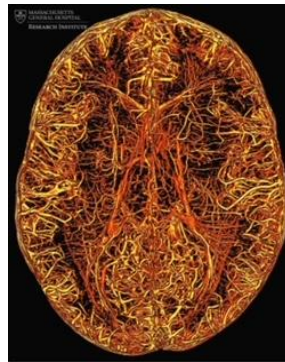
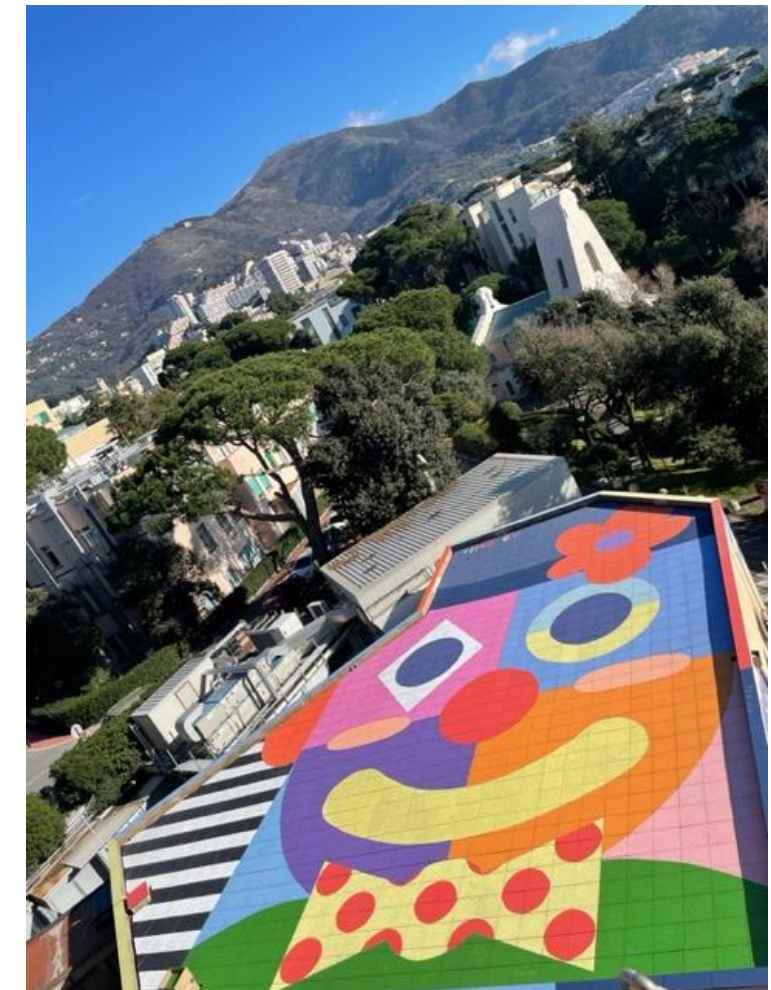
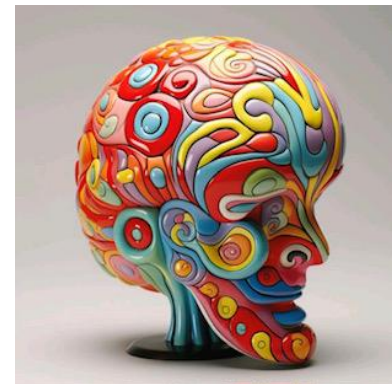


Chiari 1 e Instabilita' cranio-cervicale: aspetti chirurgici



Gianluca Piatelli
Head Neurosurgery Unit,
Dept. Of Surgery
IRCCS Istituto Giannina Gaslini, Italy



gianlucapiatelli@gaslini.org

Chiari Malformation Classification

CM-1: paraxial mesoderm disorder, with abnormalities of the posterior cranial fossa (mostly small) and the consequent herniation of the cerebellar tonsils

CM1-A: with syringomyelia on MRI

CM1-B: without syringomyelia on MRI

CM-2: associated with myelomeningocele (prevalent in childhood), hydrocephalus, and, less frequently, hydrosyringomyelia. Other types of intracranial defects (hypoplastic tentorium cerebelli, cranial lacunae, anomalies of the Sylvius aqueduct) may exist

CM-1,5: cerebellar tonsils and brainstem herniation (kinking) below the McRae line

CM-0: volumetrically small posterior fossa, obliteration of the cisterna magna, cerebellar tonsils positioned at the foramen magnum and a syringomyelia in the cervical spinal cord

“Complex CM1”: coexistence of CM1 with skeletal deformities of craniovertebral junction and cervical spine (Klippel-Feil anomaly, atlanto-occipital fusion, basilar invagination, retroversion of the odontoid process)

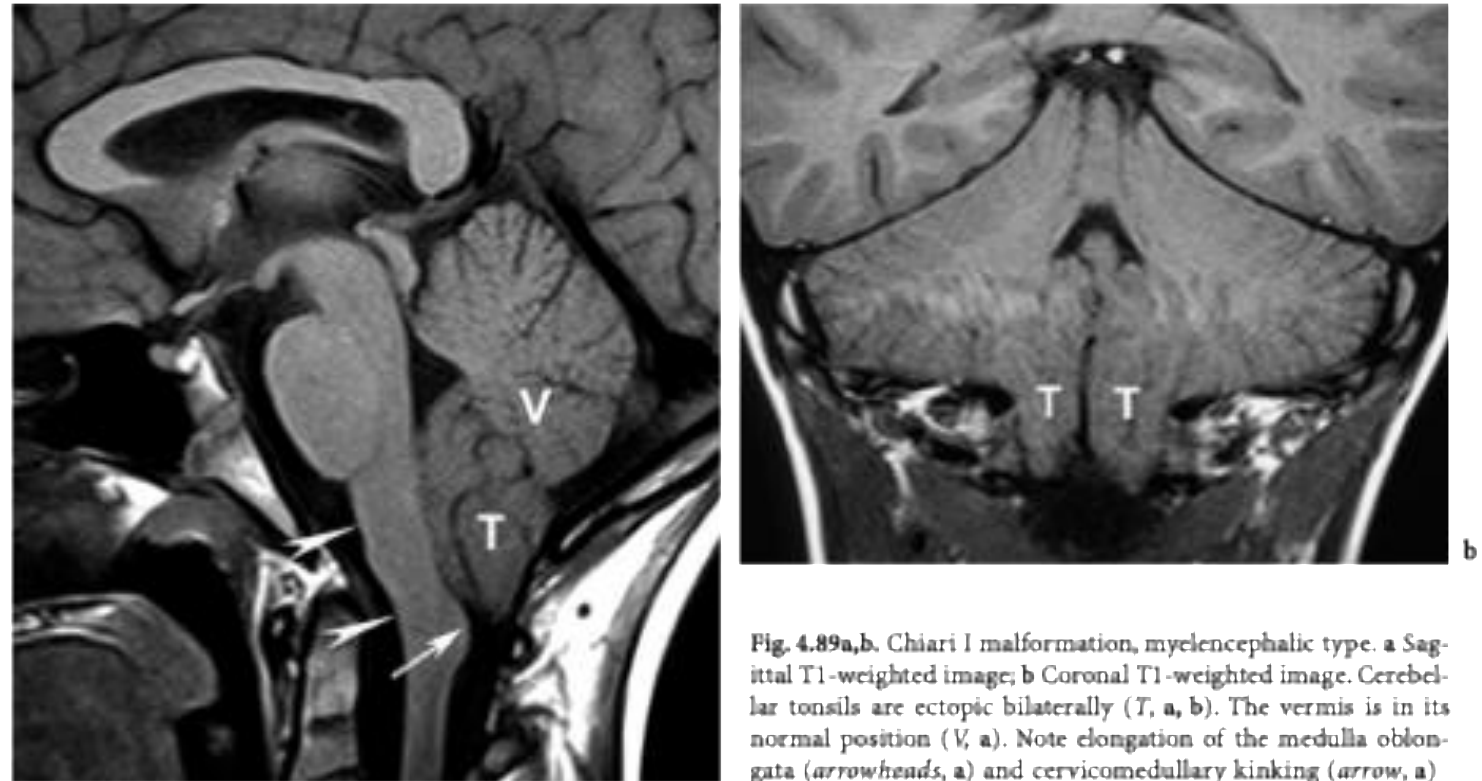
PAOLO TORTORI-DONATI

Pediatric Neuroradiology

Brain Head and Neck Spine

 Springer

Gennaio 2005

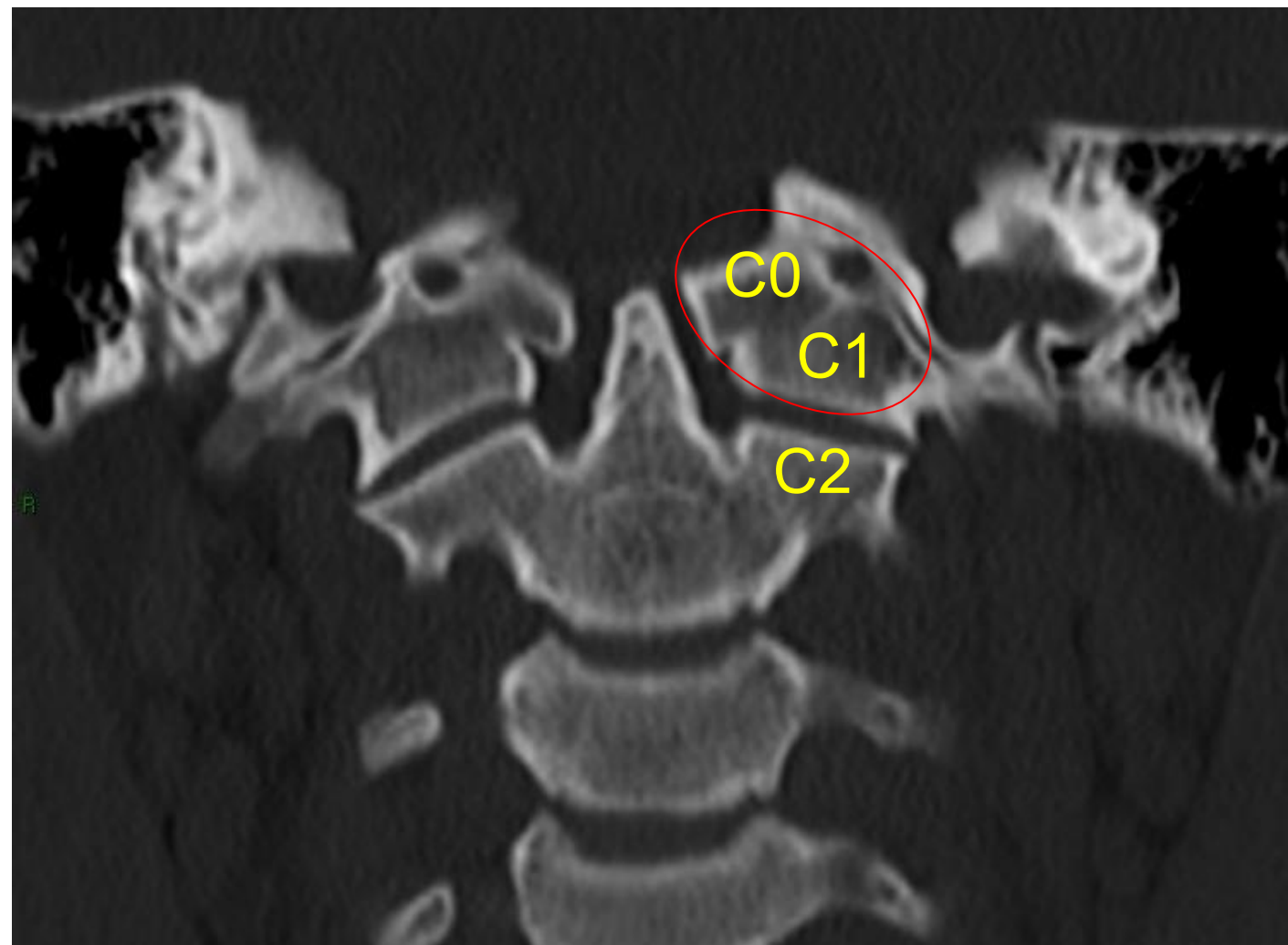
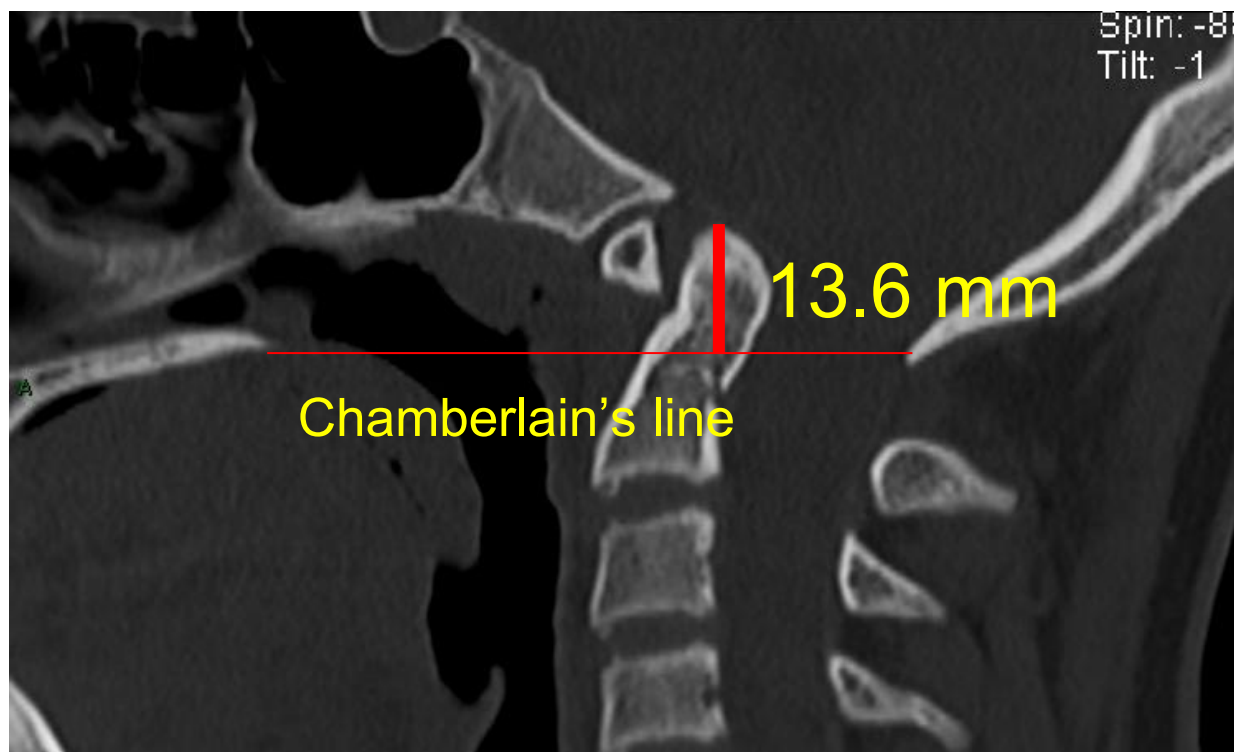


“bulbar” or *“myelencephalic”* variant of the Chiari I malformation

This variant probably results from a smaller posterior fossa as compared to classical Chiari I malformation, with increased degree of neural overcrowding and downward displacement of the medulla. It is noteworthy that associated skeletal anomalies, such as platybasia and basilar invagination, also are relatively more common in patients with the bulbar variant than in classical Chiari I's (P. Tortori-Donati and A. Rossi, unpublished observations).

CVJ anomalies and Chiari I

- CVJ abnormalities including basilar invagination, occipitalization of the atlas, and retroversion of the odontoid are more common in the Chiari 1.5 malformation
- These anomalies may coexist in individual patients





Diagnosis and treatment of Chiari malformation type 1 in children: the International Consensus Document

Luca Massimi¹ · Paola Peretta² · Alessandra Erbetta³ · Alessandra Solari⁴ · Mariangela Farinotti⁴ · Palma Ciaramitaro⁵ · Veronica Saletti⁶ · Massimo Caldarelli¹ · Alexandre Casagrande Canheu⁷ · Carlo Celada⁸ · Luisa Chiapparini³ · Daniela Chieffo⁹ · Giuseppe Cinalli¹⁰ · Federico Di Rocco¹¹ · Marika Furlanetto¹² · Flavio Giordano¹³ · George Jallo¹⁴ · Syril James¹⁵ · Paola Lanteri¹⁶ · Christian Lemarchand¹⁷ · Martina Messing-Jünger¹⁸ · Cecilia Parazzini¹⁹ · Giovanna Paternoster¹⁵ · Gianluca Piatelli²⁰ · Maria. A. Poca²¹ · Prab Prabhakar²² · Federica Ricci²³ · Andrea Righini¹⁹ · Francesco Sala²⁴ · Juan Sahuquillo²¹ · Marcus Stoodley²⁵ · Giuseppe Talamonti²⁶ · Dominic Thompson²⁷ · Fabio Triulzi²⁸ · Mino Zucchelli²⁹ · Laura Valentini¹² · International Experts - Jury of the Chiari & Syringomyelia Consensus Conference, “Milan, November 11–13, 2019”

Received: 16 January 2021 / Accepted: 8 May 2021 / Published online: 7 June 2021
© The Author(s) 2021



Conclusions

The consensus process on the controversial topics in children was fruitful and allowed the authors to get some results that can be used as a base for future studies looking for recommendations or guidelines. They can be summarized as follows:

- (1) The old definition of CM1 is still accepted, although the definition based on level of descent in millimeters alone has little clinical correlation. Attention must be paid not only on the degree of tonsillar descent but also to the morphology and volume of the posterior fossa. Transient tonsillar descent resulting from remitting primary diseases is not a CM1. Syringomyelia is considered if large and/or progressing and/or symptomatic (thin hydromyelia is a para-physiological finding);
- (2) No surgical indications are recognized in asymptomatic children with CM1 but without syringomyelia;
- (3) To date, no etiological relationship has been found between CM1 and epilepsy or autism;
- (4) Asymptomatic children should remain in contact with their physicians until the end of their growth. If needed, clinical and radiological controls are scheduled according to the characteristics of each patient;
- (5) Symptomatic subjects benefit from a multidisciplinary team to rule out symptoms/signs unrelated to CM1 (e.g., key role of the neurologist in case of migraine-like headache). Some tools (polysomnography, SSEPs, MEPs) are useful in selected, doubtful cases;
- (6) In CM1 with associated hydrocephalus, hydrocephalus should be treated first to reduce the intracranial pressure, limiting the posterior fossa decompression to the children with persisting symptoms;
- (7) CM1 can result from both non-syndromic and syndromic craniosynostoses whose treatment needs to be incorporated into the treatment strategy. Cranial vault surgery may address cerebellar ectopia. The posterior fossa decompression or the posterior distraction should be limited to selected cases;
- (8) Tethered cord is rarely and sporadically associated with CM1 and its management is independent of CM1. A manifest and symptomatic tethered cord is treated by intradural de-tethering but no effects on CM1 are expected;
- (9) Bony posterior decompression and duraplasty are both acceptable techniques of foramen magnum decompression. The first offers a very low rate of complications and a short hospital stay but shows a higher risk of persisting symptoms: it can be considered in children without severe symptoms or large syringomyelia. The second one ensures more consistent radiological results but with a higher rate of (CSF-related) complications;
- (10) The bony decompression has to be focused on a proper opening of the foramen magnum and C1 (no need for large opening of the occipital squama or C2);
- (11) The key points of duraplasty are to keep unviolated the arachnoid plane and to perform a proper dural closure (no need of tonsils coagulation unless this appears necessary to effect an effective and capacious decompression: e.g., tonsils extending below C2);
- (12) The surgical failure can be defined by the persistence of symptoms 1 year after surgery. The radiological persistence of CM1/syringomyelia should be interpreted in clinical context before a decision to re-operate;
- (13) Too small opening and incorrect indication are the main causes of failure of bony decompression alone. Re-operation with a larger decompression and duraplasty are the respective solutions;
- (14) Arachnoiditis is an important cause of failure of duraplasty. Proper surgical technique, re-operation with arachnoid lysis, and, in very selected cases, IV ventricle stenting are the solutions;
- (15) Asymptomatic postoperative CSF collection can be managed conservatively, while they should be treated promptly, if symptomatic;
- (16) CVJ instability or craniovertebral deformity (basilar impression) must be ruled out in any (CM1) patient presenting with atypical clinical and/or radiological findings. Only symptomatic subjects should be treated, using a technique chosen according to the patient's characteristics. The posterior decompression for CM1 can be added in selected cases;
- (17) Syringomyelia must be always ruled out in CM1 children by MRI of the whole spinal cord;
- (18) Asymptomatic, isolated, and stable syringomyelia shows a very low risk of evolution (no operation is indicated in this instance). It may require treatment only if symptomatic and the treatment is etiological. The direct management (shunting) should be considered only after failure of the etiologic one.

