be fused.
  - instrumentation provides immediate stability until bony fusion develops.
- ***Gallie's fusion technique*** – becomes substandard of care because of high rate of nonunion.

OS ODONTOIDEUM

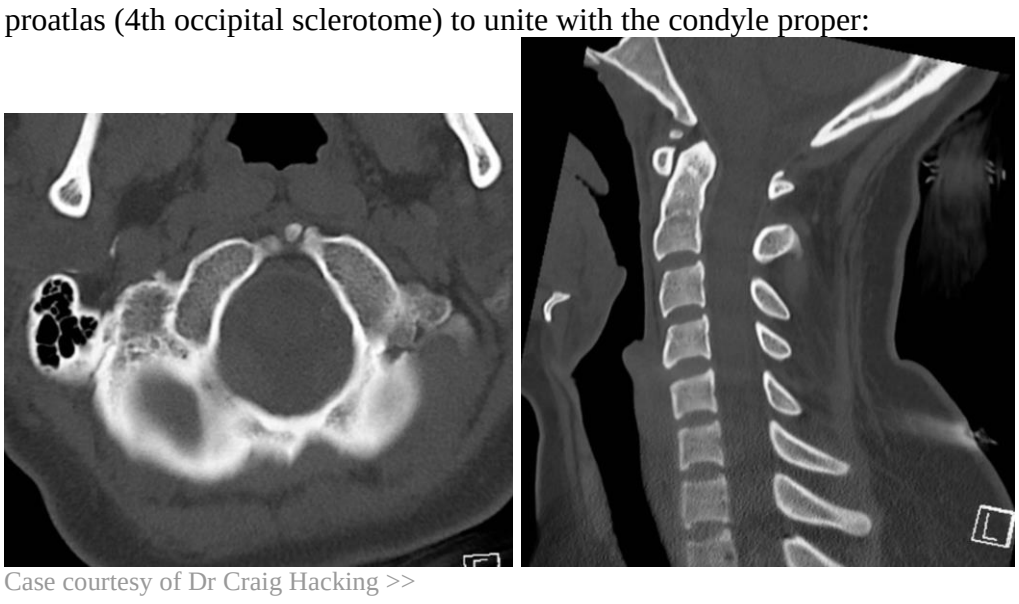
- anatomic variant of the odontoid process - abnormal dens ossification (dens center of ossification not fused with body of axis) secondary to instability and abnormal local stress (i.e. not segmentation anomaly):

- may actually represent an unremembered and/or unrecognized dens fracture through the growth plate before the age of 5-6.

Associated syndromes:

1) Morquio syndrome
2) multiple epiphyseal dysplasia

- it may be seen in association with another adjacent anatomic variant, the third condyle - small separate ossicle at the anteromedial margin of the occipital condyle formed by the failure of the embryonic proatlas (4th occipital sclerotome) to unite with the condyle proper:



Case courtesy of Dr Craig Hacking >>

CLINICAL FEATURES

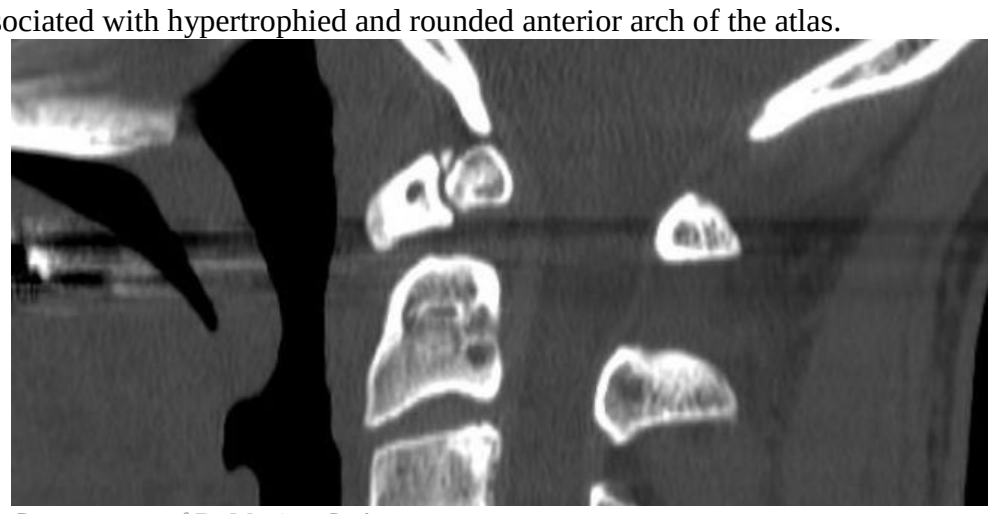
- can be associated with atlantoaxial instability and chronic symptoms.
- level of mobility is below the transverse band of the cruciform ligament and therefore results in abnormal mobility of the dens with respect to C2.