Surgical Management of Chiari Malformation Type I and Instability of the Craniocervical Junction Based on Its Pathogenesis and Classification

Misao NISHIKAWA,¹ Paolo A. BOLOGNESE,² Toru YAMAGATA,¹ Kentarou NAITO,³
Hiroaki SAKAMOTO,⁴ Mistuhiro HARA,¹ Kenji OHATA,⁵ and Takeo GOTO³

¹Department of Neurosurgery, Moriguchi-Ikuno Memorial Hospital, Moriguchi, Osaka, Japan

²Department of Neurosurgery, Chiari Ehlars Danlos Syndrome Center, Mount Sinai South Nassau, Lake Success, Nassau, New York, USA

³Department of Neurosurgery, Osaka Metropolitan University Graduate School of Medicine, Osaka, Osaka, Japan

⁴Department of Pediatric Neurosurgery, Osaka City General Hospital, Osaka, Osaka, Japan

⁵Department of Neurosurgery, Naniwa-Ikuno Hospital, Osaka, Osaka, Japan

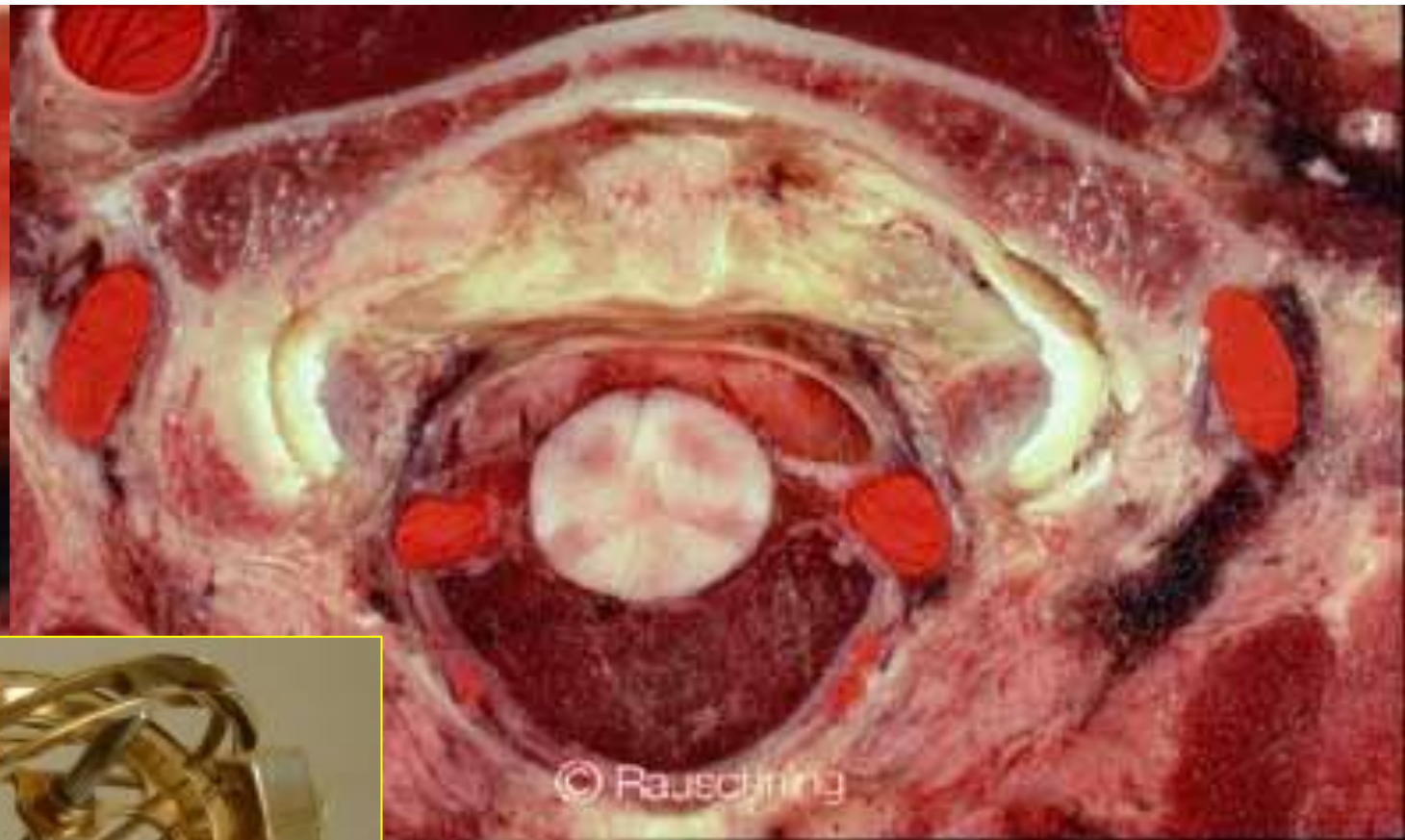
CCF was performed in a total of 70 cases,

Table 3 Results of morphometric measurements of craniocervical junction in the craniocervical traction test

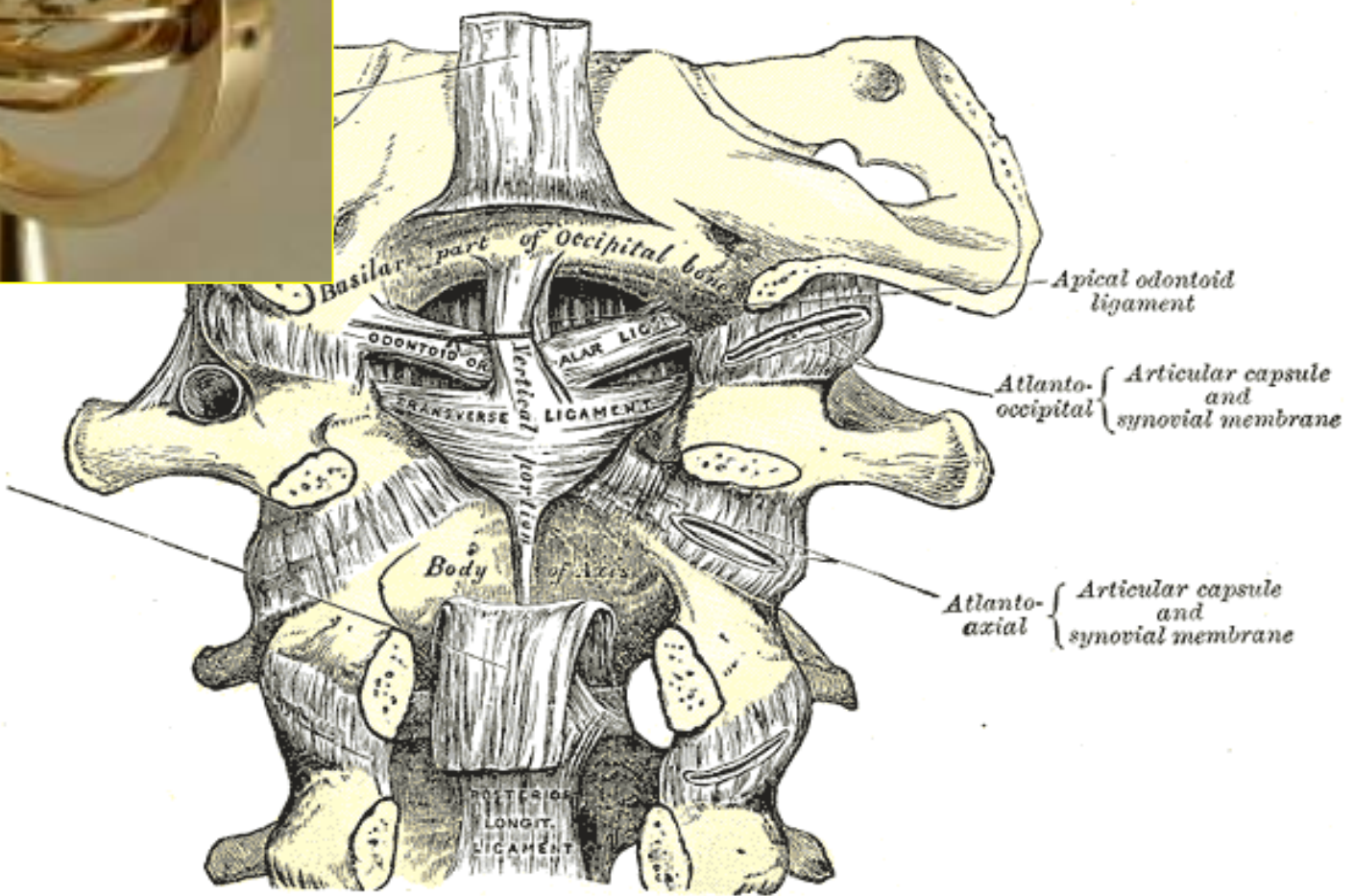
	Normal Controls 30 cases (12-year-olds)	Craniocervical test		During operation in positioning	Follow up CSJ cases	Follow up ICSJ cases
		Nontraction	Traction			
ADI (mm)						
Flexion position in sitting	3.4 ± 0.47	7.4 ± 1.24†	3.3 ± 0.55	N.A.	3.4 ± 0.53	7.4 ± 1.24†
Neutral position in sitting	2.8 ± 0.54	3.1 ± 0.63	3.0 ± 0.51	3.2 ± 0.45	3.0 ± 0.48	3.1 ± 0.63
Extension position in sitting	2.0 ± 0.48	2.3 ± 0.58	2.7 ± 0.47	N.A.	3.2 ± 0.52	2.3 ± 0.58
Supine position	2.8 ± 0.57	2.4 ± 0.34	2.5 ± 0.74	in positioning	2.2 ± 0.64	2.5 ± 0.52
Sitting position	2.4 ± 0.58	2.1 ± 0.42	1.94 ± 0.44	2.2 ± 0.781	2.4 ± 0.68	2.3 ± 0.64
BAI (mm)						
Supine position	1.8 ± 1.21	2.0 ± 1.25	2.2 ± 1.52	in positioning	2.2 ± 1.30	2.5 ± 1.48
Sitting position	2.1 ± 1.88	5.1 ± 1.22†	1.85 ± 0.71	2.0 ± 1.21	2.3 ± 1.38	5.3 ± 1.57†
BDI (mm)						
Supine position	7.4 ± 1.58	8.7 ± 1.70	8.8 ± 2.23	in positioning	8.3 ± 1.82	7.6 ± 2.01
Sitting position	7.2 ± 1.59	3.9 ± 1.81†	8.4 ± 2.20	8.7 ± 2.70	8.2 ± 1.84	3.8 ± 1.92†
AXA (°)						
Supine position	37.6 ± 6.55	35.2 ± 5.44	4.72 ± 6.23	in positioning	34.5 ± 6.04	35.3 ± 5.43
Sitting position	37.4 ± 6.84	36.5 ± 5.96	35.8 ± 5.88	36.7 ± 6.40	35.4 ± 5.44	34.4 ± 5.41
CXA (°)						
Supine position	147.6 ± 6.61	145.2 ± 5.27	146.5 ± 5.68	in positioning	144.8 ± 5.49	145.8 ± 5.58
Sitting position	147.8 ± 6.00	136.8 ± 6.47†	145.4 ± 5.88	146.8 ± 6.45	145.2 ± 5.58	134.2 ± 5.58†

CCF was not performed in children younger than 12 years.

In children younger than 12 years, the relationships between anatomical structures are under development, so there is a possibility of overestimating instability.



Membrana tectoria, divi



CCJ instability

Instability occurs each time the anatomical relationships between the articular components are altered or have the propensity of becoming abnormal under the influence of certain solicitations



Broken red
circle



risk of neurological involvement

- Atlanto-occipital dislocation (C0-C1 instability)
- Atlanto-axial subluxation (C1-C2 instability)
- Sub-axial subluxation

osteologia

- Scheletro largamente cartilagineo: rachide ipermobile

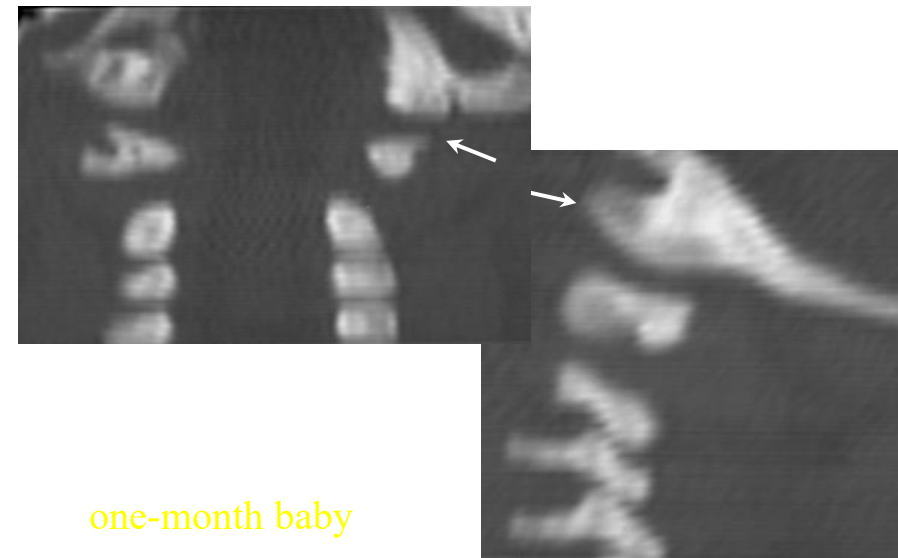
- Processi iposviluppati

- condili
- Processo uncinato (assenti <10 aa)
- Faccette articolari: orizzontali

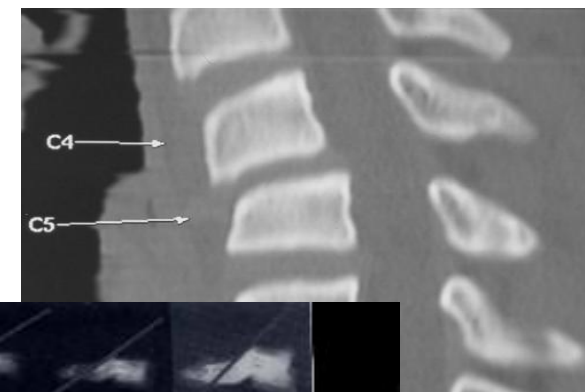
- Corpi vertebrali: orizzontalizzati e

cuneizzati

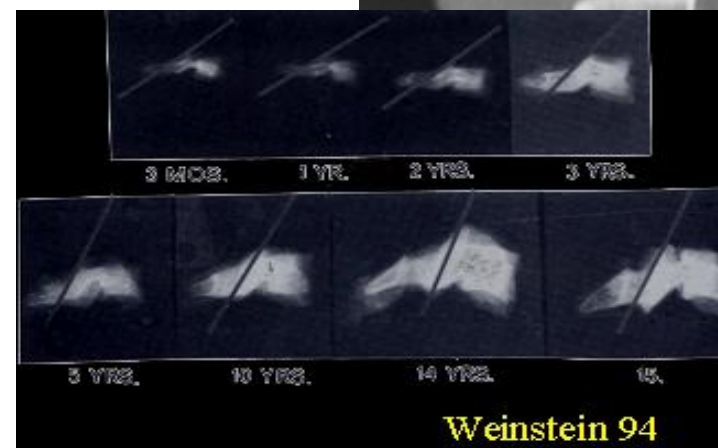
- Soggetti alla lussazione



one-month baby



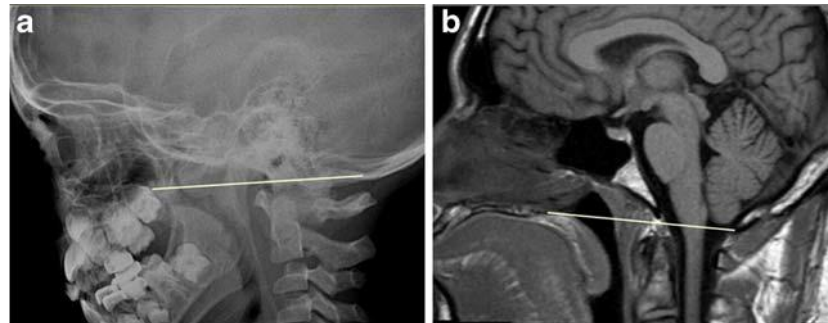
ar boy



Weinstein 94

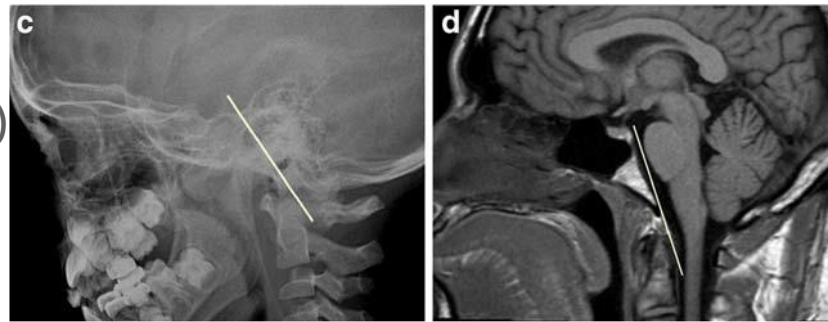
Lussazioni+frequenti delle fratture!!!!

Chamberlain's line:
posterior margin of hard
palate to opisthion



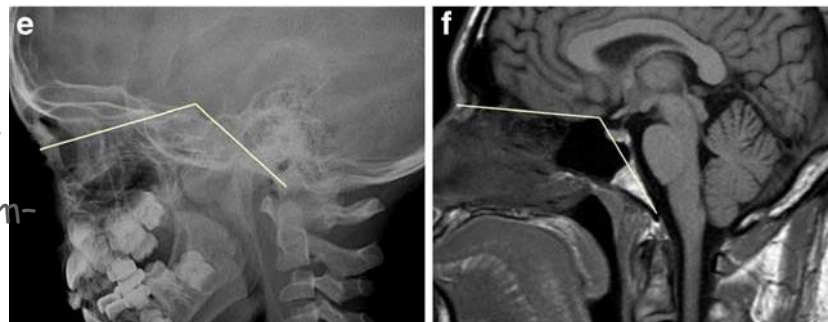
Tip of the odontoid
should be no more
than 5 mm above this
line

**Wackenheim's clivus
baseline (basilar line)**
Line extrapolated along
dorsal surface of the
clivus



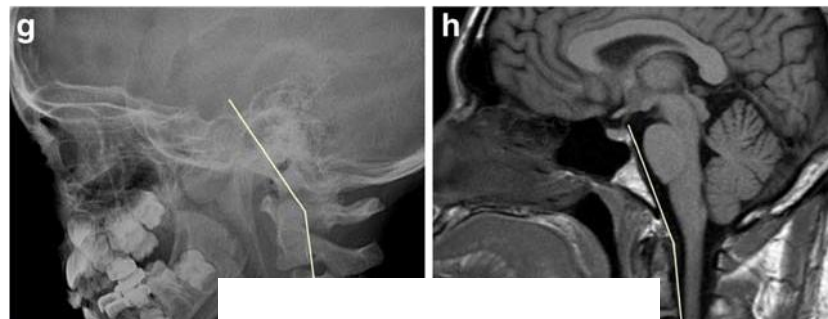
Line should fall
tangent to, or
intersect, the
posterior one third
of the odontoid

Basal angle:
Angle subtended by the
junction of the nasion-
tuberculum and tuberculum-
basion tangents



Should be less than
140°

Clivus-canal angle:
Angle formed at junction
of Wackenheim's line
and posterior vertebral
body line



Range= 150° in flexion
to 180° in extension
< 150° considered
abnormal

Criteria for unstable configurations at the CVJ

- Predental space >5mm in patients <8 yrs of age, >3 mm in those 8-10yrs & older
- Separation of atlas lat masses >6 mm
- Vertical clivus odontoid traslation > 2mm
- Gap between occipital condyles & atlas facets on lat cervical Xray
- Abnormal relationship between spinal canal e FM
- Abnormal craniocervical motion dynamics

INDICATIONS

- a) Anterior and posterior bifid C1 arches with instability, absent occipital condyles
- b) Severe reducible basilar invagination, unstable dystopic Os odontoideum and unilateral atlas assimilation
- c) Acquired phenomenon with traumatic occipito-cervical dislocation, complex CVJ fractures of C1 and C2, after transoral CVJ decompression, cranial settling in Down's Syndrome and inflammatory diseases such as Grisel's Syndrome.
- d) Clivus chordoma and osteoblastoma

"Fusion at the craniovertebral junction"
Ahmed R, Traynelis VC, Menezes AH.
Childs Nerv Syst. 2008 oct;24 (10) 1209-24

CVJ Instability in CM1: Rationale

- .Association with CVJ malformations
- .Association with genetic syndromes presenting cervical spine anomalies
- .Association with disorders and bone diseases(see fibrodisplasia)
- .Association with hereditary disorders of connective tissue (HDTC), see Ehlers-Danlos syndrome and Marfan syndrome