SUBTYPES

orthotopic: ***normal position*** with a wide gap between C2 and os odontoideum.
dystopic s. "os avis": ***displaced***

RADIOGRAPHIC FEATURES

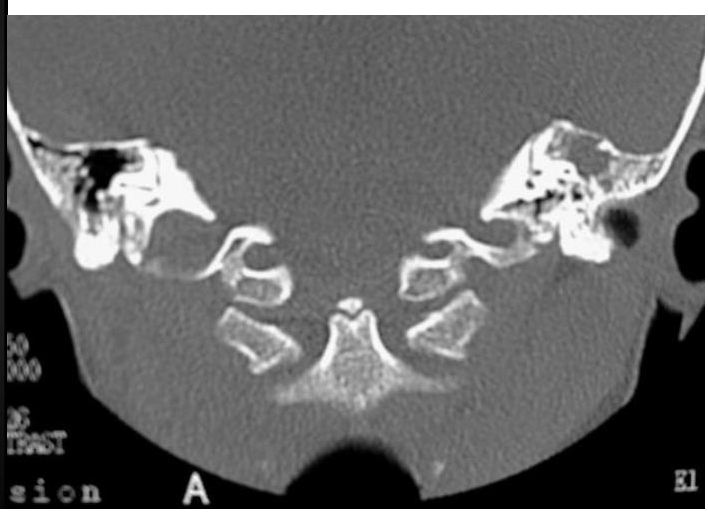
- smooth, well-corticated ossicle.
- around half the size of a normal dens.
- associated with hypertrophied and rounded anterior arch of the atlas.



Case courtesy of Dr Maxime St-Amant >>

DIFFERENTIAL DIAGNOSIS

1) type 2 odontoid fracture
2) ossiculum terminale:



TREATMENT

- Os odontoideum is one of the skeletal "don't touch" lesions.

OCCIPITALIZATION of ATLAS (s. ASSIMILATION of ATLAS)

- fusion of anterior arch and ring of C1 with foramen magnum.

ETIOLOGY

1) Chiari type I malformation
2) Klippel-Feil anomaly
3) achondroplasia.

CLINICAL FEATURES

- cervical myelopathy when AP diameter of foramen magnum (behind odontoid process) decreases to < 19 mm.

DENS HYPOPLASIA

- usually ***misdiagnosis*** - os odontoideum in fact being present but overlooked because it is not ossified, small or malplaced.
- TRUE DENS HYPOPLASIA only occurs ***in association with more complex fusion anomalies*** (esp. those which restrict rotation at $C_{1/2}$).

KLIPPEL-FEIL ANOMALY

- congenital fusion of cervical vertebrae into one or more separate masses:
    a) failure of segmentation (most likely)
    b) secondary fusion.
- fusion is most commonly restricted to C2-3* or C5-6 (but can extend beyond cervical spine – esp. upper thoracic vertebrae).

*frequently associated with occipital assimilation of atlas.

- *sum in height of congenitally fused bodies* is equal to normal height of two vertebrae plus expected height of intervertebral disc if one were present (vs. fusion due to disease – height is less).
- bony structure of fused vertebra is normal except for fusion.
- in cases of PARTIAL fusion, it is anterior aspect that fuses, while rudiment of disc remains in posterior portion.

ETIOLOGY

1) **part of syndromes** (e.g. Turner's, Noonan's, Wildervanck's)
2) **isolated**:
    a) sporadic
    b) inherited (autosomal dominant or autosomal recessive).

CLINICAL FEATURES

- fusion of vertebrae in itself is not of any great clinical importance:
    1) **short neck**
    2) **low posterior hairline**
    3) **limitation of neck movements** (esp. rotation and bending to sides).
- accentuation of symptoms in presence of cervical osteoarthritis.

N.B. clinical symptoms are usually due to presence of other developmental defects.

Associated anomalies:

- ***atlanto-occipital anomalies*** are frequent (one of major reasons for associated morbidity).
    N.B. main neurological complications result from ***craniocervical instability*** → spinal cord compromise!

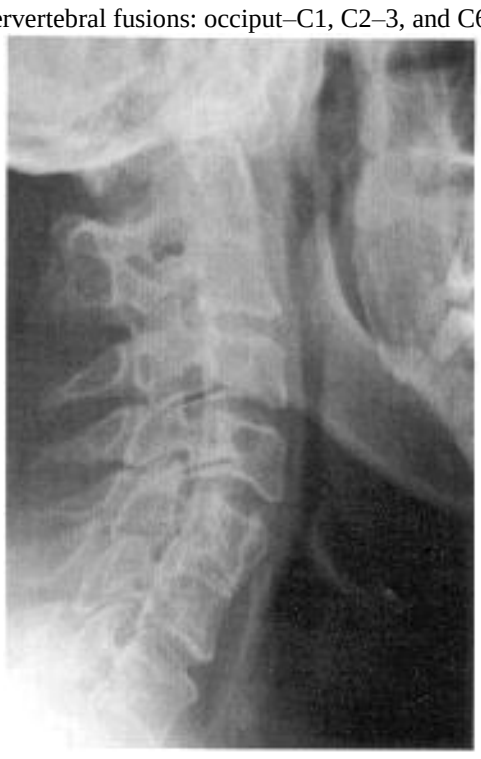
- ***kyphosis & scoliosis*** are frequent.
- ***other skeletal malformations*** (esp. Sprengel's deformity – congenital scapula elevation and medial rotation) can be associated.
- ***congenital deafness*** (faulty development of osseous inner) – 20-30% patients.
- patients may have ***GU*** anomalies (incl. unilateral renal agenesis), ***cardiovascular*** anomalies.