Embryology and bony malformations of the craniovertebral junction

Dachling Pang • Dominic N. P. Thompson

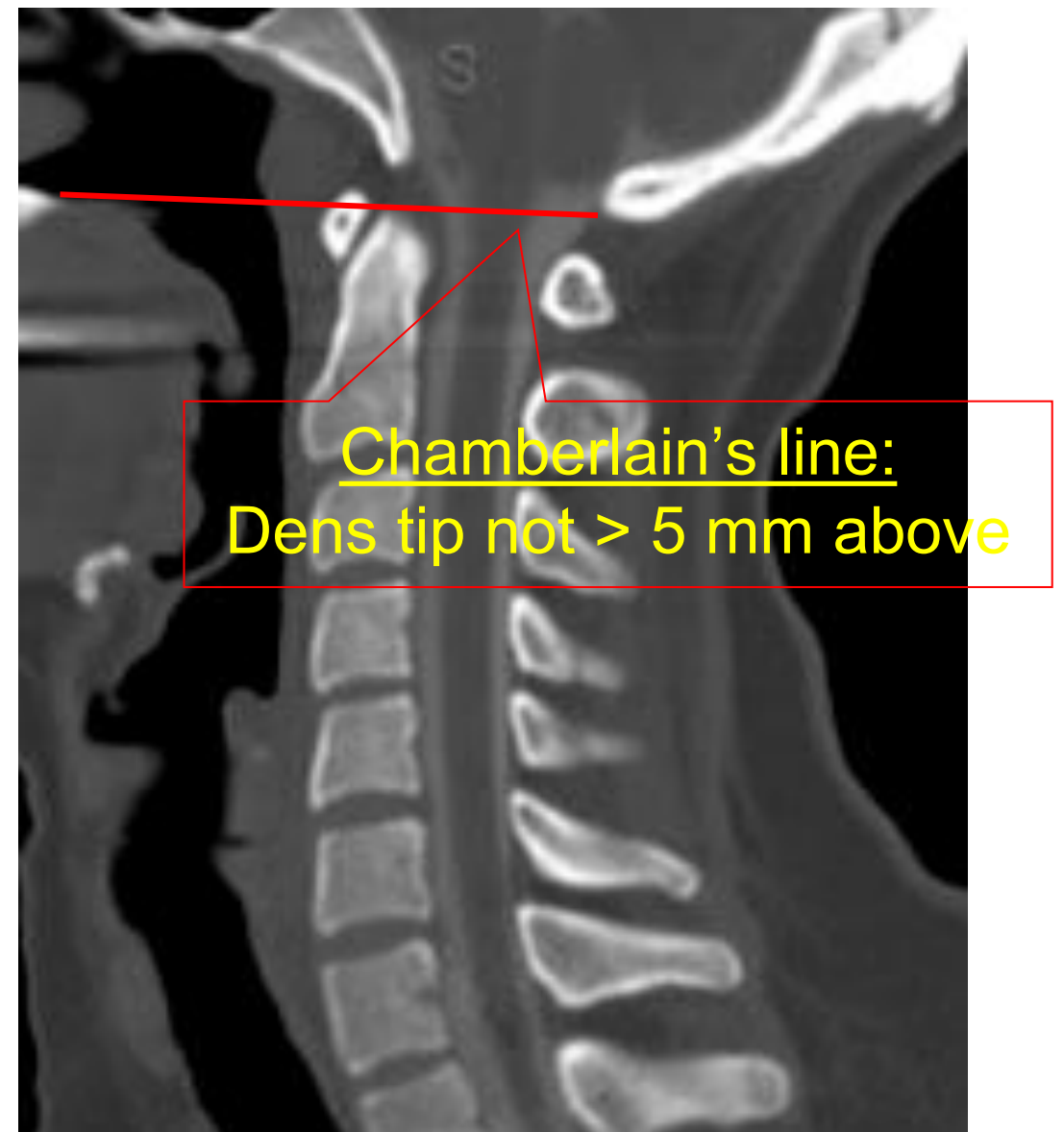
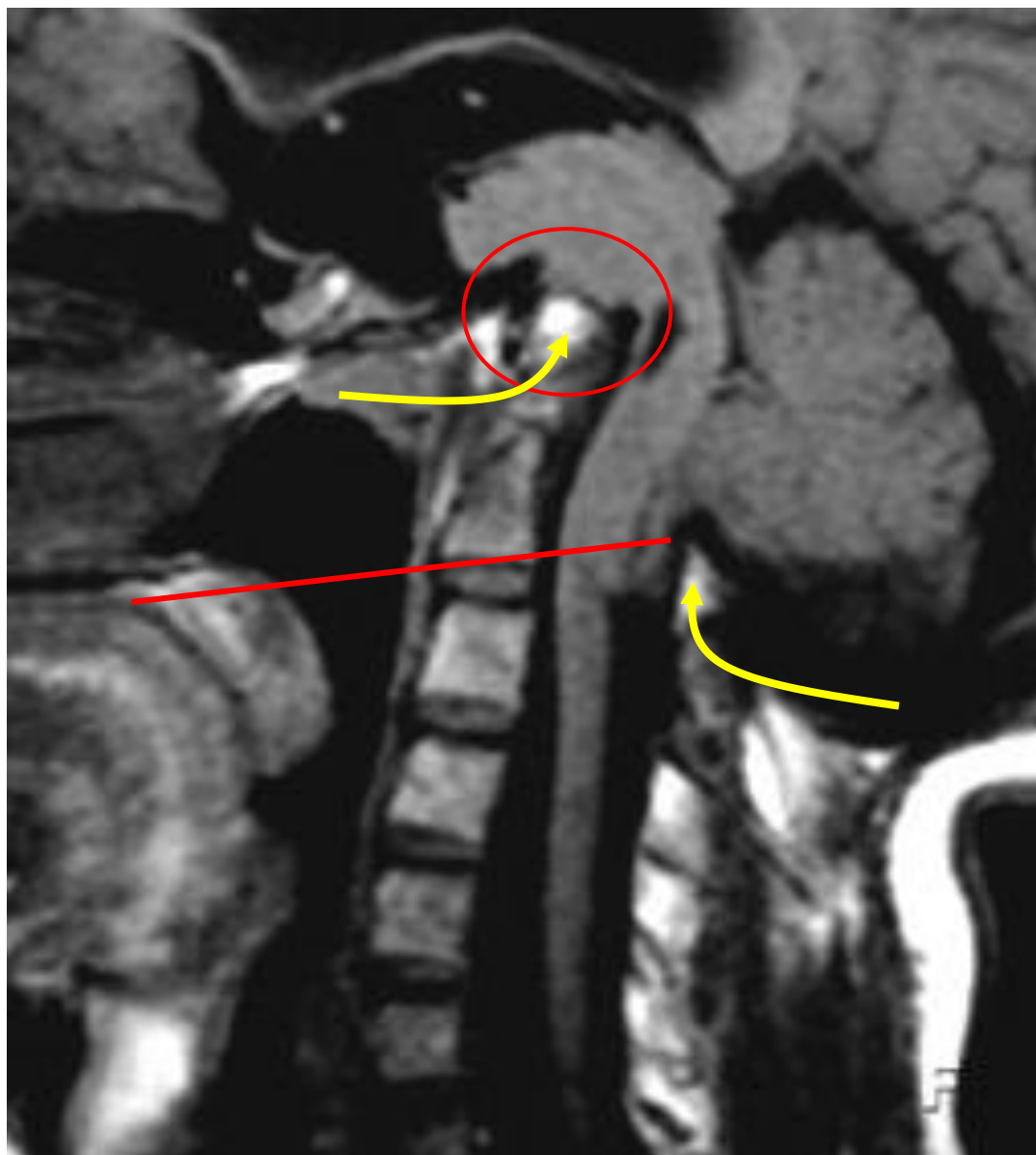
Table 1 Classification of bony malformations of the CVJ according to embryogenesis

Clinically significant CVJ bony malformations

Malformations of central pillar		Malformations of surrounding rings	
Disturbance of axial component of occipital sclerotome, proatlas and C ₁ resegmented sclerotome		Disturbance of lateral component and hypochordal bows of proatlas and C ₁ resegmented sclerotome	
Odontoid dysgeneses	Basioccipital dysgeneses	Proatlas anomalies	C ₁ sclerotome anomalies
▪ Aplasia/hypoplasia of odontoid components ▪ Disturbance of odontoid synchondroses (IBZ) – Os odontoideum – Ossiculum terminale persistens ▪ Abnormal resegmentation of proatlas centrum – Os avis ▪ Failed midline integration of basal dental segment – Bifid dens	▪ Failed midline integration of basioccipital primordium – Bifid clivus ▪ Basioccipital dysplasia – Basilar impression – Platybasia – Retroflexed dens – Basilar invagination – Basilar kyphosis	▪ Hyperplasia of hypochordal bow of proatlas – Third occipital condyle – Pre-basioccipital arch ▪ Hyperplasia of exoccipital sclerotome – Hypertrophic occipital condyle ▪ Non-resegmentation of proatlas (anterior homeotic transformation) – Atlas assimilation ▪ Posterior homeotic transformation	▪ Aplasia of hypochordal bow of C₁ – Aplasia and hypoplasia of anterior C₁ arch ▪ Aplasia/hypoplasia of lateral sclerotome – Posterior C₁ arch agenesis ▪ Combined hypochordal bow and lateral sclerotome dysplasia – Aplasia of lateral mass and anterior C₁ arch – Combined anterior and posterior C₁ arch defects – Bifid anterior and posterior C₁ arch

Basilar invagination

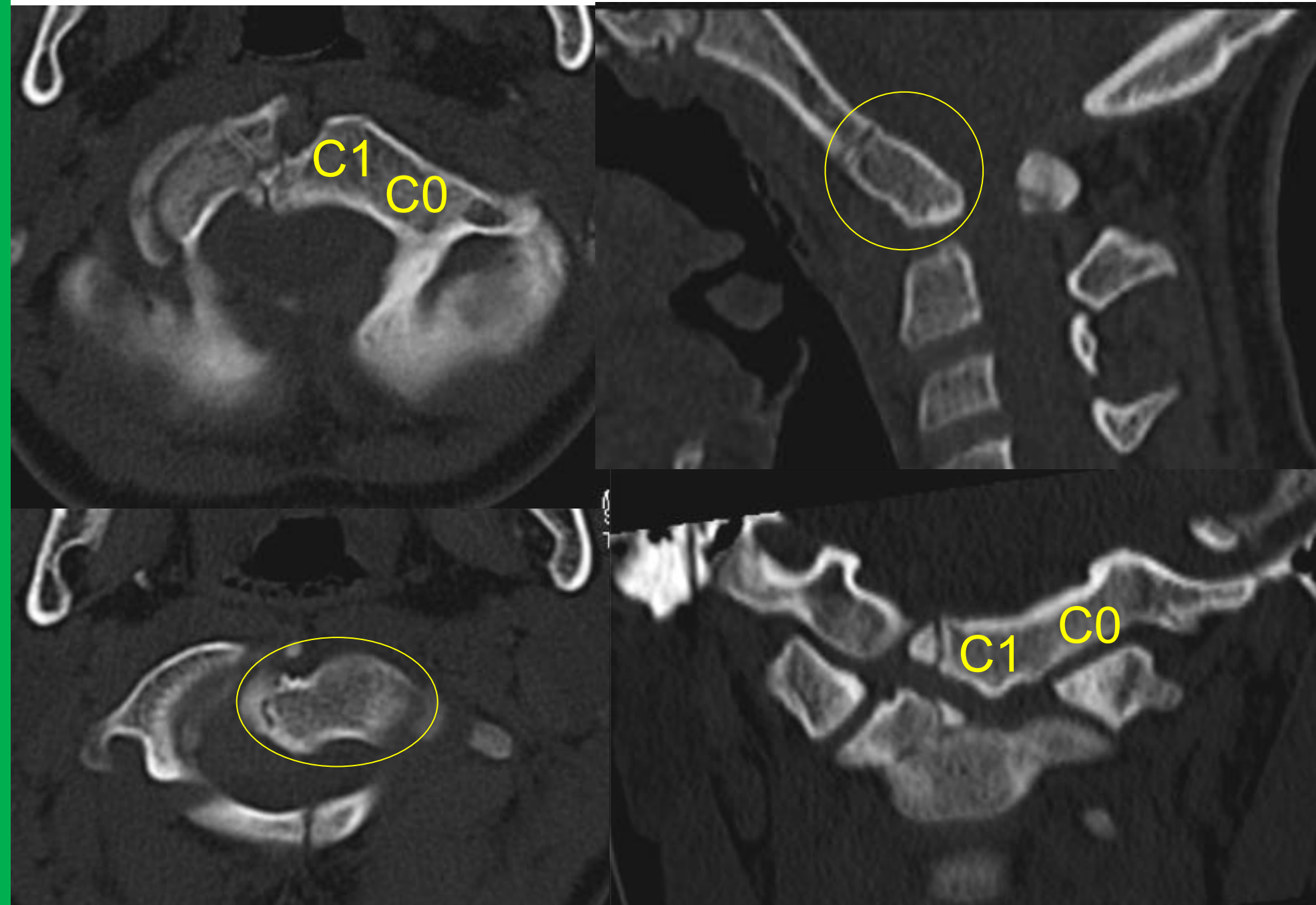
- Upward herniation of the margins of the foramen magnum into the posterior fossa
- Impingement of structures, such as the odontoid on the medulla oblongata with flexion of the head
- Abnormal development of C0/1/2
- **Basilar impression**: secondary form of invagination caused by softening of bone