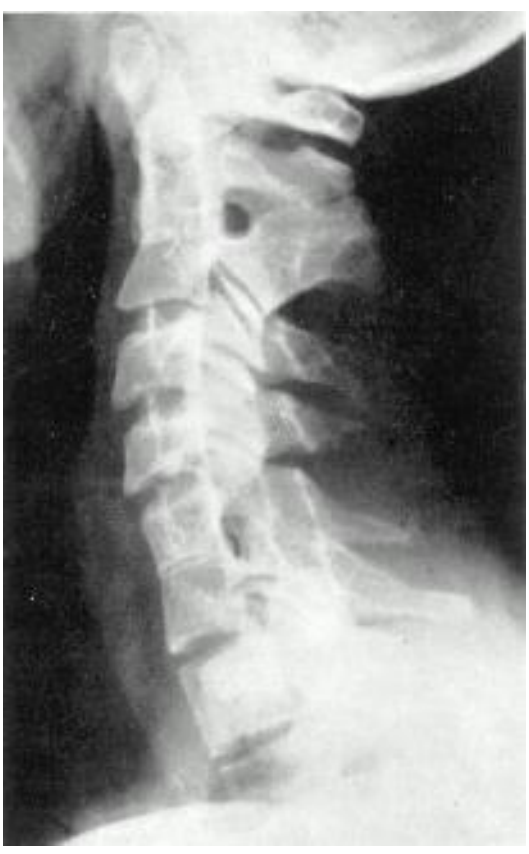
MURCS syndrome (Müllerian duct aplasia, renal aplasia, cervicothoracic somite dysplasia) - Klippel-Feil anomaly with absence of vagina & uterus, renal agenesis or ectopy; hearing and GI defects.

ESCOBAR syndrome - autosomal recessive cervical vertebral fusion and other bony defects, ptosis, hypertelorism, pterygia of neck, axillae, and other joints; genital anomalies; small stature.

DIAGNOSIS

Intervertebral fusions: occiput–C1, C2–3, and C6–7:





ATLANTO-AXIAL ROTATORY FIXATION (AARF)

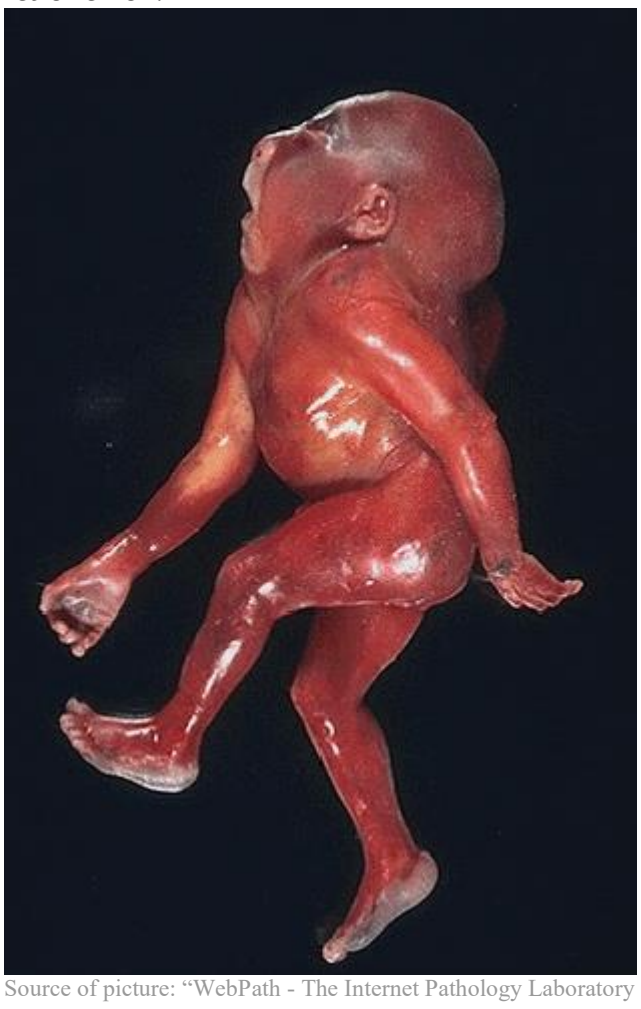
- rare form of torticollis in children: rotatory fixation between C_1 and C_2 (within normal range of motion) without subluxation.

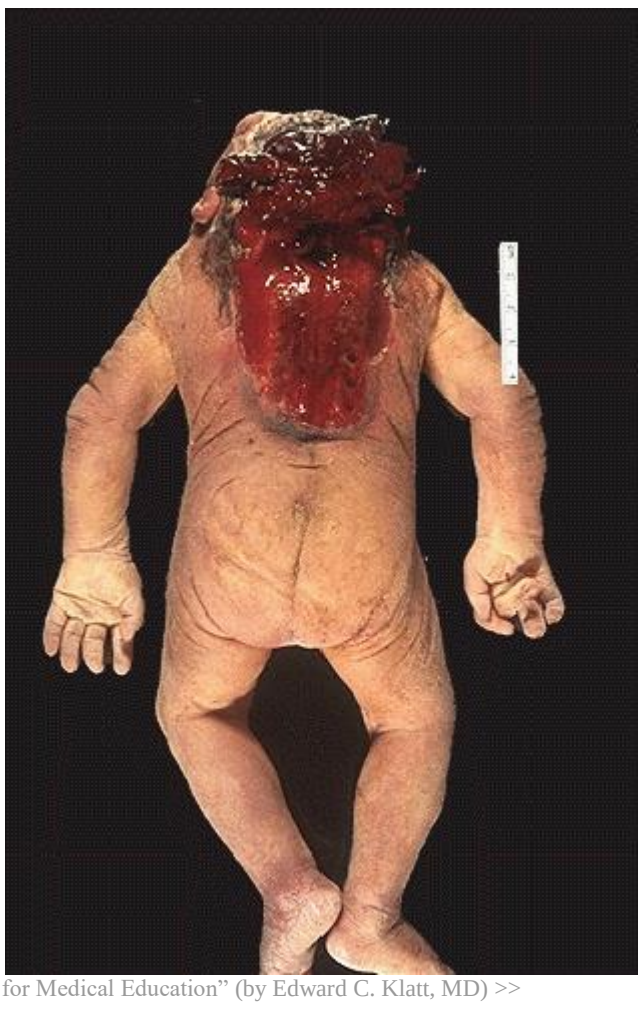
- mechanism - *lax ligaments* and possibly *synovium interposition* in intervertebral joints.
- clinically - torticollis persisting for > 2 weeks.
- requires careful diagnostic work-up - **total muscle relaxation** (under *general anaesthesia*) and **CT** of upper cervical spine in neutral position as well as in maximum rotation to right and left;
  AARF is present if there is asymmetrically reduced rotation between C1 and C2.
- requires aggressive* therapy – **traction**.

*if unsuccessful, AARF will result in permanent rotatory malalignment with ankylosis

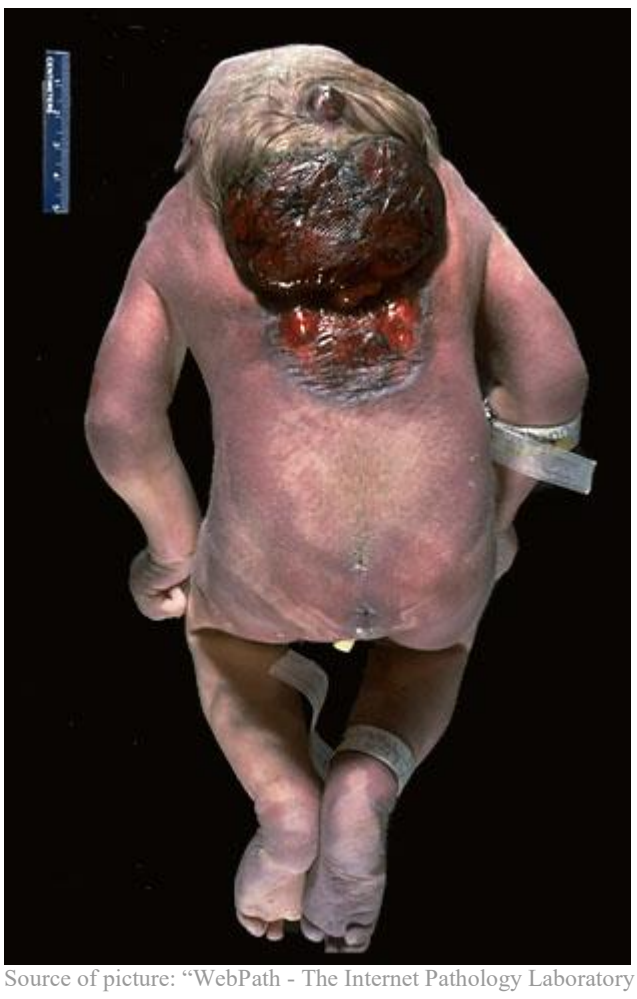
INIENCEPHALY

- cranial defect at occiput, with brain exposed; often in combination with cervical rachischisis and retroflexion:





Source of picture: "WebPath - The Internet Pathology Laboratory for Medical Education" (by Edward C. Klatt, MD) >>



Source of picture: "WebPath - The Internet Pathology Laboratory for Medical Education" (by Edward C. Klatt, MD) >>

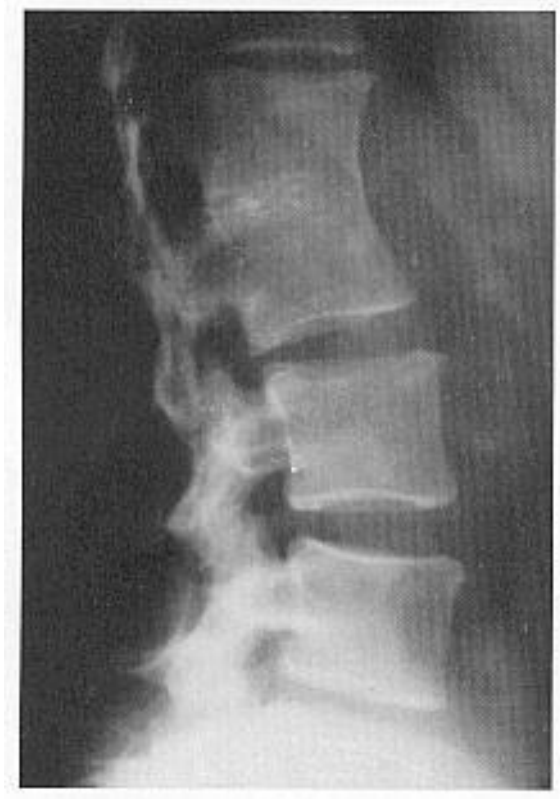
SPINE

VERTEBRAL FUSION ANOMALIES

- intervertebral discs are narrow and partly bridged by regions where disc material never developed.