Occipitalization of C1

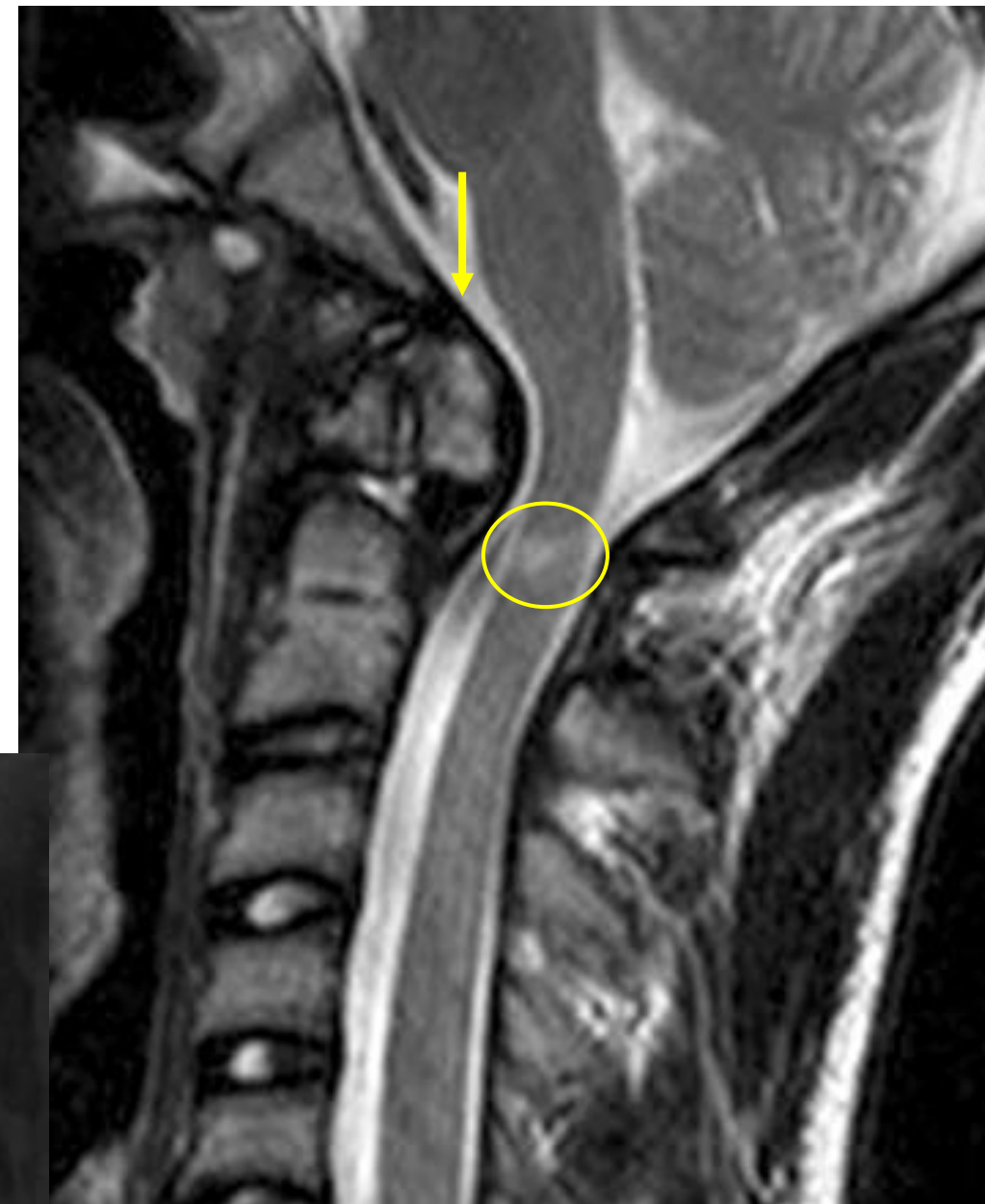
Fusion of the atlas to the occiput

Frequent association with foramen magnum stenosis



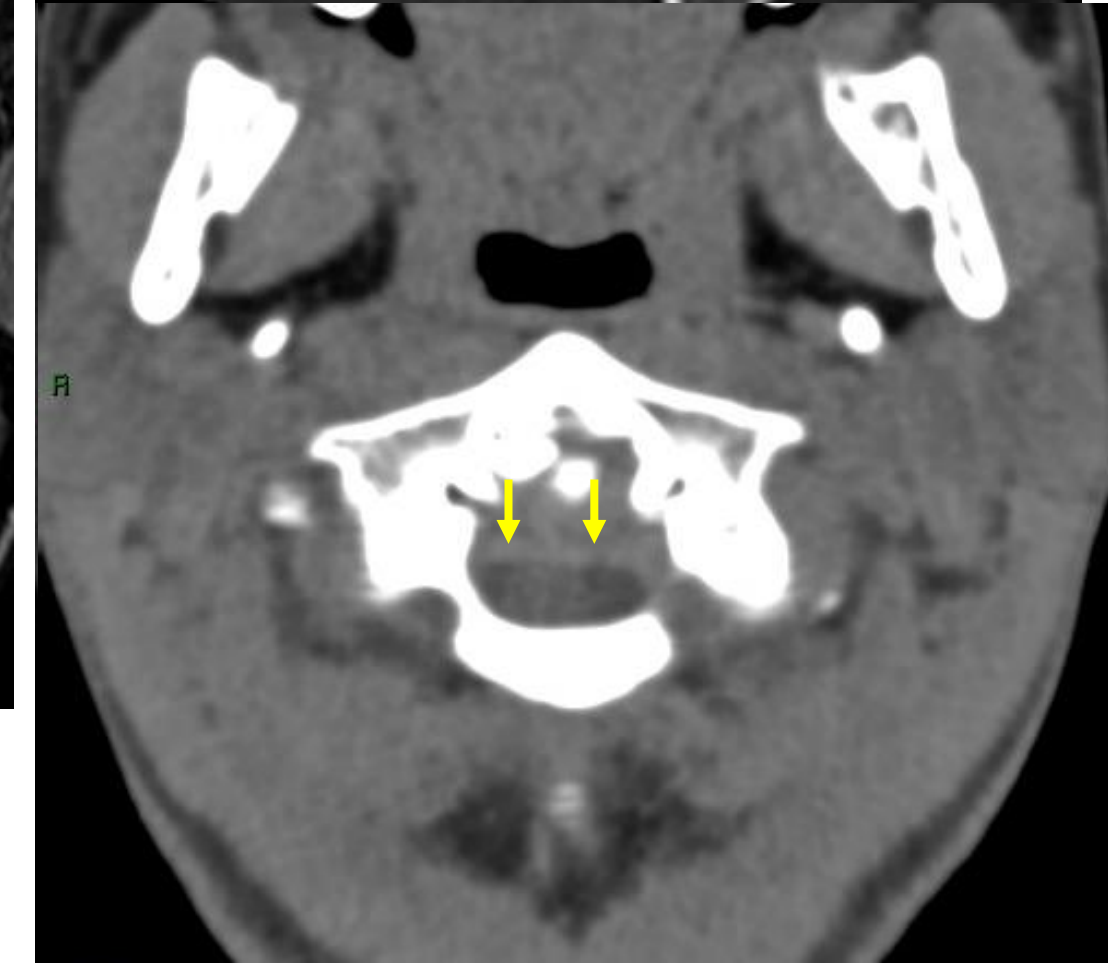
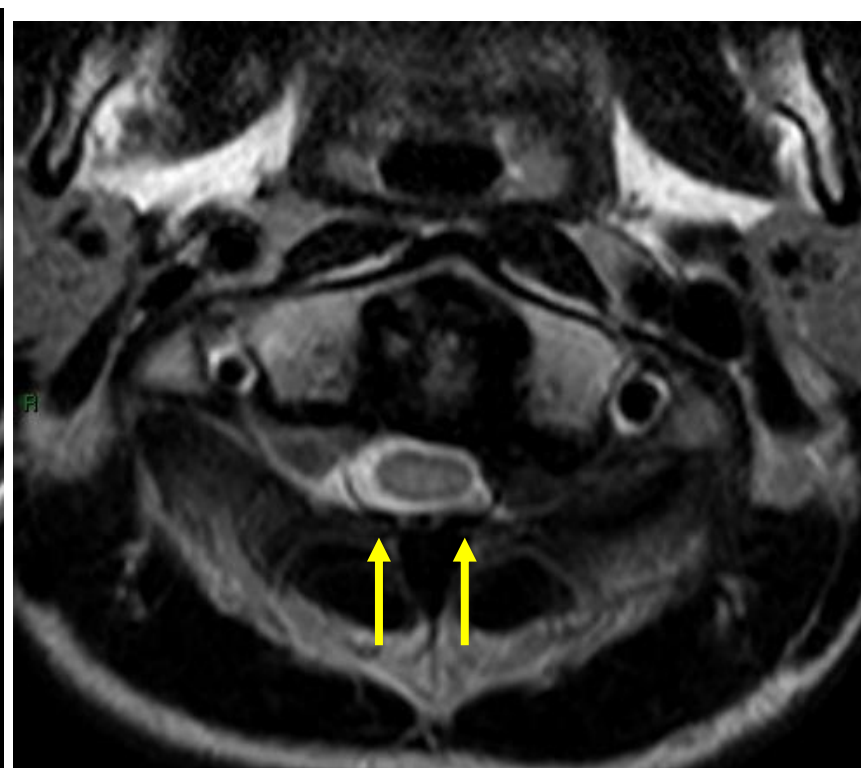
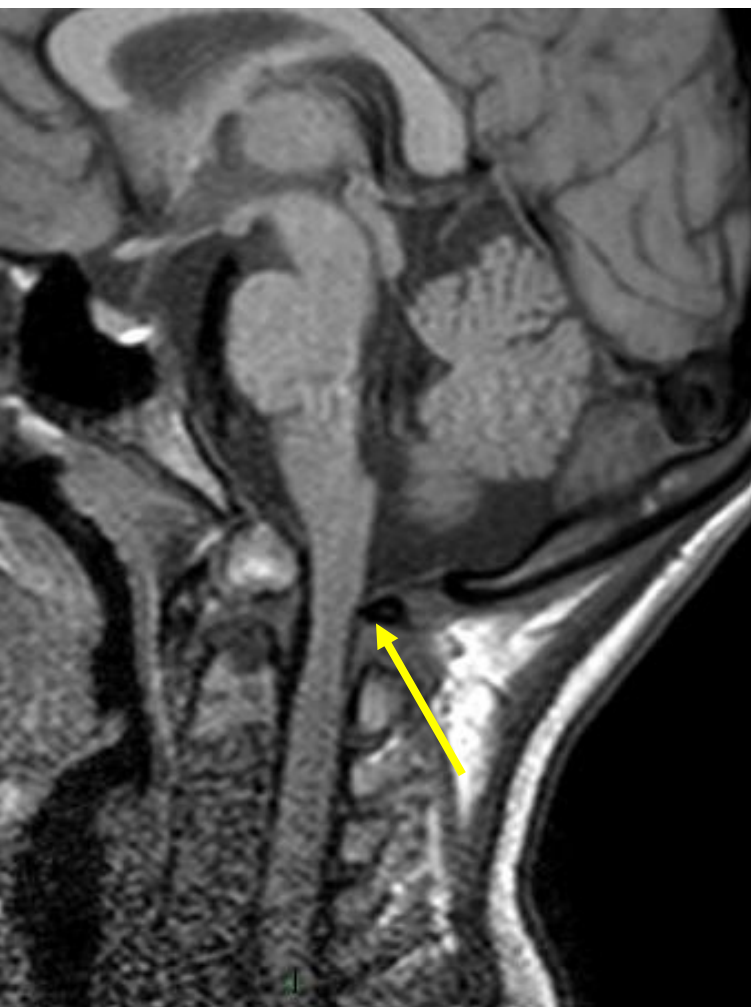
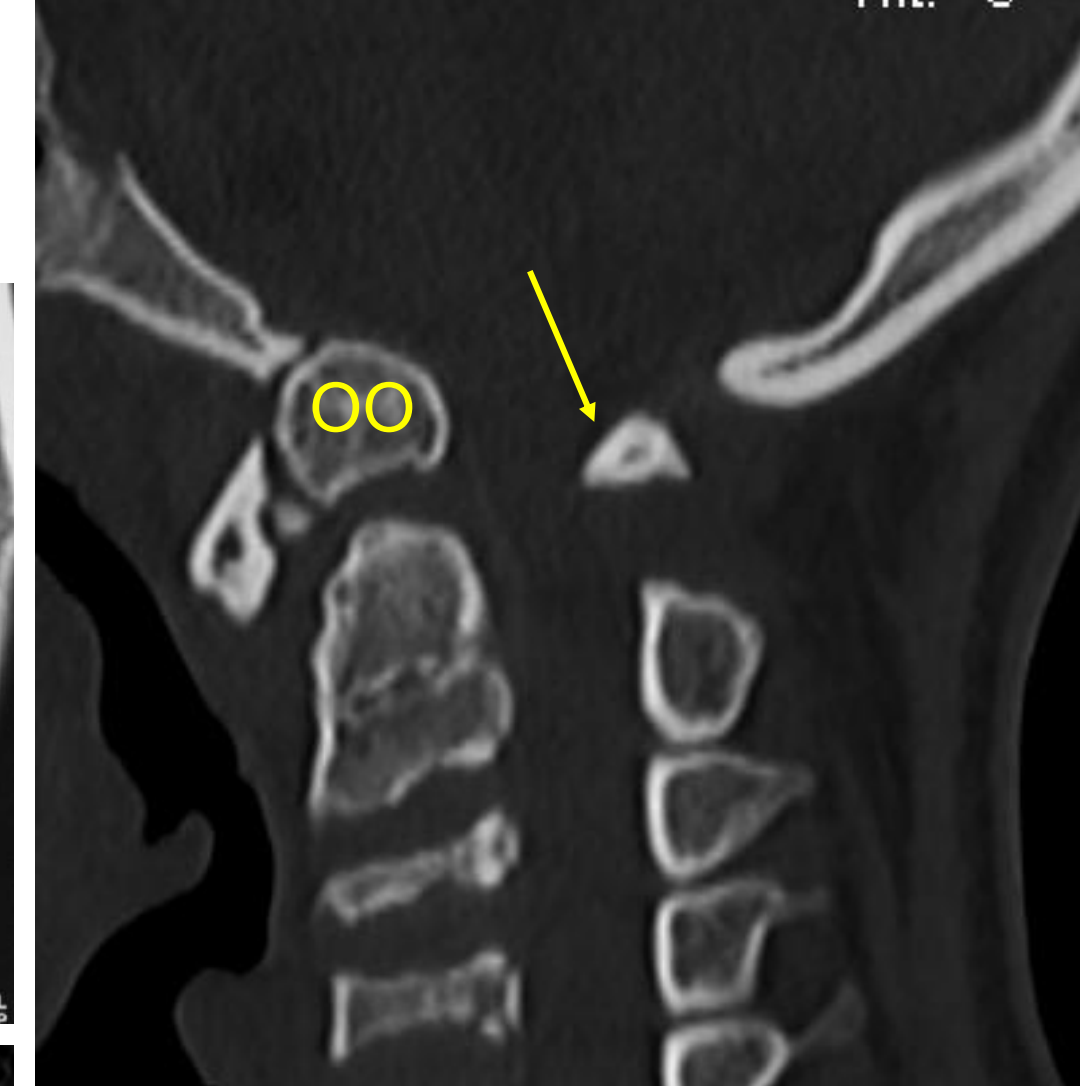
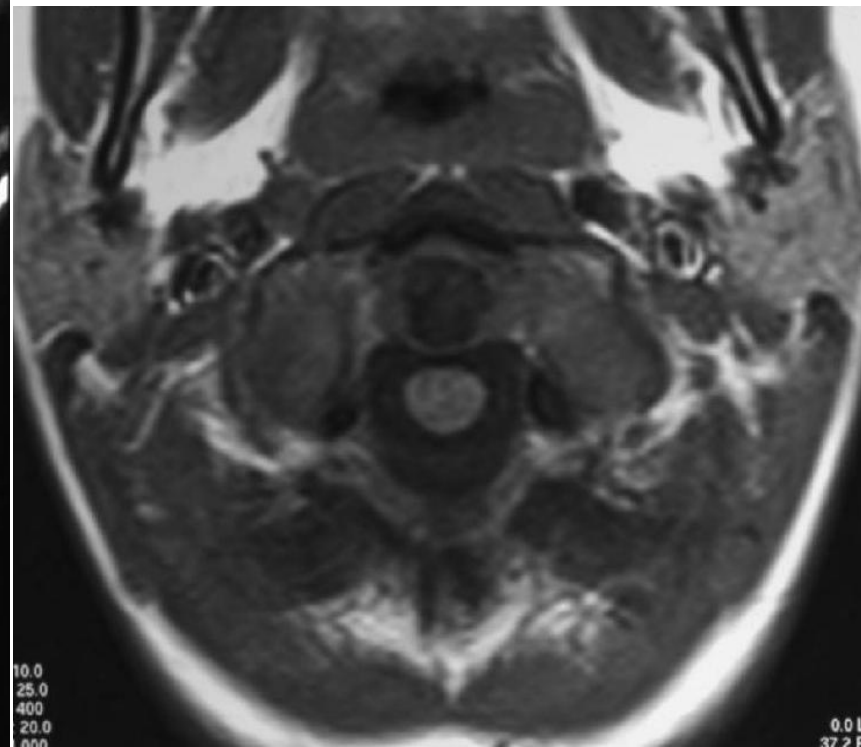
Os odontoideum

- Ossicle with smooth cortical margins and no connection to the body of C2
- Congenital (centrum of proatlas)
- Acquired (remote trauma with chronic nonunited dens fracture)
- Intrinsically unstable, possible acute neurologic findings



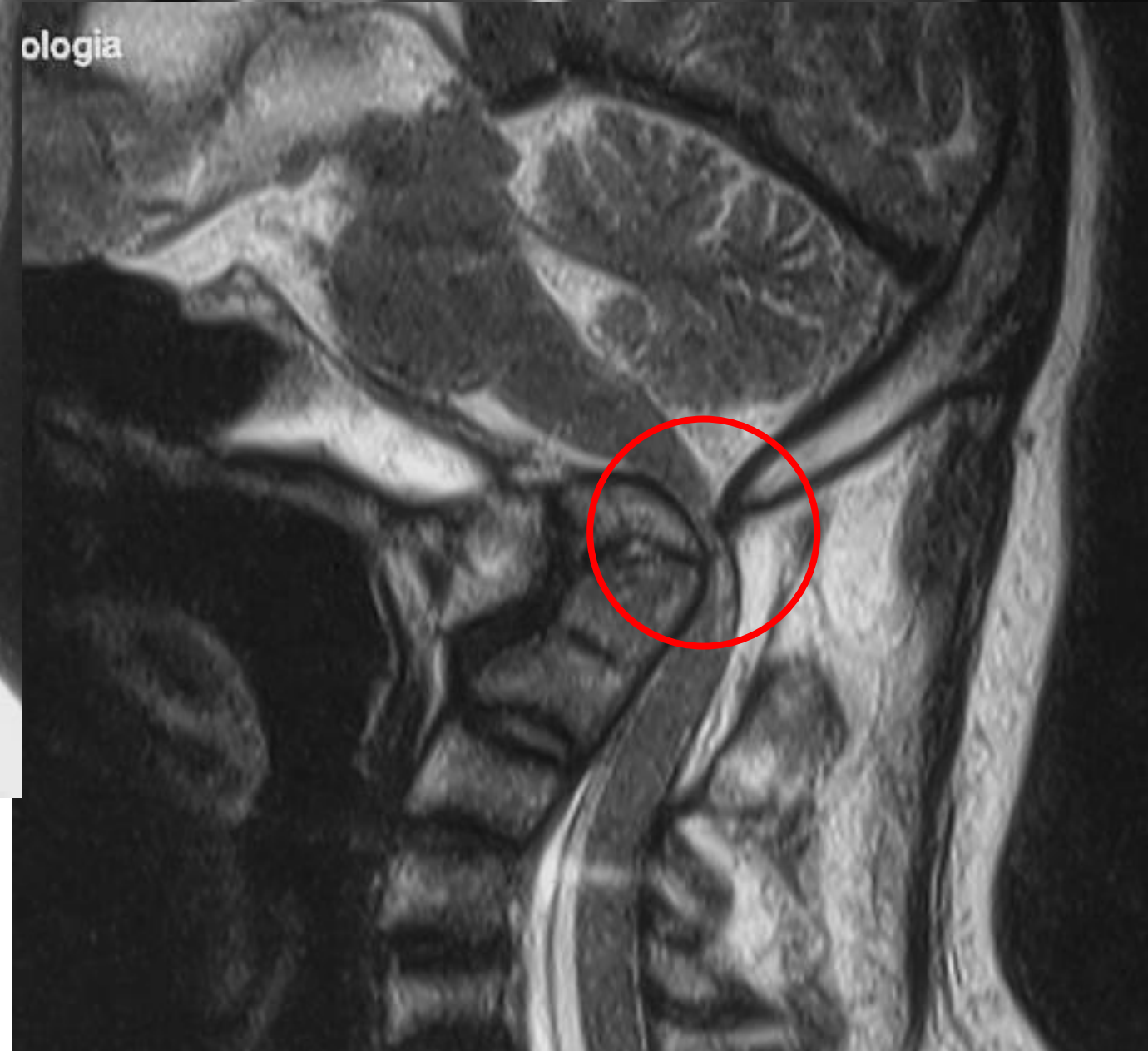
Klimo P et al.
J Neurosurg Spine 9:332-342, 2008