- determined very early in development!
- fused segments show *varying degrees of hypoplasia* (when multiple segments are involved, *marked dysplasias* such as hemivertebrae are also often present).
- in marked cases, term **SYNSPONDYLISM** is used (term **Klippel-Feil syndrome** is appropriate when *cervical* region is predominantly involved).

Congenital fusion of two lumbar vertebrae ("block vertebra"): note concavity of anterior vertebral contour at level of expected disc space (not seen in surgical fusions!) and posterior remnant of intervertebral disc:

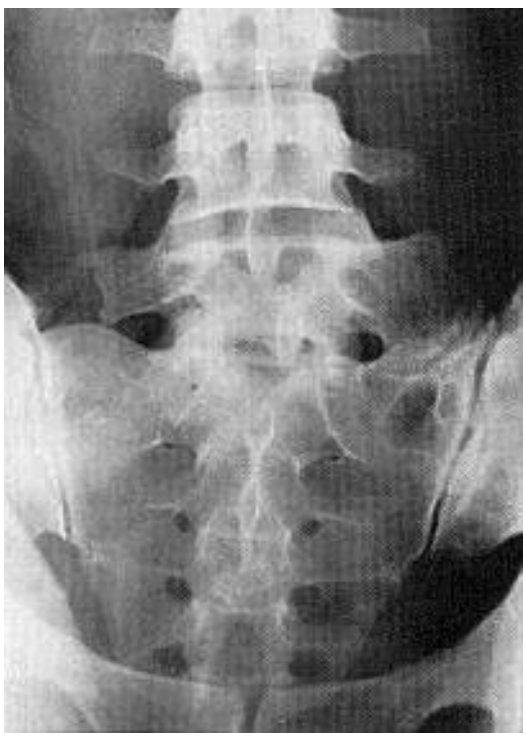


Source of picture: John H. Juhl "Paul and Juhl's Essentials of Radiologic Imaging", 7th ed. (1998); Lippincott Williams & Wilkins; ISBN-10: 0-397-58421-0 >>

TRANSITIONAL VERTEBRAE

- vertebra ***at junction of major divisions of spine*** has CHARACTERISTICS OF BOTH DIVISIONS:
  - **C7** may have ***ribs*** (unilateral or bilateral) attached to transverse processes (≈ 6% of normal population);
    - cervical ribs may be short or may be long enough to articulate with sternum.
    - cervical rib may be fused or may form pseudarthrosis with first rib.
    - cervical rib (as it passes anteriorly) may *compress subclavian vessels* → venous thrombosis / arterial insufficiency.
    - even if cervical rib is short, fibrous band may extend from its tip to first rib or to sternum (source of compression of subclavian vessels).
  - **L1** may have ***rudimentary ribs*** articulating with transverse processes.
  - **L5** may be ***partially sacralized*** (often one transverse process fused with sacrum, other free) with rudimentary disc between them (≈ 6% of normal population).
  - **S1** segment may become ***partially lumbarized***.
- most frequent at *thoracolumbar* and *lumbosacral* junctions:
- transition may be complete (e.g. 6 lumbar vertebrae and 4 sacral segments; 13 thoracic and 4 lumbar); usually, addition of segment to one division of spine is corrected at another level.
- main significance of transitional vertebrae - may result in level being wrongly identified preoperatively (e.g. when MRI is used without X-ray).

Partially sacralized L5 vertebra - left transverse process is enlarged and articulates with sacrum; right transverse process is free:

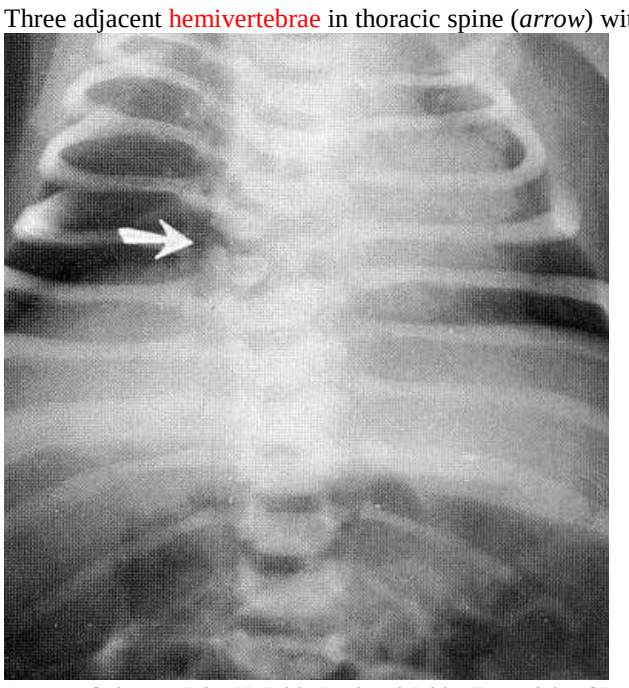


HEMIVERTEBRAE

LATERAL HEMIVERTEBRA - incomplete development of lateral half of vertebral body.

- failure of development of one of lateral centers of chondrification.
- in AP radiograph, hemivertebra has triangular shape.
- causes **scoliosis** with acute lateral angulation of spine.
- hemivertebra in thoracic region has only one rib (on side of ossified center).

Three adjacent hemivertebrae in thoracic spine (*arrow*) with associated convex rightward scoliosis:



Source of picture: John H. Juhl "Paul and Juhl's Essentials of Radiologic Imaging", 7th ed. (1998); Lippincott Williams & Wilkins; ISBN-10: 0-397-58421-0 >>

DORSAL HEMIVERTEBRA - failure of development of ventral fetal ossification center.

- causes progressive **kyphosis** that may require posterior fusion at level of hemivertebra.

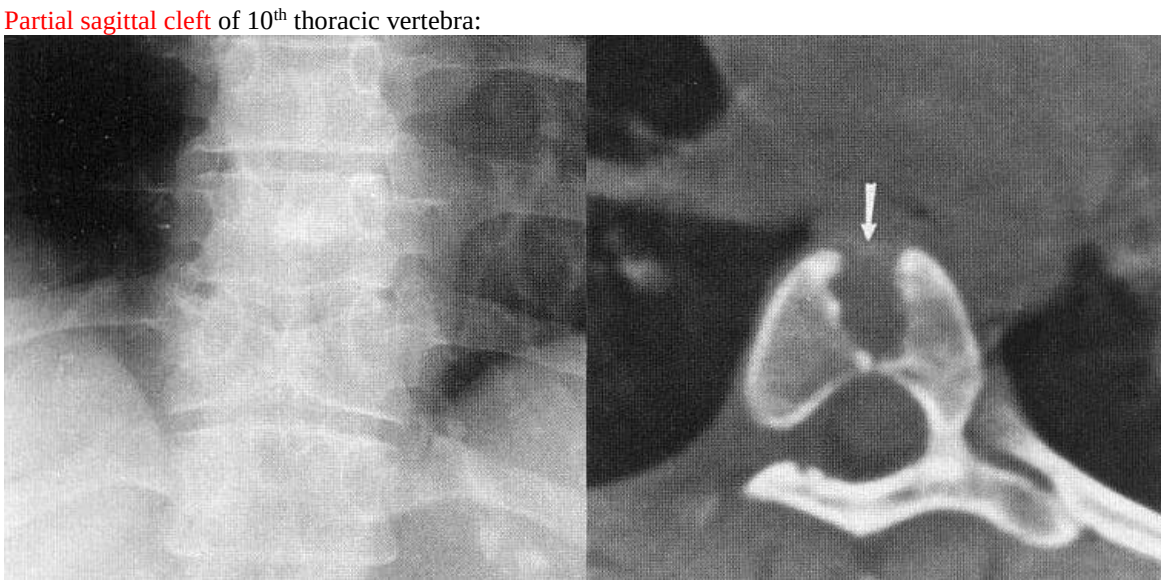
VENTRAL HEMIVERTEBRA (rare) - failure of development of dorsal ossification center.

BUTTERFLY VERTEBRAE

- failure to fuse of two lateral centers of chondrification for vertebral body → cleft in midsagittal plane (dividing body into two lateral halves).

- more frequently cleft is only partial, resulting in characteristic shape "butterfly vertebra".

Partial sagittal cleft of 10th thoracic vertebra:



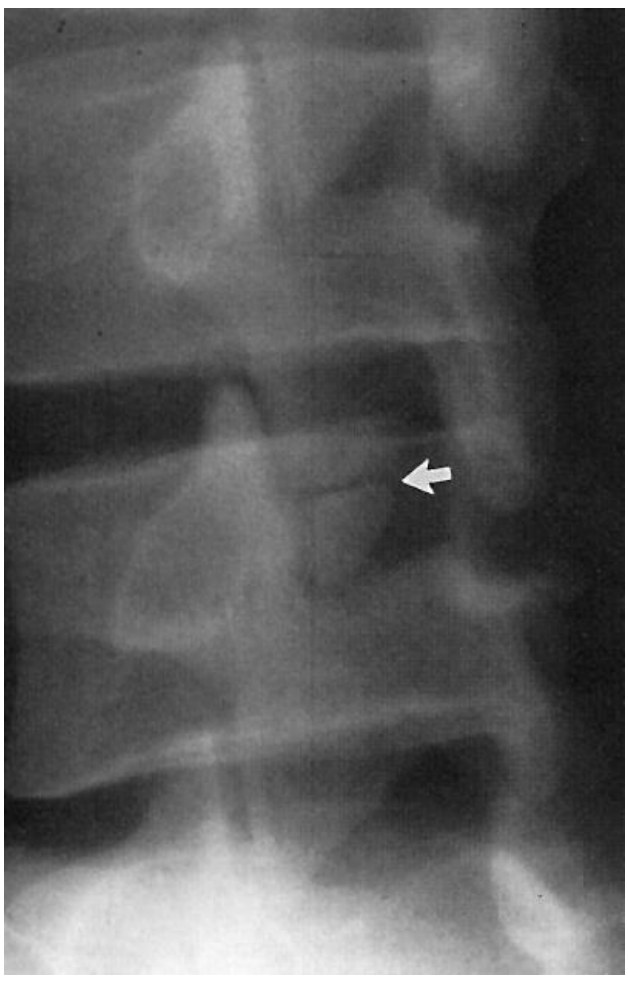
FAILURE of FUSION of SECONDARY OSSIFICATION CENTERS

- secondary ossification center* fails to unite to vertebral body → persist into adult life as ***separate bony fragment*** (esp. at inferior articular process).