Anterior dislocation

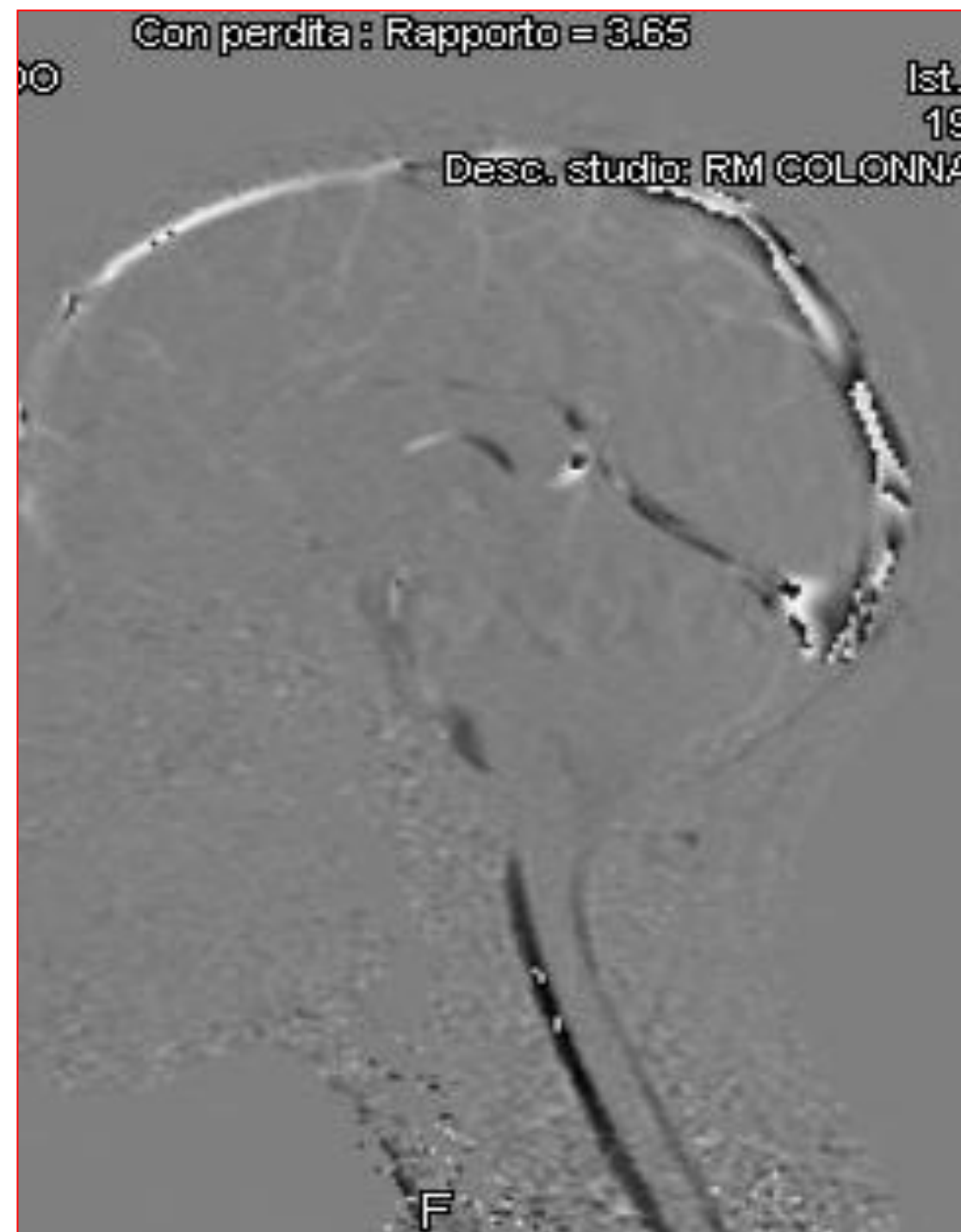


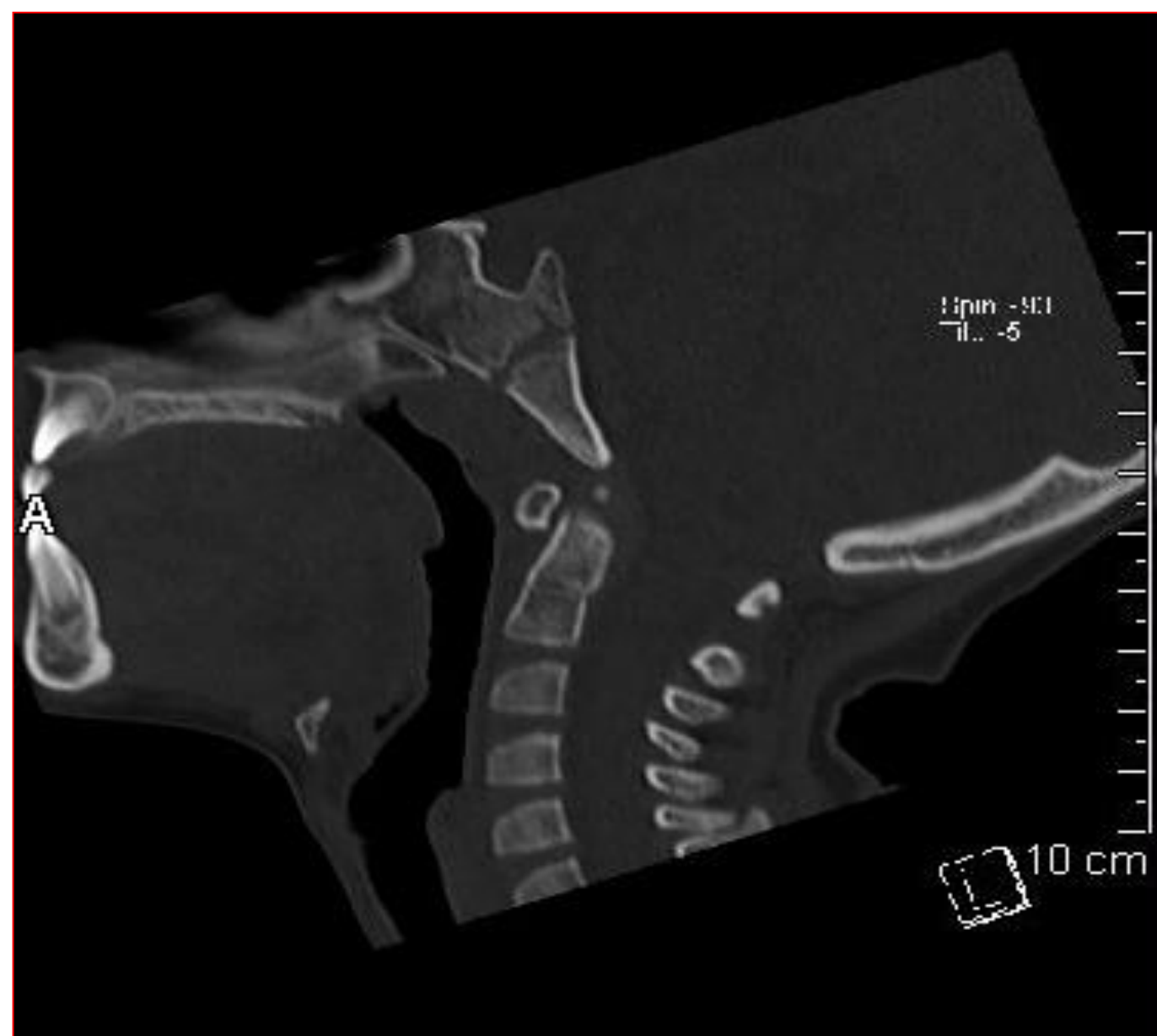
Os odontoideum

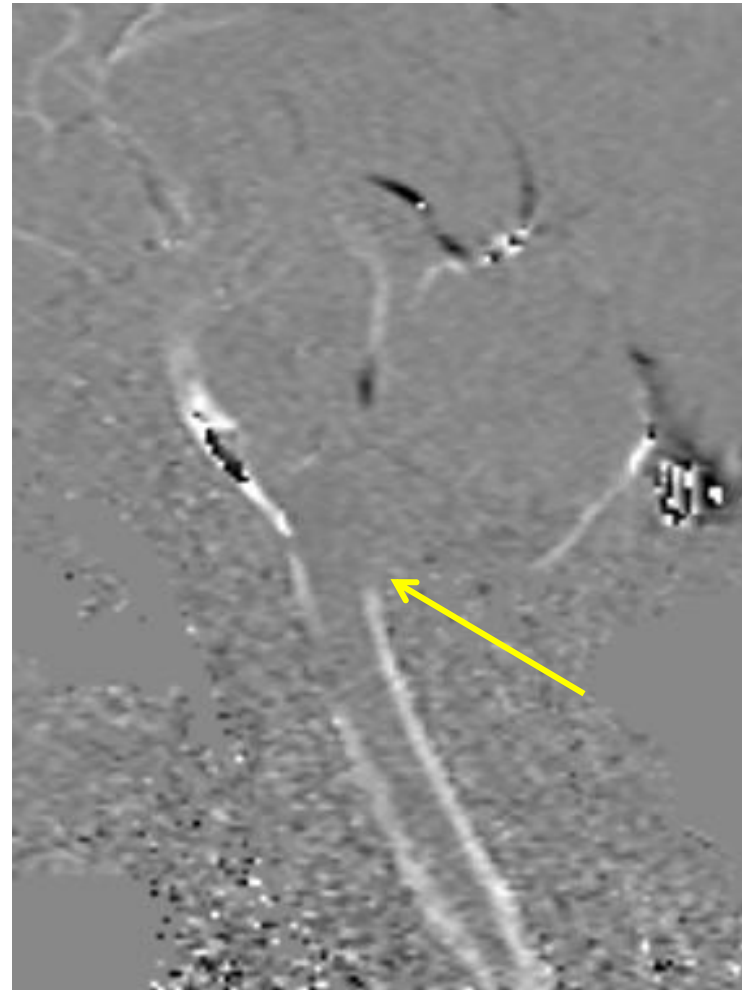
Posterior dislocation



HEADACHE and NEUROVEGETATIVE DISORDERS

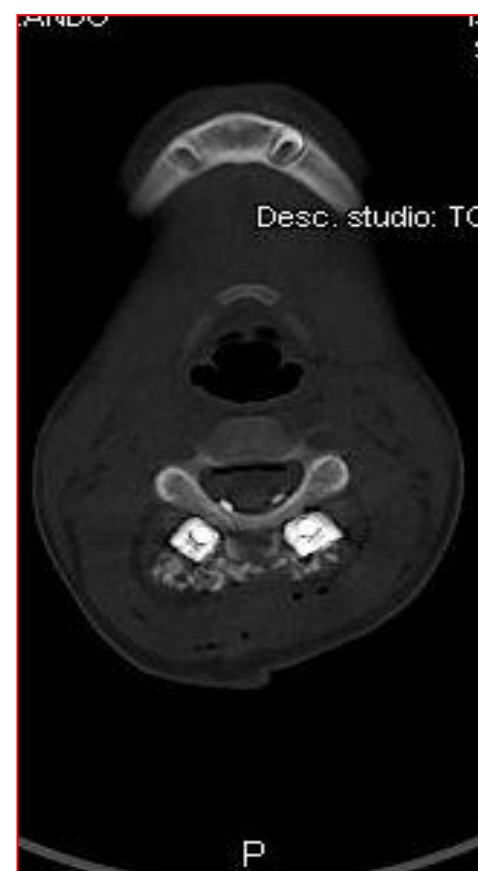
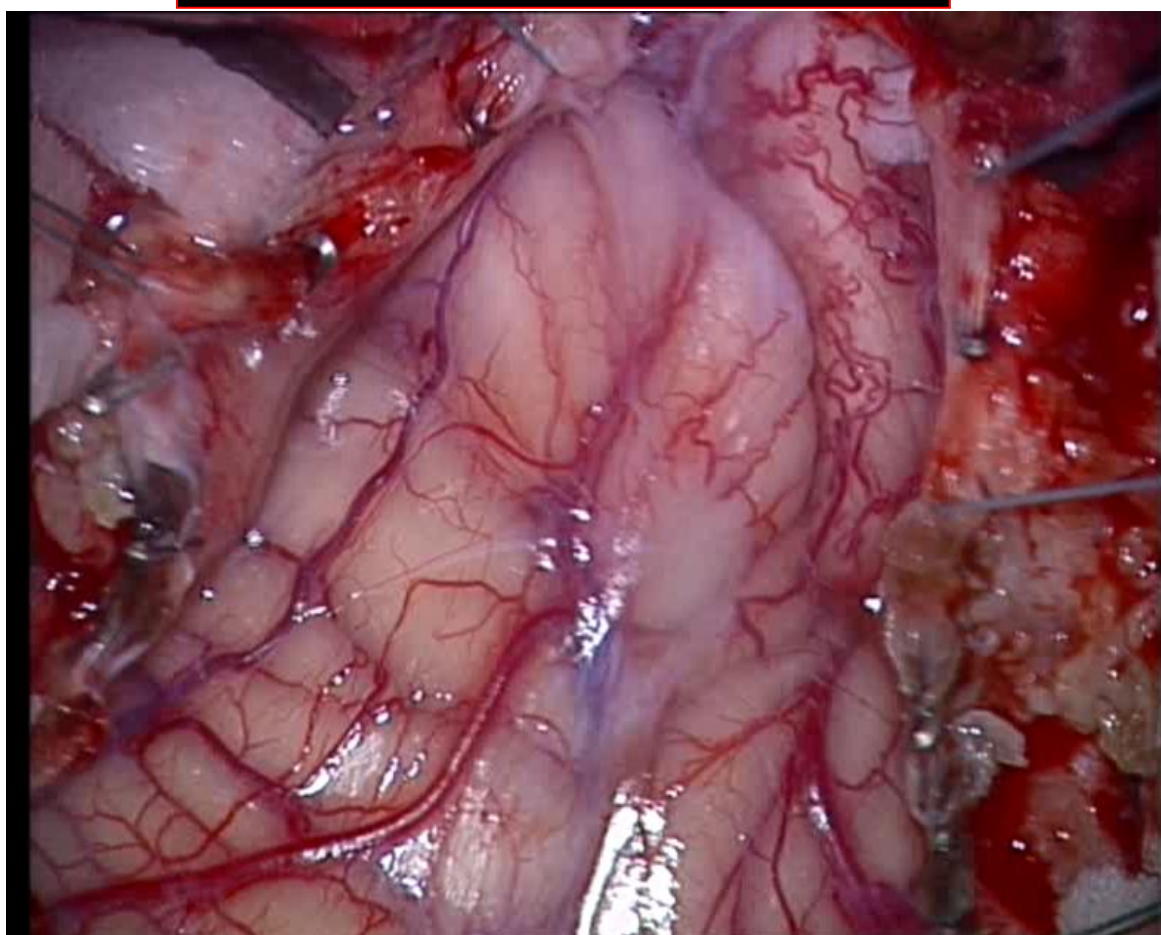


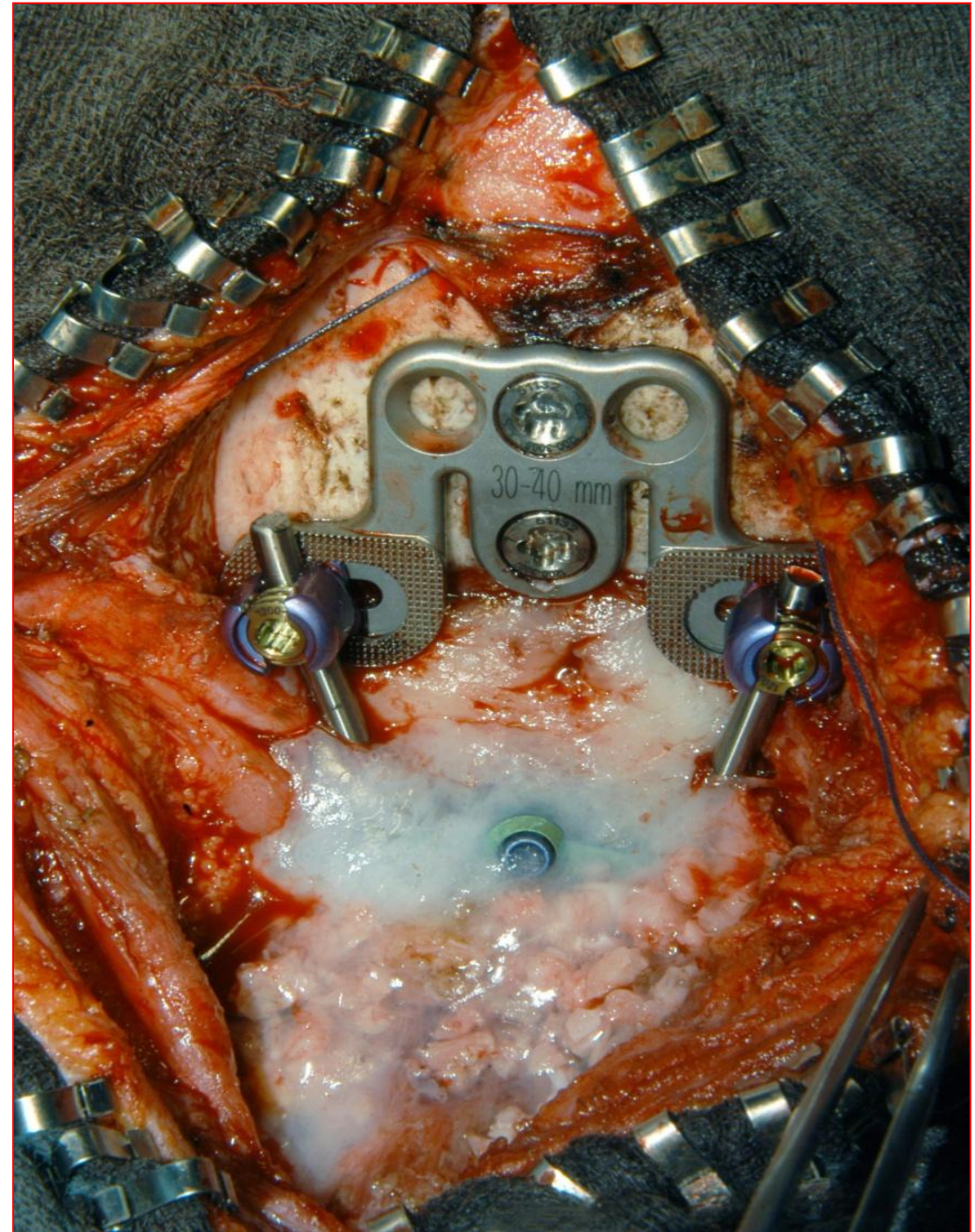




Severely reduced CSF flow in the posterior subarachnoid spaces at the foramen magnum









Syndrome	Cervical Spine Involvement
Aarskog syndrome	Cervical vertebral anomalies including hypoplasia and synostosis of 1 or more cervical vertebrae
Apert syndrome	Fusion of cervical vertebrae usually at C5 to C6
Atelosteogenesis, type 1	Abnormally segmented and fused cervical vertebrae
Campomelic dysplasia	Short and somewhat flat cervical vertebrae
Cervico-oculo-acoustic syndrome (Wildervanck syndrome)	Klippel-Feil anomaly (fusion of 2 or more cervical vertebrae, torticollis)
Chondrodysplasia punctata	Subaxial canal stenosis, coronal clefts or hypoplasia of vertebral bodies, upper cervical instability from os odontoideum
Diastrophic dysplasia	Hypoplasia of cervical vertebral bodies with kyphosis and subluxation, spina bifida occulta from C3 to C4 to upper thoracic vertebrae
22q11.2 deletion syndrome	Upper cervical instability from odontoid hypoplasia or os odontoideum, congenital C2 to C3 fusion
Cleidocranial dysostosis	Basilar impression with enlarged foramen magnum
Down syndrome (trisomy 21)	Upper cervical instability from odontoid hypoplasia, os odontoideum, and ligamentous laxity
Dyggve-Melchoir-Clausen syndrome	Odontoid hypoplasia, cervical spine instability, flattened vertebrae
Ehlers-Danlos syndrome IV	Atlantoaxial instability
Fetal alcohol syndrome	Cervical vertebral malformations
Fibrodysplasia ossificans progressive	Large posterior elements, tall narrow vertebral bodies, enlarged pedicles, fusion of the facet joints, lateral masses, and spinous processes. May lead to severe chin-on-chest deformity
Frontometaphyseal dysplasia	Wide foramen magnum, cervical vertebral anomalies, wide interpedicular distance
Goldenhar syndrome	Occipitalization of the atlas, fusion of cervical spine, odontoid hypoplasia, and basilar impression
Hajdu-Cheney syndrome	Cervical vertebrae osteolysis; cervical spine instability
Hurler syndrome	Odontoid hypoplasia; atlantoaxial instability
Klippel-Feil syndrome	Congenital fusion of cervical vertebrae, occipitalization of the atlas, stenosis of cervical spine, instability
Kniest dysplasia	Atlantoaxial instability
Kozłowski spondylometaphyseal dysplasia	Odontoid hypoplasia, cervical spine instability
Larsen syndrome	Cervical kyphosis, which can be progressive
Marfan syndrome	Atlantoaxial and other cervical vertebrae subluxation, focal kyphosis, absence of cervical lordosis, basilar impression
Maroteaux-Lamy mucopolysaccharidosis syndrome	Odontoid hypoplasia, cervical myopathy due to thickening of the cervical dura mater
Marshall-Smith syndrome	Instability of the craniocervical junction with severe spinal stenosis
Metaphyseal chondrodysplasia	Atlantoaxial instability due to ligamentous laxity
Metatropic dysplasia	Odontoid hypoplasia, C1 to C2 subluxation
Morquio syndrome	Odontoid hypoplasia, C1 to C2 subluxation, C2 to C3 subluxation
Mucopolysaccharidoses	Odontoid hypoplasia, abnormal soft tissue thickening C1 to C2
Multiple epiphyseal dysplasia	Upper cervical instability due to os odontoideum
Osteopathin striata	Kyphosis due to vertebral dysplasia and hypotonia

Review of Cervical Spine Anomalies in Genetic Syndromes.
 McKay, Scott; Al-Omari, Ali; Tomlinson, Lauren; Dormans, John
 Spine. 37(5):E269-E277, March 01, 2012.

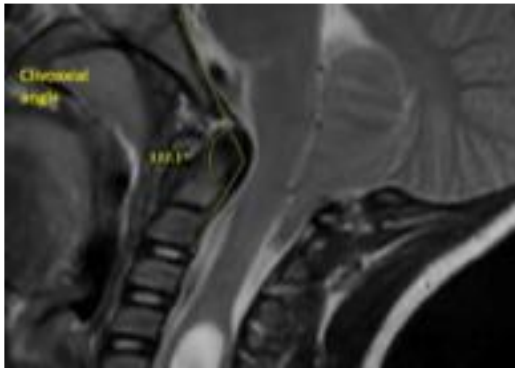
Syndrome	Cervical Spine Involvement
Osteopoikilosis	Cervical central canal stenosis
Pseudoachondroplasia	Odontoid aplasia or hypoplasia, C1 to C2 instability
Pyknodysostosis	Kyphosis due to C2 and/or C3 spondylolysis
Spondylocarpotarsal synostosis syndrome	Failure of vertebral segmentation, odontoid hypoplasia, cervical spine instability
Spondyloepimetaphyseal dysplasia	Upper cervical instability due to odontoid hypoplasia
Spondyloepiphyseal dysplasia congenital	Upper cervical instability and underlying stenosis
Weaver syndrome	Instability of the upper cervical spine

Craniovertebral Junction Instability in the Setting of Chiari Malformation

Yosef M. Dastagirzada, MD^{a,*}, David B. Kurland, MD, PhD^a,
Todd C. Hankinson, MD^b, Richard CE. Anderson, MD^a

Genetic syndromes associated with
craniovertebral junction instability^{8,59}

- Ehlers-Danlos syndrome
- Marfan syndrome
- Down syndrome
- VACTERL
- Klippel-Feil syndrome
- Achondroplasia
- Down syndrome
- Osteogenesis imperfecta
- Skeletal dysplasia
- Goldenhar syndrome
- Crouzon syndrome
- Conradi syndrome
- Morquio syndrome
- Hurler syndrome
- Noonan syndrome
- Kniest syndrome



Osseous abnormalities associated with Chiari I
malformation and craniovertebral junction
instability

Abnormality	Definition
Retroflexed odontoid process	Odontoid process is angled toward the brainstem, can use pBC2 distance (>9 mm) ³³
Basilar invagination	Prolapse of odontoid into the foramen magnum ³⁴
Occipital condyle hypoplasia	Flattening of the condyles with elevation of the atlas-axis
Platybasia	Flattening of the skull base (Welcher basal angle >140°) ³⁵
Proatlas segmentation failures	Embryologic failure of segmentation (somite > vertebra); can affect foramen magnum, occipital condyles, cruciate/alar ligaments, lateral atlantal masses, posterior arch of atlas ³⁶
Basioccipital/clivus hypoplasia	Shortened clivus, associated with CHARGE syndrome ³⁷
Atlas assimilation	Partial or complete congenital fusion of the atlas with occiput

Radiographic lines used to assess for basilar invagination

Line	Landmarks	Basilar Invagination is Considered if...
Chamberlains	Hard palate to opisthion	Odontoid tip >5 mm above line
McGregor	Hard palate to most caudal part of occipital bone	Odontoid tip >7 mm above line
McRae's	Basion to opisthion (AP diameter of foramen magnum)	Odontoid tip is above line
Wackenheims (clivus canal line)	Along the clivus, extending into cervical canal	Odontoid tip transects line

The CXA—normally varying from 144 in flexion to 170 in extension

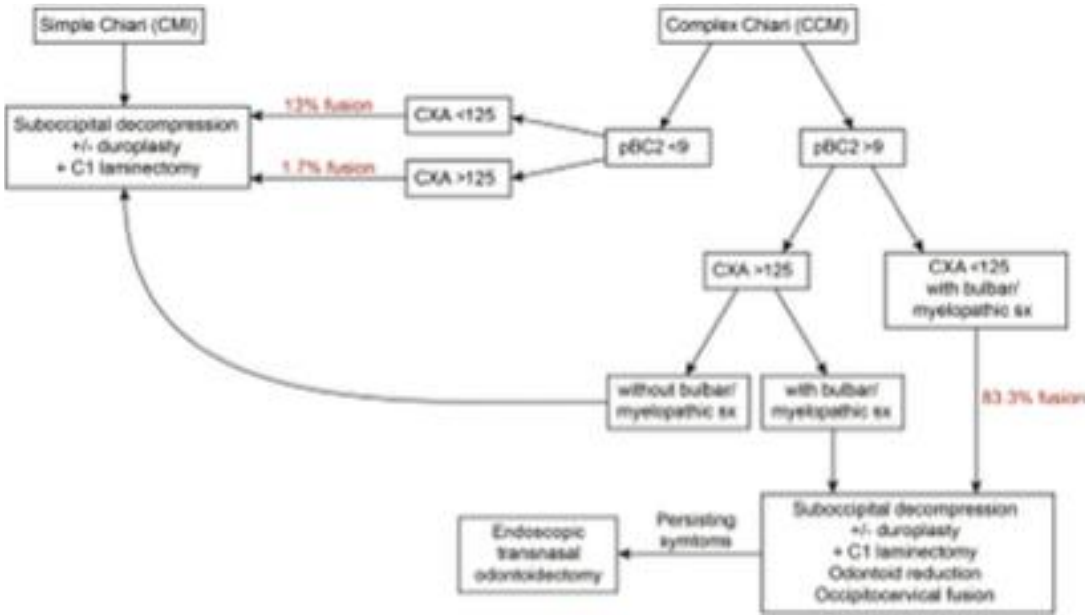


Fig. 5. Surgical algorithm for patients with complex Chiari I¹⁶

Prevalence of Craniovertebral Junction Instability with Chiari I Malformation

Overall, among surgical patients with CMI and syr- inx, the prevalence of coexisting CVJ instability is **extremely low**. In a multicenter study of 637 patients who underwent posterior fossa decompression for CMI and syrinx, **only 1.9% went on to have OCF for instability.**

CLINICAL CARE POINTS

Craniovertebral Junction Instability in the Setting of Chiari Malformation

Yosef M. Dastagirzada, MD^{a,*}, David B. Kurland, MD, PhD^a,
Todd C. Hankinson, MD^b, Richard CE. Anderson, MD^a

- Understanding the biomechanical properties and normal motion at the CVJ allow a greater understanding of abnormal pathologies and instability.
- Descriptions of atypical Chiari malformations (Chiari 1.5, complex Chiari, and so forth) have provided insights to Chiari phenotypes that have a higher risk for associated CVJ instability and may necessitate stabilization in addition to decompression.
- Osseous craniocervical abnormalities (retroflexed odontoid process, basilar invagination, platybasia, and so forth) often raise suspicion for CVJ instability among patients with CM1.
- Radiographic measurements including the Grabb-Oakes Index (pB-C2 line), CXA, and the occipital condyle-C2 sagittal vertical alignment (C-C2SVA), can help physicians to make decisions whether to include craniocervical arthrodesis in surgical management.
- Surgical goals for patients with CMI and coexisting CVJ instability are to relieve compression and stabilize the craniovertebral junction.

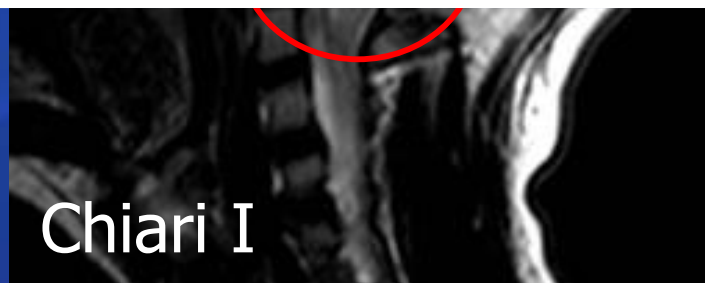
S. di Rubinstein-Taybi

anomalie osteo-legamentose della giunzione

chiari I / siringomielia

tethered cord

scoliosi



Chiari I



Siringomielia

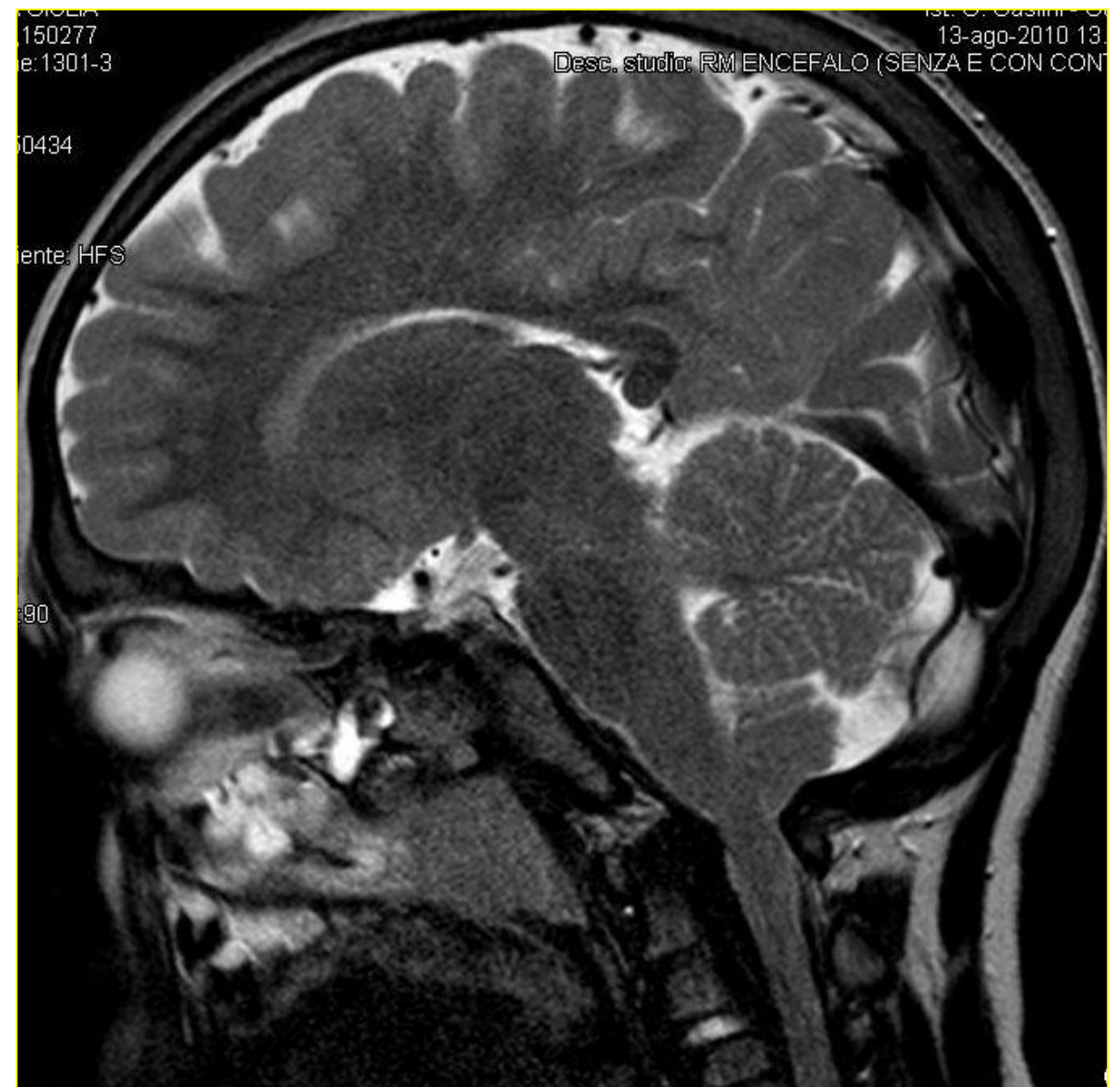
S. NOONAN



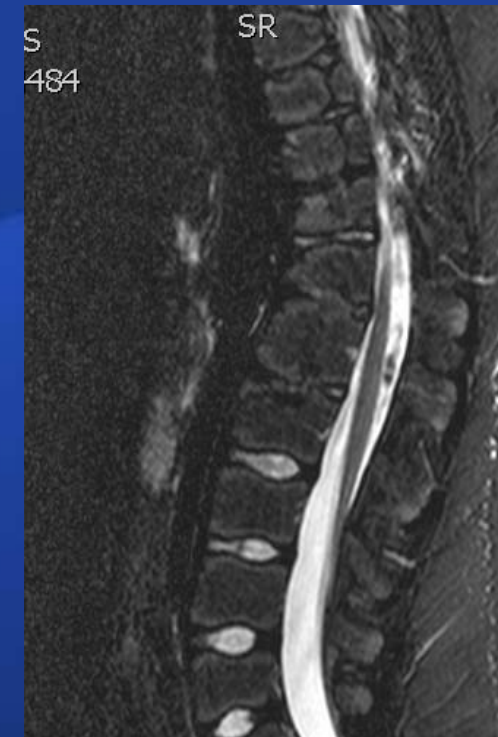
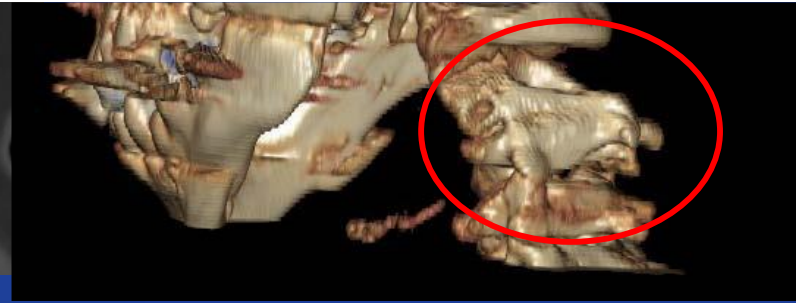
- Ipertelorismo
- Orecchie bassoposte
- Collo corto e pterigio
 - Cardiopatía
- Ritardo psico-motorio
 - Bassa statura
- Difetti di coagulazione
 - Ipoacusia

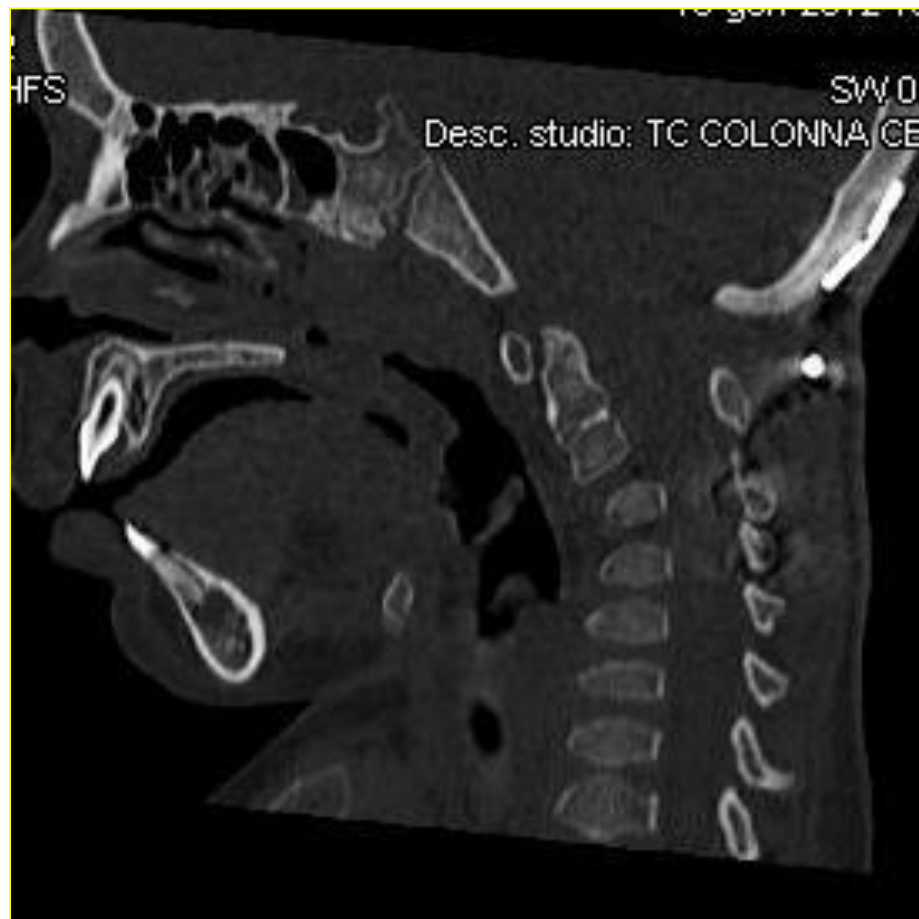


Female 7 yrs
Headache, increased by
Valsalva maneuver



Pseudoarthroplasia
Chiari 1.
Lumbar Kyphoscoliosis











Chiari I Malformation and Basilar Invagination in Fibrous Dysplasia: Prevalence, Mechanisms, and Clinical Implications

Kristen S Pan,¹ John D Heiss,² Sydney M Brown,¹ Michael T Collins,¹ and Alison M Boyce¹

¹Skeletal Disorders and Mineral Homeostasis Section, National Institute of Dental and Craniofacial Research, National Institutes of Health, Bethesda, MD, USA

²Surgical Neurology Branch, National Institute of Neurologic Disorders and Stroke, National Institutes of Health, Bethesda, MD, USA

ABSTRACT

Fibrous dysplasia (FD) is a mosaic disorder of benign fibro-osseous lesions, which may be associated with extraskeletal features as part of McCune-Albright syndrome (MAS). Cranial base abnormalities, including Chiari I malformation (CM1), in which the cerebellum extends below the foramen magnum, and secondary basilar invagination (BI), in which the odontoid prolapses into the posterior cranial fossa, are potentially serious complications of metabolic bone disorders. The purpose of this study was to determine the prevalence, natural history, and risk factors for CM1 and BI in patients with FD/MAS, and to determine mechanisms of cranial base deformities. Clinical and radiographic data from subjects in an FD/MAS natural history study were evaluated and compared to normal controls. In 158 patients with craniofacial FD, 10 (6.3%) cases of CM1 and 12 (7.6%) cases of BI were diagnosed. No cranial base abnormalities were identified in 10 control subjects. Craniomorphometric and volumetric analyses identified cranial constriction and cranial settling as the primary mechanisms of cranial base abnormalities, whereas intracranial hypertension was a contributing factor in a minority of subjects. Longitudinal analyses found progression of odontoid position with age, but no progression of tonsillar position. No endocrinopathies were associated with CM1. MAS endocrinopathies associated with BI included hyperthyroidism (odds ratio [OR] 12.0; 95% confidence interval [CI], 2.9 to 55.6; $p < 0.01$), precocious puberty (OR 5.6; 95% CI, 1.2 to 26.0; $p < 0.05$), and hypophosphatemia (OR 7.7; 95% CI, 1.9 to 27.0; $p < 0.01$). Scoliosis was associated with both CM1 (OR 4.8; 95% CI, 1.1 to 22.8; $p < 0.05$) and BI (OR = infinity; 95% CI, 4.7 to infinity; $p < 0.01$). This study successfully characterized cranial base abnormalities in FD/MAS and the pathophysiological connection between them. These findings support routine screening for cranial base abnormalities in patients with craniofacial FD, as well as aggressive management of contributory risk factors. © 2018 American Society for Bone and Mineral Research.

KEY WORDS: DISEASES AND DISORDERS OF/RELATED TO BONE; OSTEOMALACIA AND RICKETS; BONE-BRAIN-NERVOUS SYSTEM INTERACTIONS; SYSTEMS BIOLOGY; BONE INTERACTORS

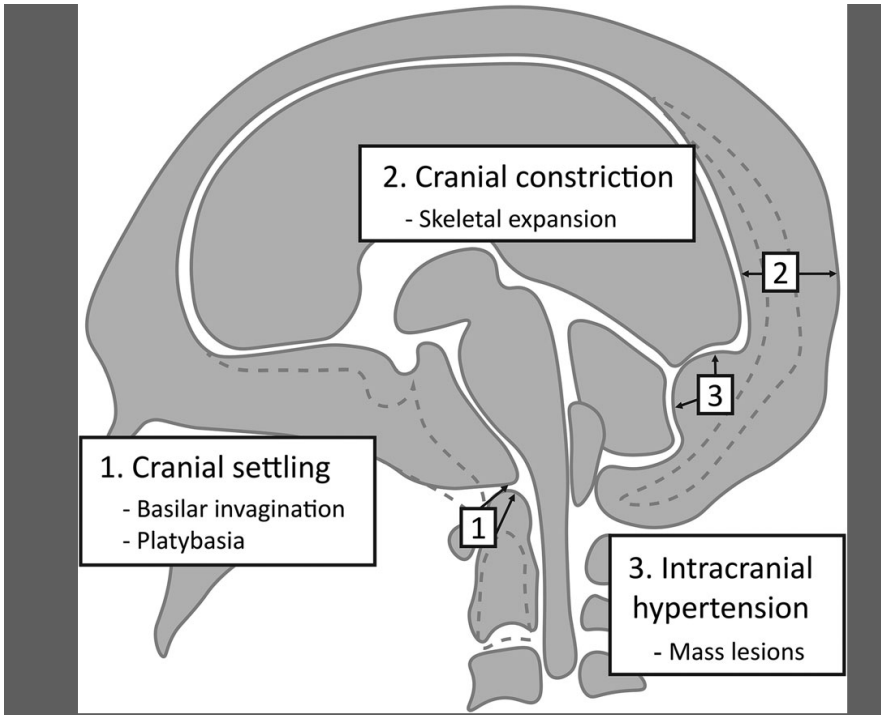
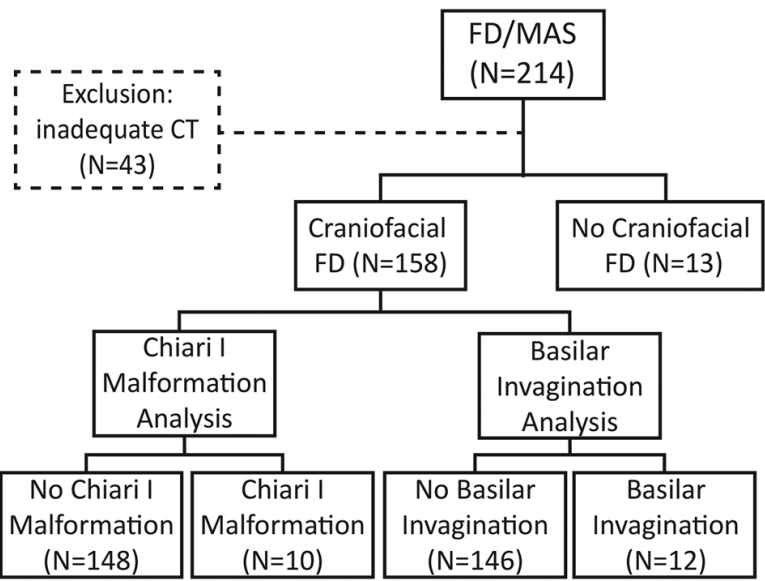
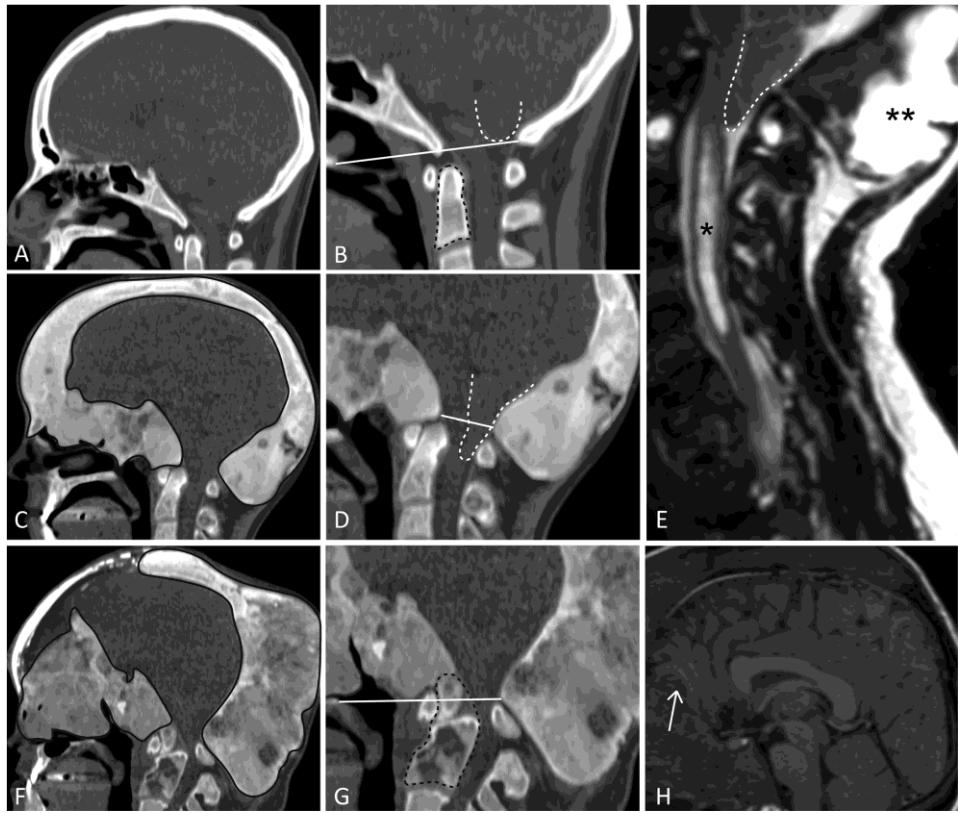
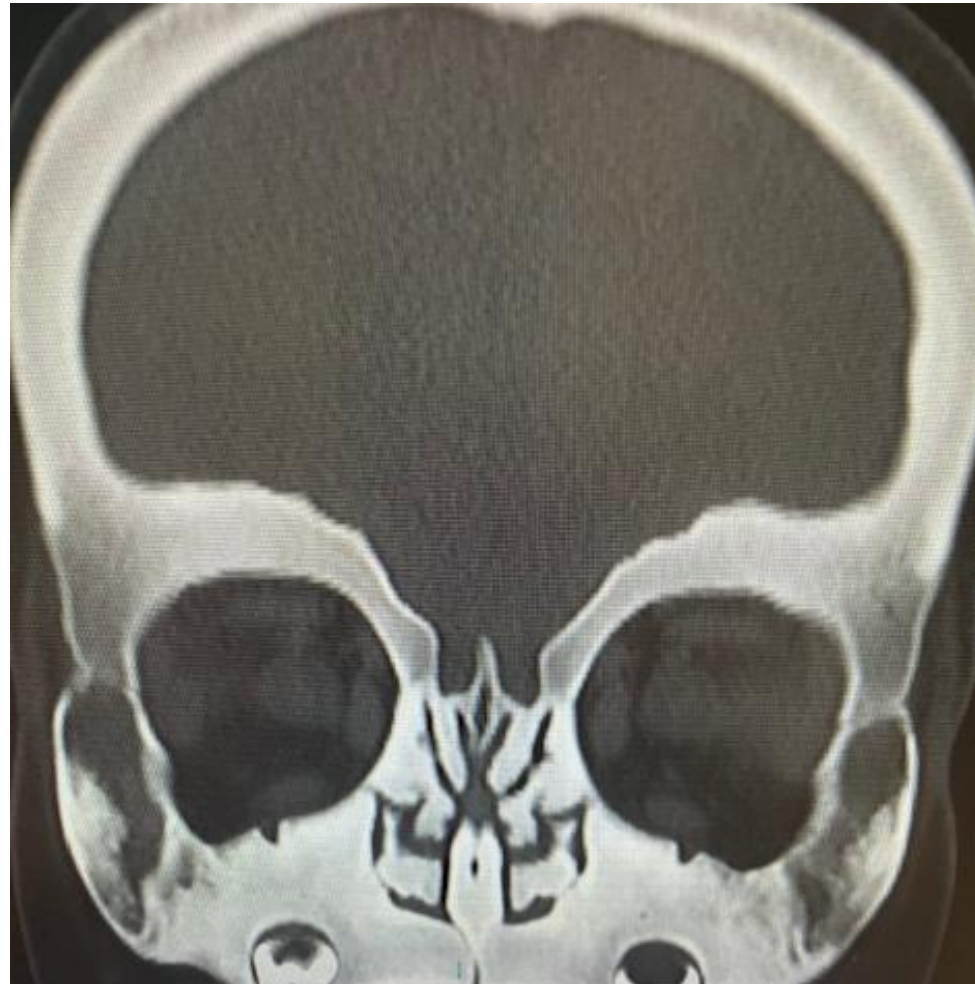
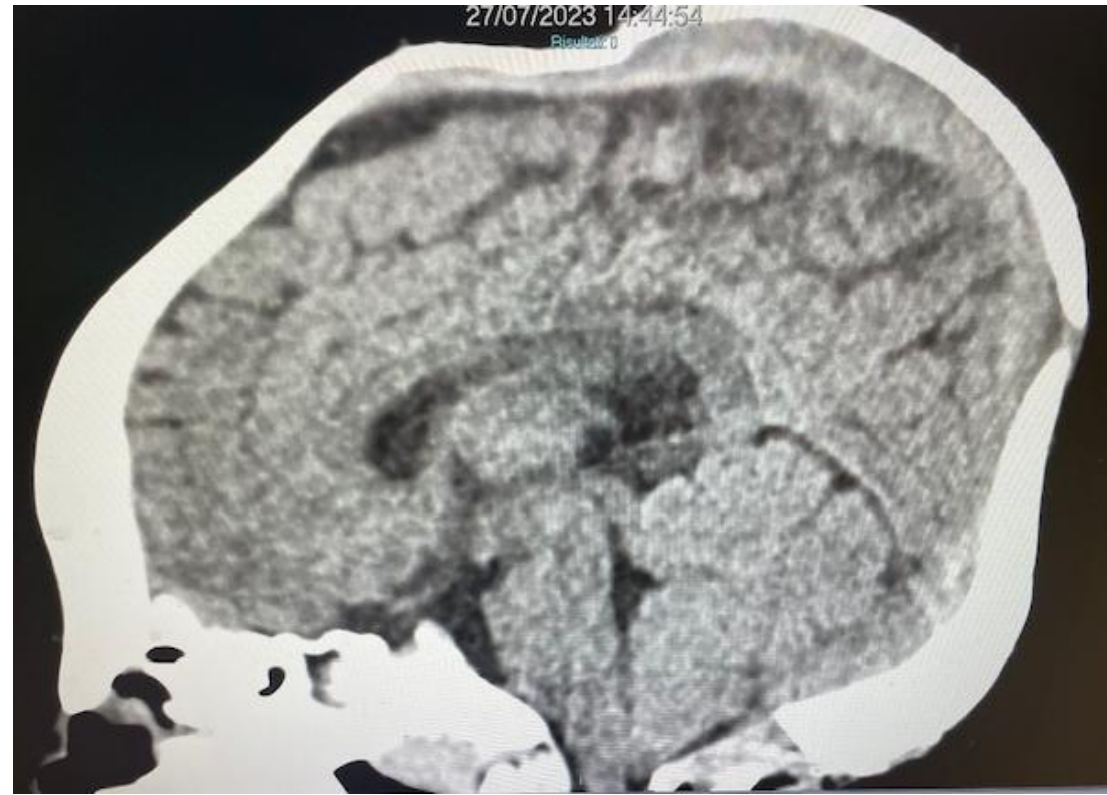
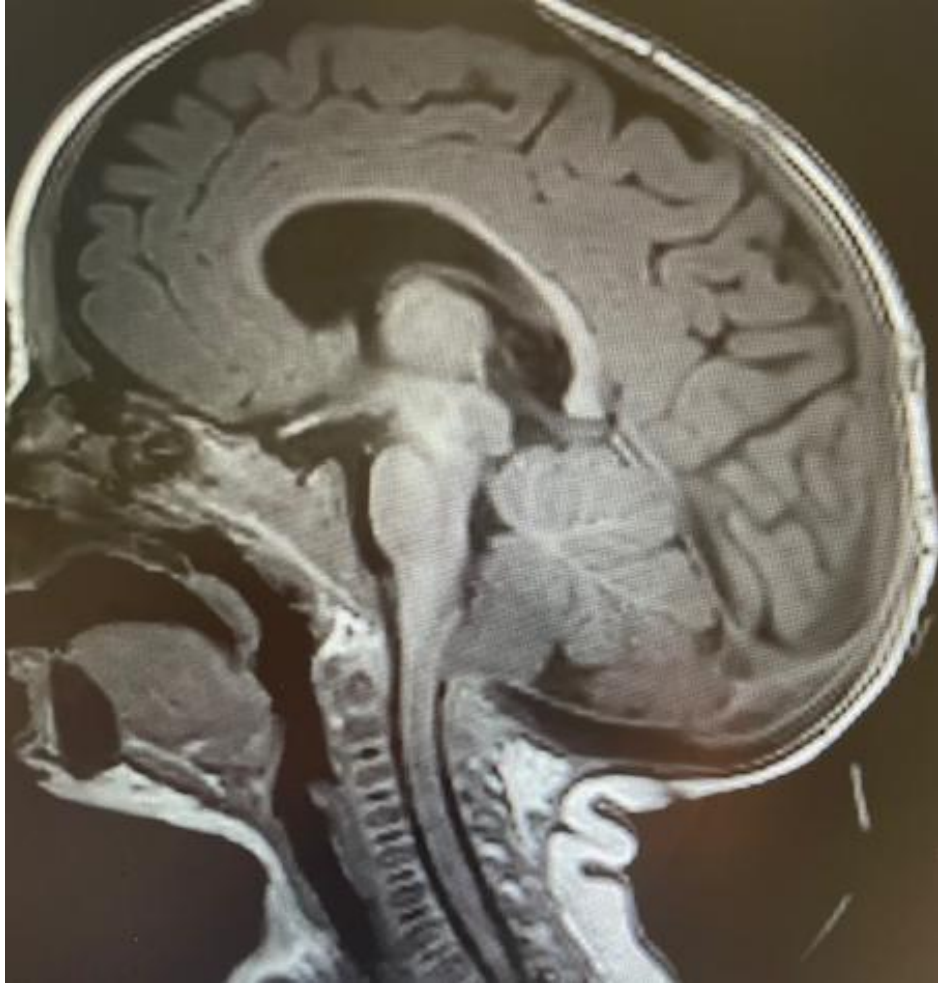