*normally, secondary ossification centers appear at tips of all spinous and transverse processes.

- can be confused with fracture!
  - smooth corticated margins of fragment help differentiate this anomaly from acute fracture.

Nonunited apophysis (*white arrow*) of L4 inferior articular process (oblique lumbar radiograph); smooth, sclerotic margins indicate that this is not acute fracture:

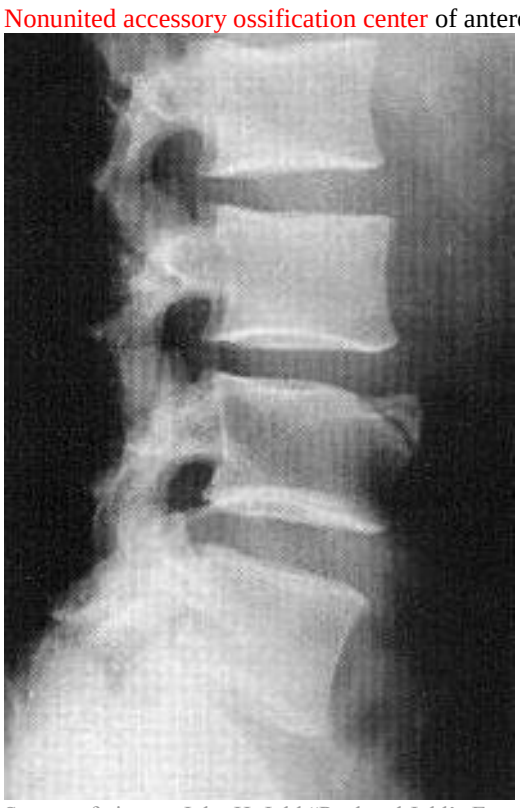


LIMBUS VERTEBRA

- ***anterior interposition of intravertebrally herniated nuclear material*** prevents fusion of portion of peripheral ring apophysis with adjacent vertebral end-plate.

- on lateral radiographs: triangle-shaped bony mass along anterosuperior corner with corresponding defect in adjacent vertebral body.
- smooth bony margins and characteristic shape and location differentiate from fracture.

Nonunited accessory ossification center of anterosuperior corner of L4:



Source of picture: John H. Juhl "Paul and Juhl's Essentials of Radiologic Imaging", 7th ed. (1998);
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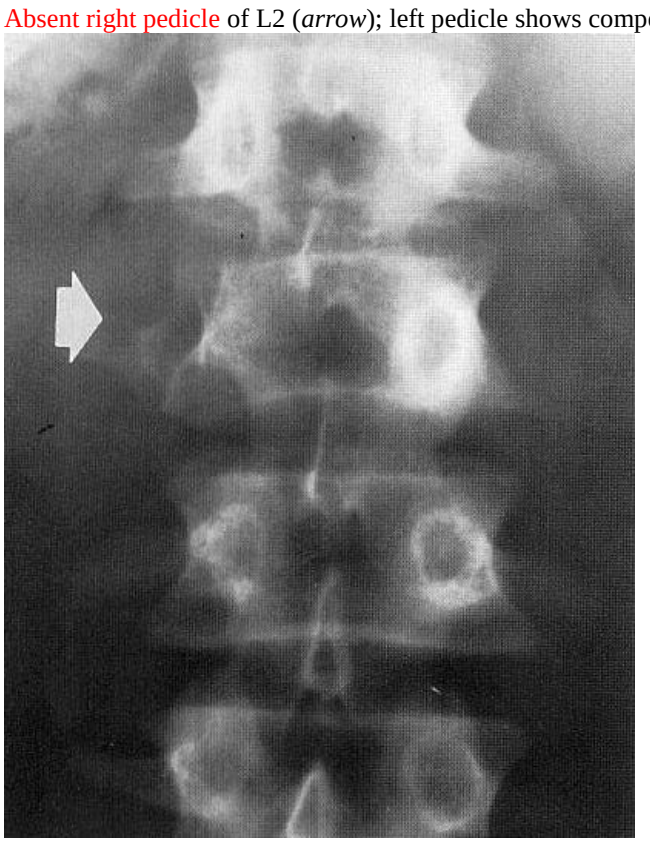
PEDICLE ANOMALIES

- absence or hypoplasia of pedicle with compensatory hypertrophy* of opposite pedicle.
*differentiates from destructive lesion of pedicle.

- ipsilateral intervertebral foramen is widened; posterior displacement of maldeveloped lateral mass.
- flattening / thinning of pedicles at Th12 / L1 is common anatomic variant.

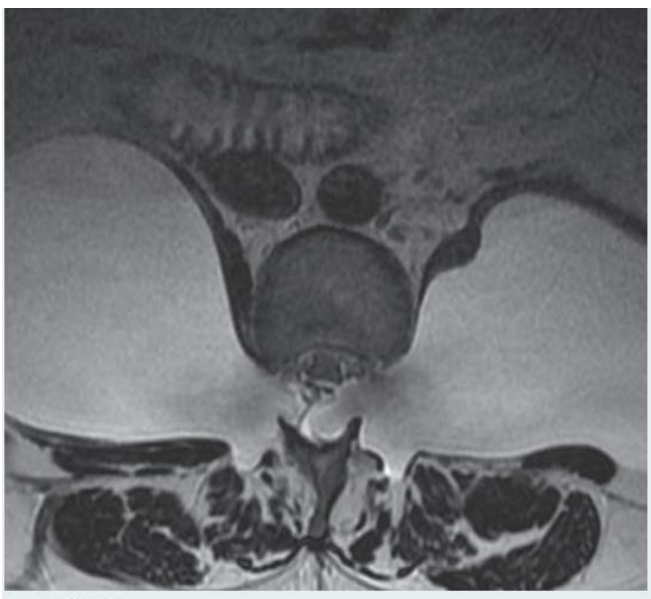
Absent right pedicle of L2 (*arrow*); left pedicle shows compensatory sclerosis and hypertrophy:



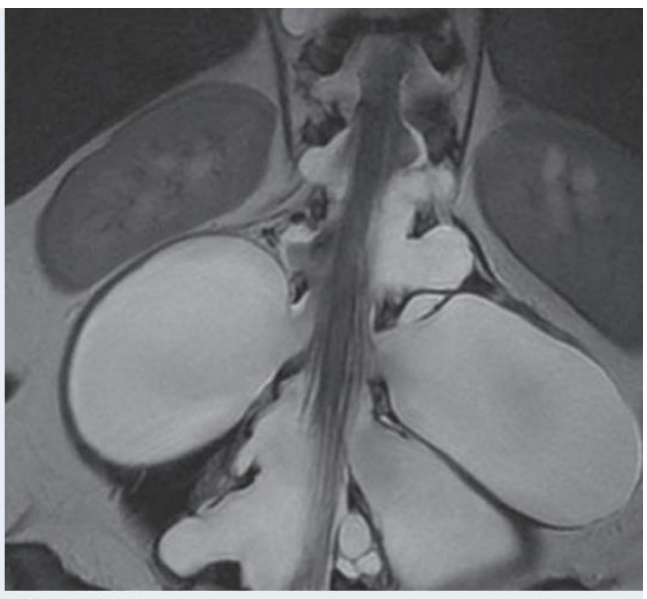
LATERAL MENINGOCELE SYNDROME

- rare hereditary autosomal dominant connective tissue disorder
- pan-spinal meningoceles secondary to dural ectasia

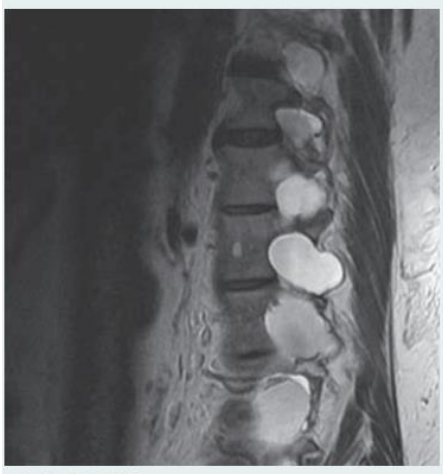
T2 MRI of thoracolumbar spine (a-c) - intraspinal and paraspinal meningoceles.
Plain lateral radiograph (d) - large neuroforamina, thinned out pedicles and congenital fusion of T10-L4 vertebral bodies.
Axial non-contrast CT (e) - thin stretched out pedicles.



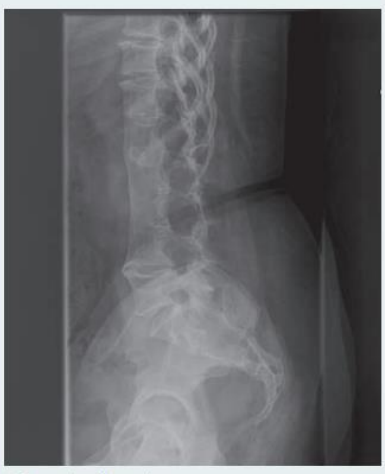
a. Axial T2 MRI



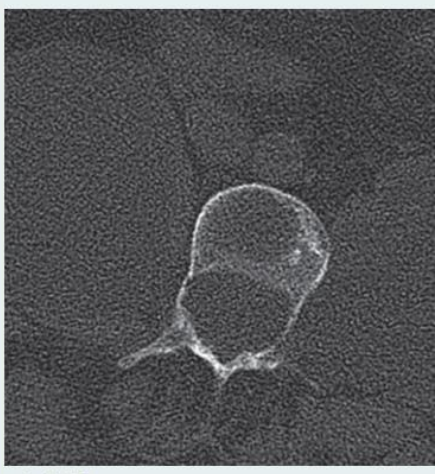
b. Coronal T2 MRI



c. Sagittal T2 MRI



d. Lateral radiograph



e. Axial CT scan

BIBLIOGRAPHY for ch. "Developmental Anomalies" → follow this LINK >>