Fig. 4. Potential mechanisms of Chiari I malformation in craniofacial FD.



Syndrome of occipitoatlantoaxial hypermobility, cranial settling, and Chiari malformation Type I in patients with hereditary disorders of connective tissue

THOMAS H. MILHORAT, M.D.,¹ PAOLO A. BOLOGNESE, M.D.,¹ MISAO NISHIKAWA, M.D.,¹ NAZLI B. McDONNELL, M.D., PH.D.,² AND CLAIR A. FRANCOMANO, M.D.³

¹Department of Neurosurgery, The Chiari Institute, North Shore-Long Island Jewish Health System, Manhasset, New York; ²Medical Genetics, NIH National Institute on Aging; and ³Harvey Institute for Medical Genetics, Greater Baltimore Medical Center, Baltimore, Maryland

Object. Chiari malformation Type I (CM-I) is generally regarded as a disorder of the paraxial mesoderm. The authors report an association between CM-I and hereditary disorders of connective tissue (HDCT) that can present with lower brainstem symptoms attributable to occipitoatlantoaxial hypermobility and cranial settling.

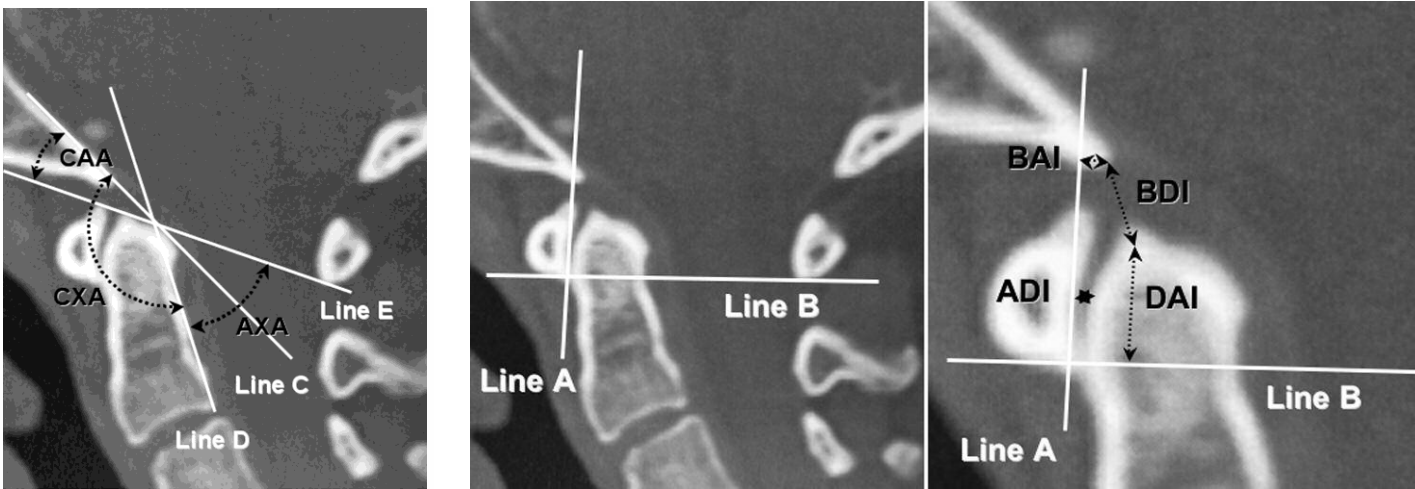
Methods. The prevalence of HDCT was determined in a prospectively accrued cohort of 2813 patients with CM-I. All patients underwent a detailed medical and neuroradiological workup that included an assessment of articular mobility. Osseous structures composing the craniocervical junction were investigated morphometrically using reconstructed 3D computed tomography and plain x-ray images in 114 patients with HDCT/CM-I, and the results were compared with those obtained in patients with CM-I (55 cases) and healthy control individuals (55 cases).

Results. The diagnostic criteria for Ehlers–Danlos syndrome and related HDCT were met in 357 (12.7%) of the 2813 cases. Heritability was generally compatible with a pattern of autosomal dominant transmission with variable expressivity. The diagnostic features of HDCT/CM-I were distinguished from those of CM-I by clinical stigmata of connective tissue disease, a greater female preponderance (8:1 compared with 3:1, $p < 0.001$), and a greater incidence of lower brainstem symptoms (0.41 compared with 0.11, $p < 0.001$), retroodontoid pannus formation (0.71 compared with 0.11, $p < 0.001$), and hypoplasia of the oropharynx (0.44 compared with 0.02, $p < 0.001$). Measurements of the basion–dens interval, basion–atlas interval, atlas–dens interval, dens–atlas interval, clivus–atlas angle, clivus–axis angle, and atlas–axis angle were the same in the supine and upright positions in healthy control individuals and patients with CM-I. In patients with HDCT/CM-I, there was a reduction of the basion–dens interval (3.6 mm, $p < 0.001$), an enlargement of the basion–atlas interval (3.0 mm, $p < 0.001$), and a reduction of the clivus–axis angle (10.8° , $p < 0.001$), clivus–atlas angle (5.8° , $p < 0.001$), and atlas–axis angle (5.3° , $p < 0.001$) on assumption of the upright position. These changes were reducible by cervical traction or returning to the supine position.

Conclusions. The identification of HDCT in 357 patients with CM-I establishes an association between two presumably unrelated mesodermal disorders. Morphometric evidence in this cohort—cranial settling, posterior gliding of the occipital condyles, and reduction of the clivus–axis angle, clivus–atlas angle, and atlas–axis angle in the upright position—suggests that hypermobility of the occipitoatlantal and atlantoaxial joints contributes to retroodontoid pannus formation and symptoms referable to basilar impression. (DOI: 10.3171/SPI-07/12/601)

Characteristics of patients with CM-I with and without HDCT*

Variable	Patient Group (%)	
	HDCT/CM-I	CM-I
total no. of cases	357	2456
mean age of symptom onset (yrs)	13.3 ± 4.6	22.0 ± 3.1
mean age at presentation (yrs)	25.7 ± 4.3	31.7 ± 3.9
sex		
male	39 (11)	647 (26)
female	318 (89)	1809 (74)
no prior op	232 (65)	1319 (54)
failed CM op	125 (35)	1137 (46)
classification of HDCT		
hypermobile EDS	149 (42)	
classic EDS	96 (27)	
arthrochalasia EDS	5 (1)	
Marfan syndrome	17 (5)	
MASS phenotype	19 (5)	
overlap disorder	71 (20)	



Conclusions. The identification of HDCT in 357 patients with CM-I establishes an association between two presumably unrelated mesodermal disorders. Morphometric evidence in this cohort—cranial settling, posterior gliding of the occipital condyles, and reduction of the clivus–axis angle, clivus–atlas angle, and atlas–axis angle in the upright position—suggests that hypermobility of the occipitoatlantal and atlantoaxial joints contributes to retroodontoid pannus formation and symptoms referable to basilar impression. (DOI: 10.3171/SPI-07/12/601)

RESEARCH REVIEW

Neurological and Spinal Manifestations of the Ehlers–Danlos Syndromes

FRASER C. HENDERSON SR.,* CLAUDIU AUSTIN, EDWARD BENZEL,
PAOLO BOLOGNESE, RICHARD ELLENBOGEN, CLAIR A. FRANCOMANO, CANDACE IRETON,
PETRA KLINGE, MYLES KOBY, DONLIN LONG, SUNIL PATEL, ERIC L. SINGMAN,
AND NICOL C. VOERMANS

Chiari malformation Type I (CMI) has been reported as a comorbid condition in hypermobile EDS (hEDS). The precise incidence of the CMI and EDS association is unknown, but the female to male ratio is higher (9:1) in the CMI and EDS subgroup than in the general CMI population (3:1).

ATLANTOAXIAL INSTABILITY

Epidemiology

Atlantoaxial instability (AAI) is a potential complication of all forms of EDS. Motor delay [Jelsma et al., 2013], headache associated with “connective tissue pathological relaxation” and quadri-paresis have all been attributed to ligamentous laxity and instability at the atlantooccipital, and atlantoaxial joints [Nagashima et al., 1981; Halko et al., 1995].

CRANIOCERVICAL INSTABILITY

Epidemiology

Cranio cervical instability (CCI) is recognized as a manifestation of ligamentous laxity in EDS [Nagashima et al., 1981; Milhorat et al., 2010]. Ligamentous laxity has been shown to result in neuraxial injury [Lindenburg and Freytag, 1970; Henderson et al., 1993; Menezes and Traynelis, 2008].

Diagnostic Exam for CVJ Instability

Imaging techniques

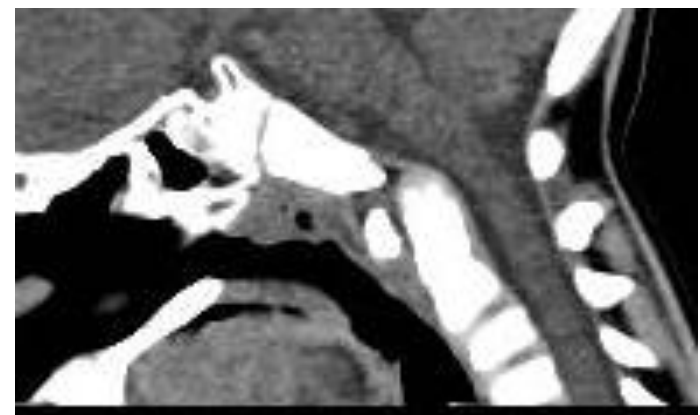
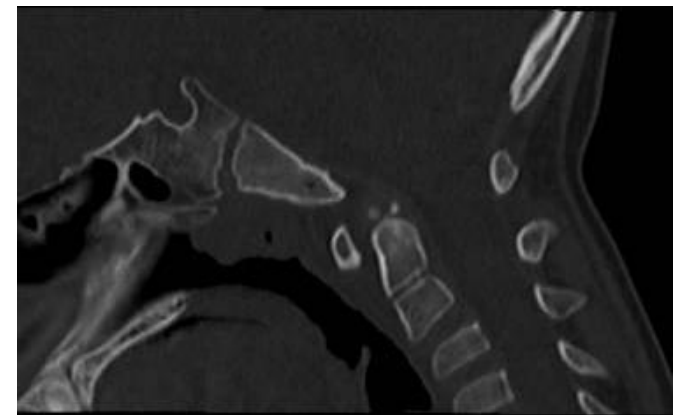
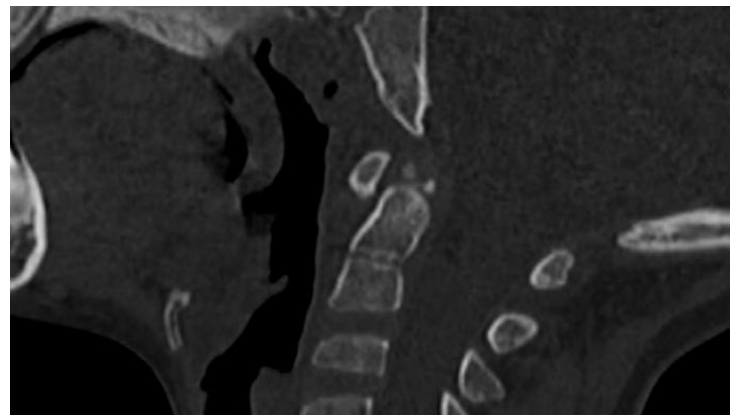
- Plain radiographs
(static - dynamic)

evaluate specific distances
between structures of the CCJ



- CT (static - dynamic)

optimally defines the
osseous structures

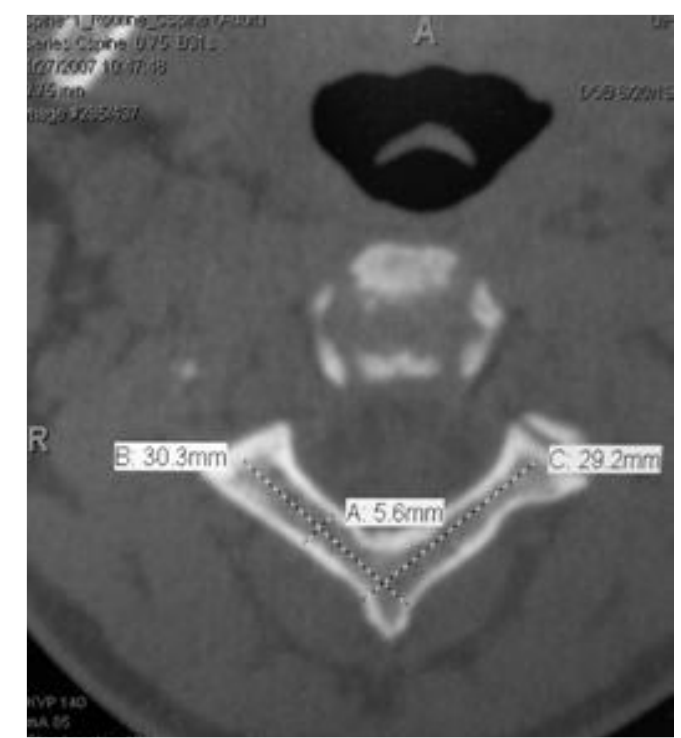
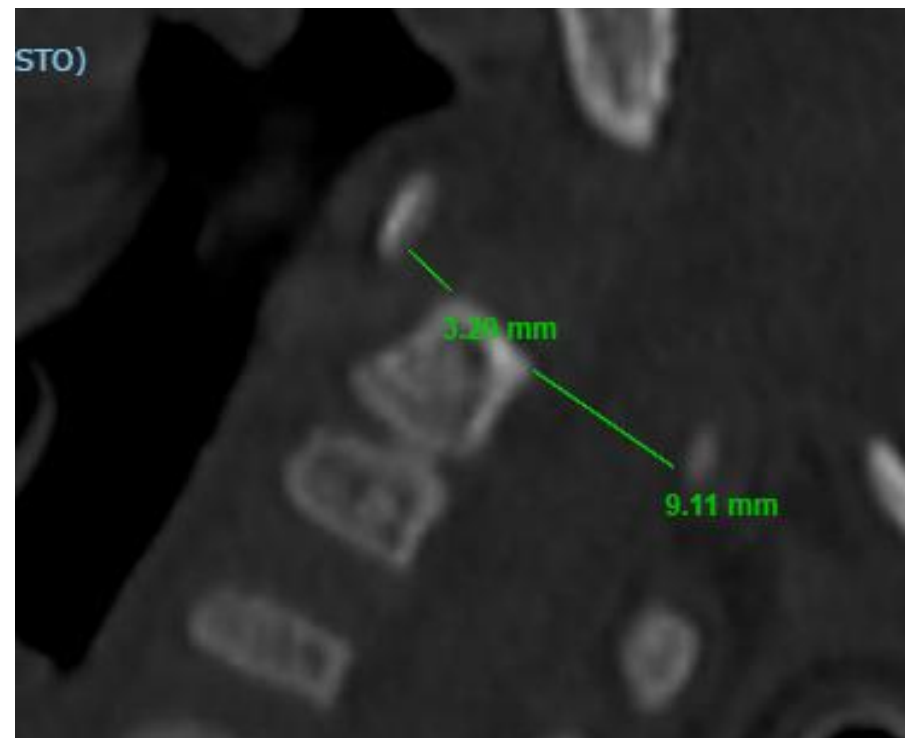
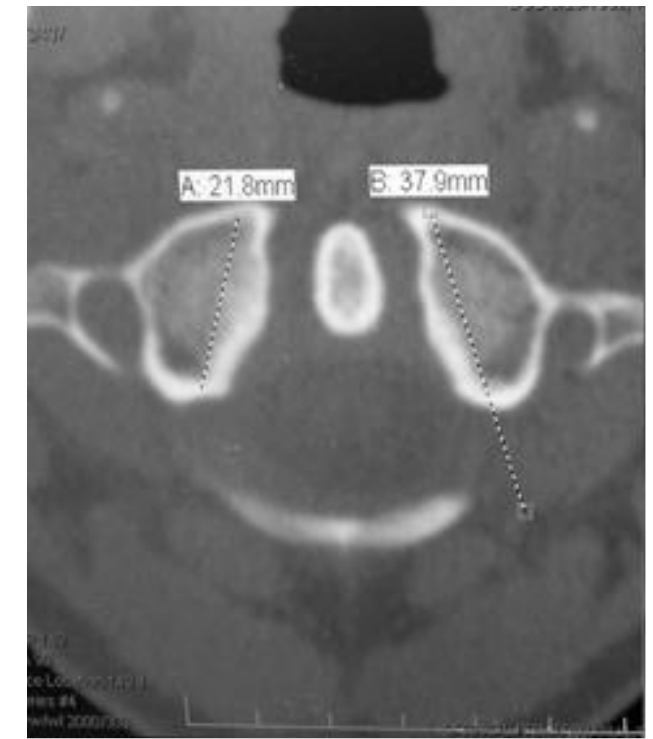
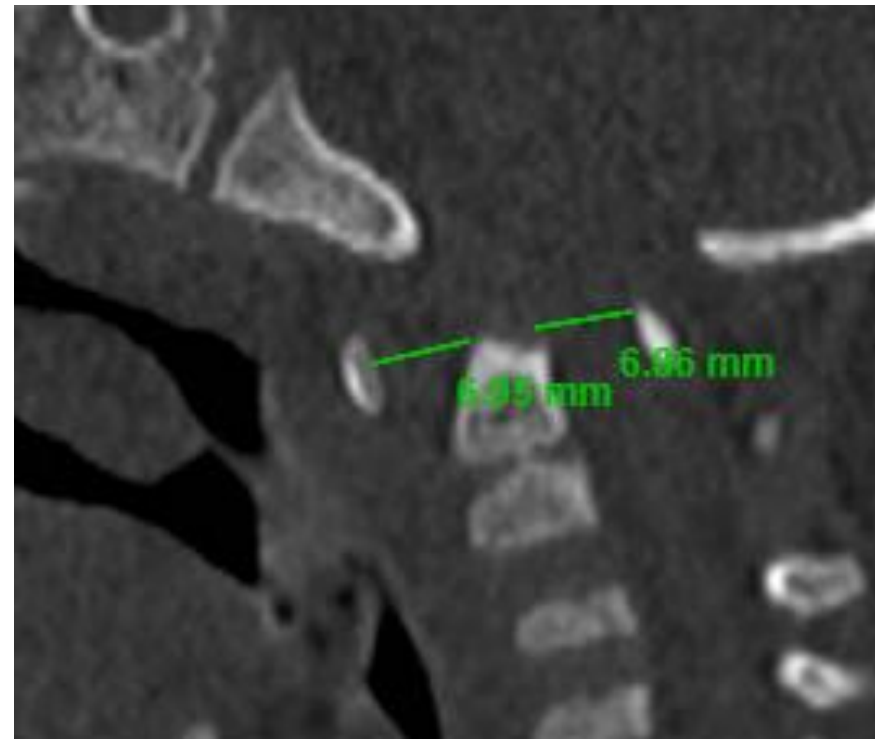
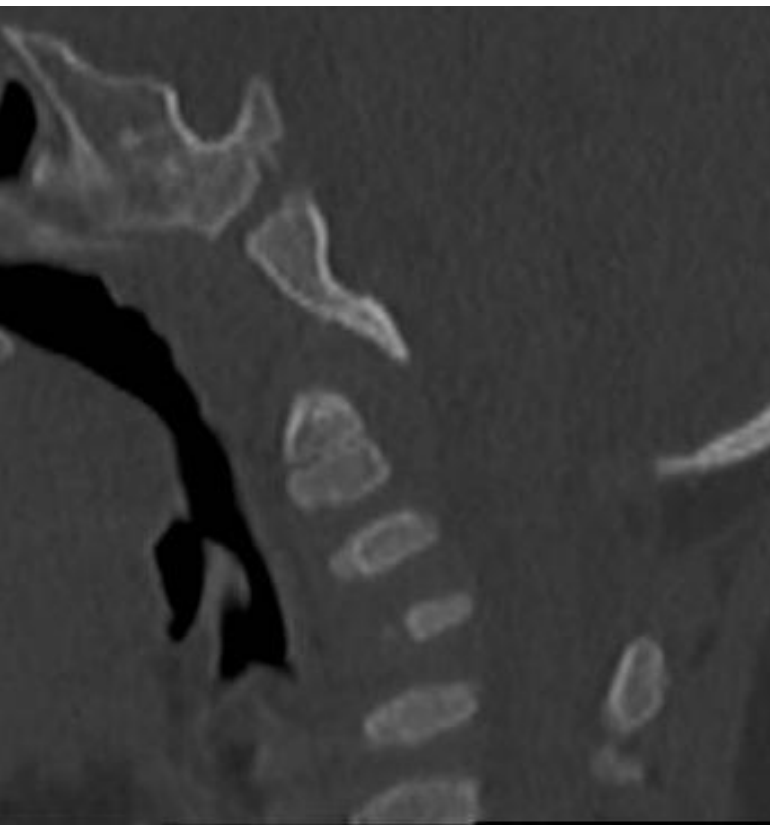


**RADIATION
EXPOSURE!!**

!

Imaging techniques

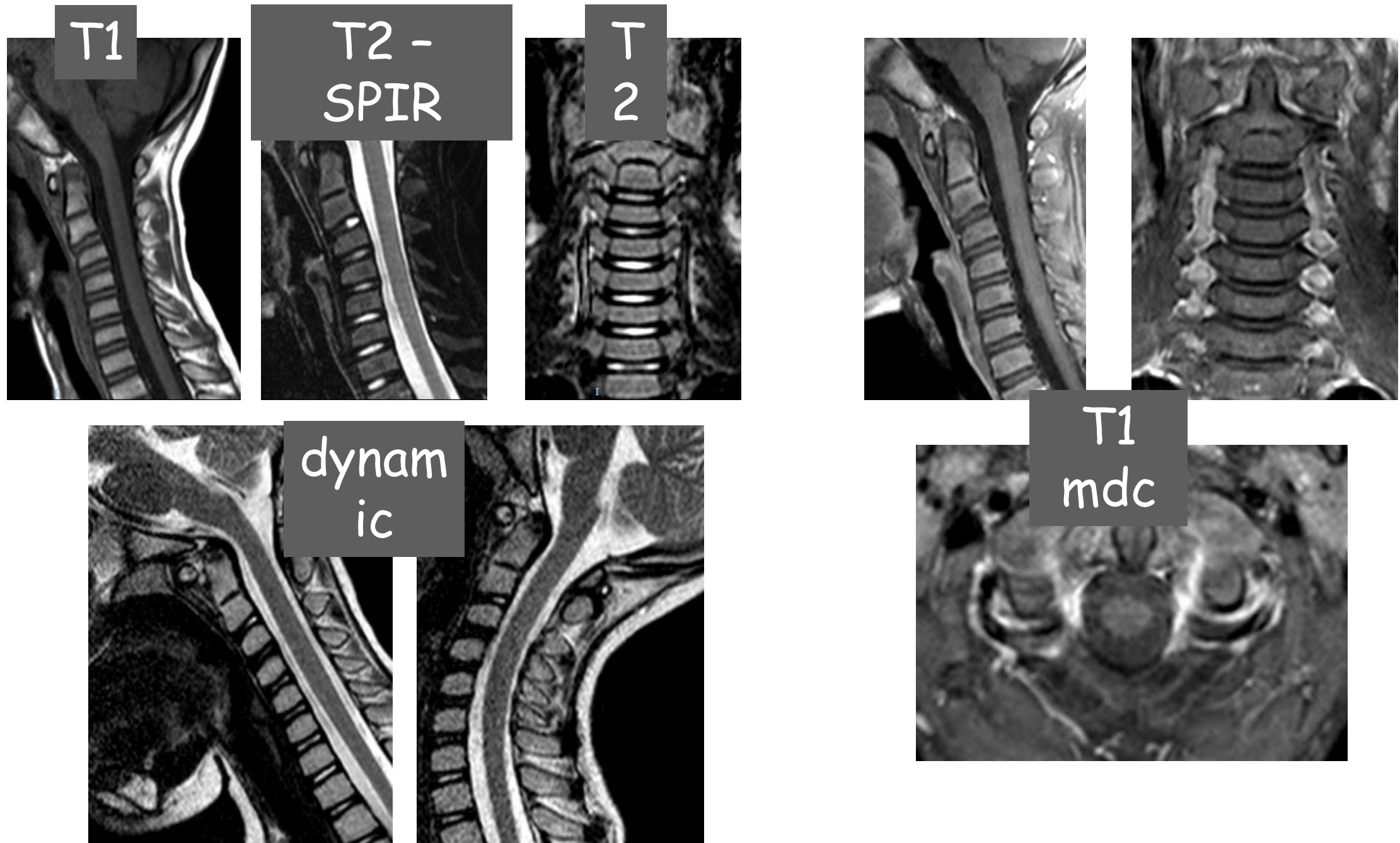
3D CT reconstructions



Imaging techniques

- MRI (static - dynamic)

ideal for evaluation of soft tissues, neural structures, and ligaments / bone edema



Clinical Indication and timing for Fixation

clinical presentation

Paresthesias

Weakness/stiffness

torticollis

Hoarseness

Neck
pain

headache

Breathing
difficulty

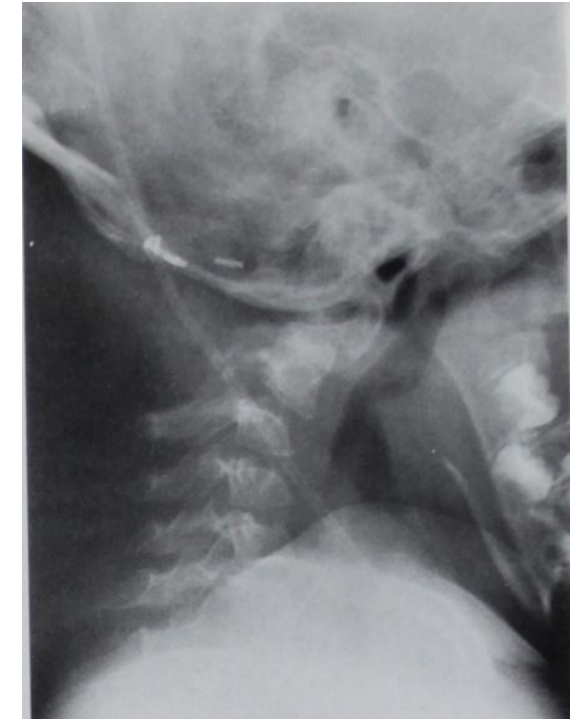
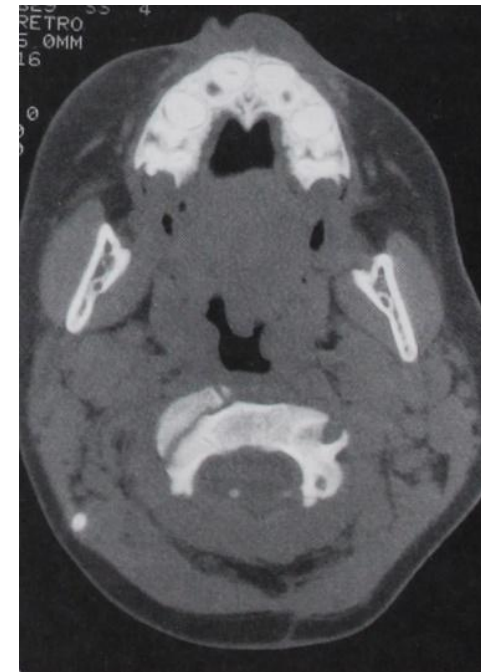
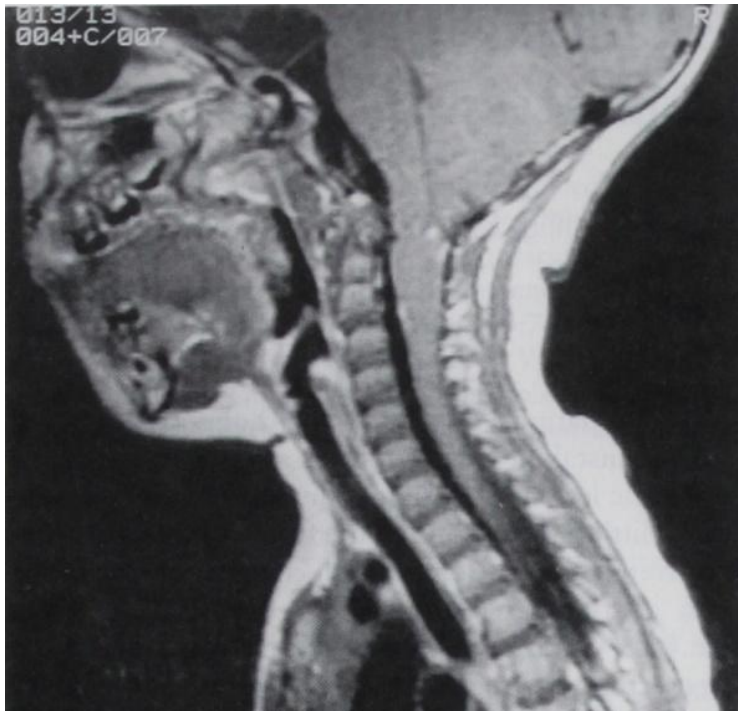
Arm Pain

Ataxia

Quadriparesis(may
be transient)

Incidence of Postlaminectomy Kyphosis After Chiari Decompression

Mark R. McLaughlin, MD,* John B. Wahlig, MD,* and Ian F. Pollack, MD†



In summary, we believe that careful attention to technique with minimal bony disruption of laminae and preservation of the facet joints is a critical factor in decreasing the incidence of PLK in this patient population.

Placement of occipital condyle screws for occipitocervical fixation in a pediatric patient with occipitocervical instability after decompression for Chiari malformation

Case report

**KIMON BEKELIS, M.D.,¹ ANN-CHRISTINE DUHAIME, M.D.,¹ SYMEON MISSIOS, M.D.,¹
CLIFFORD BELDEN, M.D.,² AND NATHAN SIMMONS, M.D.¹**

¹Section of Neurosurgery and ²Department of Radiology, Dartmouth-Hitchcock Medical Center, Lebanon, New Hampshire

In cadaveric studies and recently in one adult patient the occipital condyle has been studied as an option to allow bone purchase by fixation devices. In the current case the authors describe the use of occipital condyle screws in a child undergoing occipitocervical fixation. To the best of the authors' knowledge this case is the first reported instance of this technique in a pediatric patient.

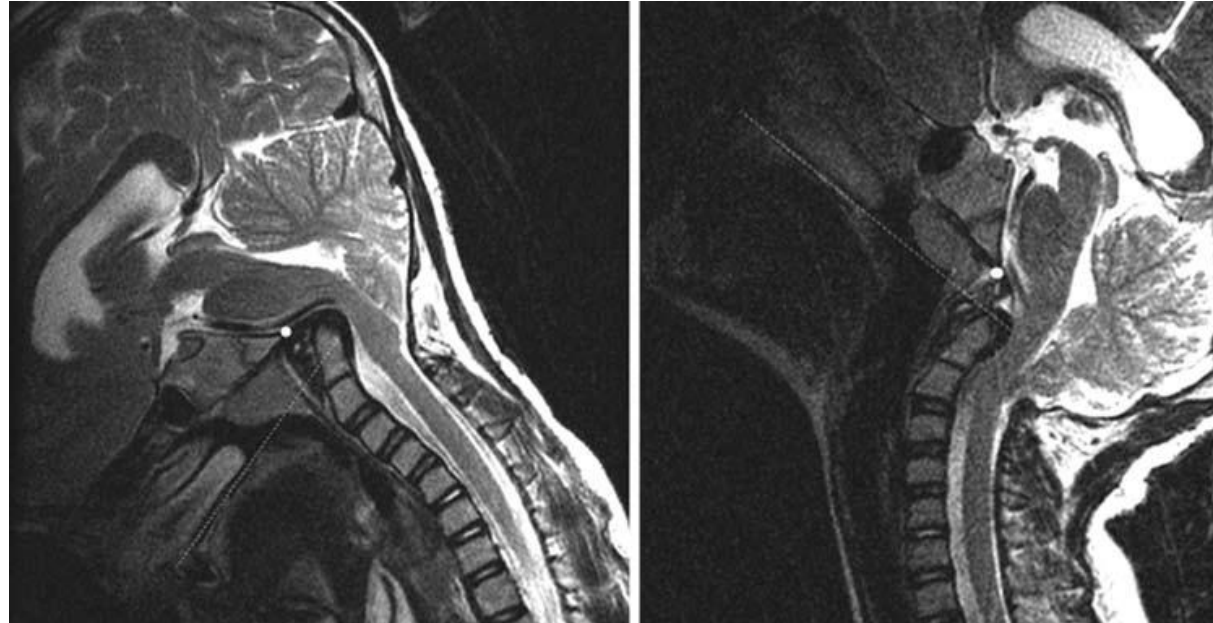
This girl had a history of posterior fossa decompression for Chiari malformation Type I when she was 22 months of age. When she was 6 years old she presented with neck pain on flexion and extension of her head. Magnetic resonance imaging in flexion and extension revealed occipitocervical instability. She underwent an occiput to C-2 posterior arthrodesis with bilateral screw placement in the occipital condyles, C-2 lamina, and C-1 lateral masses. Postoperatively, she was neurologically intact. Computed tomography demonstrated a stable construct, and her cervical pain had resolved on follow-up. (DOI: 10.3171/2010.4.PEDS09551)

KEY WORDS • Chiari malformation Type I • occipital condyle screw •
atlantooccipital instability



FIG. 1. Sagittal T2-weighted MR image obtained before initial posterior fossa decompression at the age of 22 months, showing extension of the tonsils to C-2. Basilar invagination is not present, but the odontoid is tilted posteriorly. *White dot* indicates the tip of the basion; *dotted line* shows the plane of the hard palate.

Operato a 22 mesi



Impressione basilare Con instabilità

FIG. 2. Preoperative sagittal T2-weighted MR images of the head and cervical spine, flexion (**left**) and extension (**right**) views, demonstrating basilar invagination and instability of the craniocervical junction with significant deformity of the lower brainstem and upper cervical cord. The findings worsen with flexion. *White dot* indicates the tip of the basion; *dotted line* shows the plane of the hard palate. The increased laxity allows the basion to slip forward and inferior with flexion and, coupled with the retroflexion of the dens, results in compression of the medulla.

A 6 aa dolore
Occipitale accentuato
Da Valsalva



FIG. 3. Preoperative reconstructed sagittal (**left**) and 3D (**right**) CT of the cervical spine, demonstrating basilar invagination (**left**) and changes due to suboccipital craniectomy and C1-2 laminectomies (**right**).



FIG. 5. Postoperative axial CT at the level of the occipital condyles demonstrating the position of the occipital condyle screws.

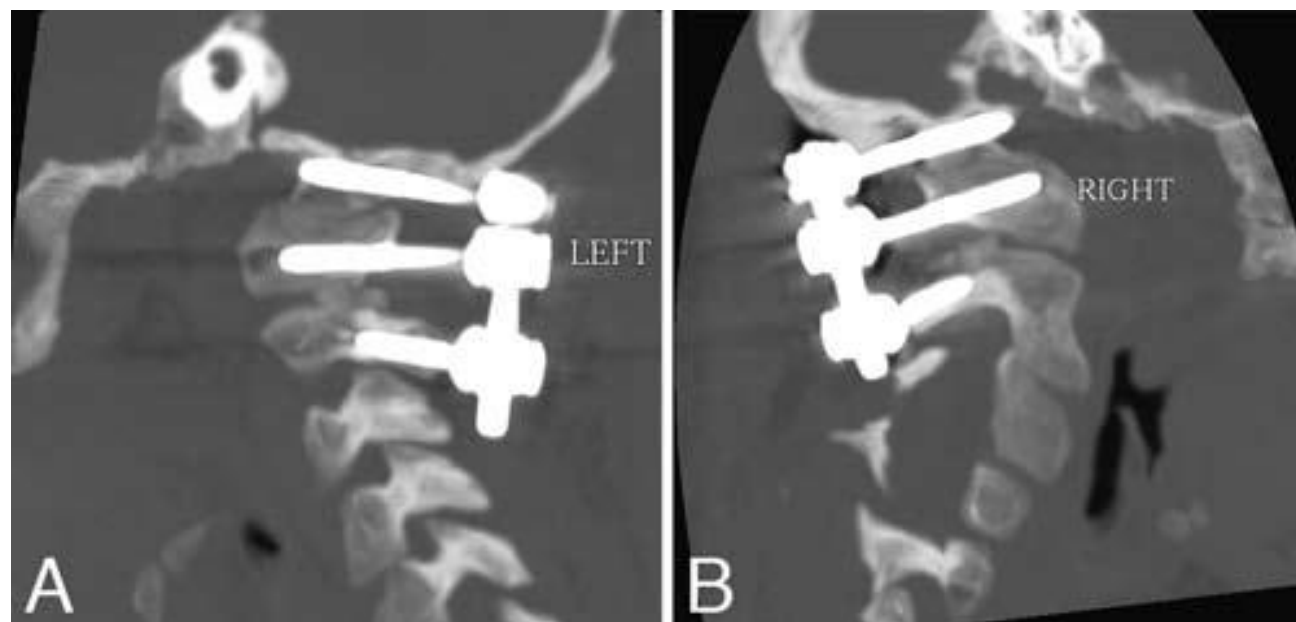


FIG. 6. Postoperative reconstructed sagittal CT of the cervical spine showing the position of the left **(A)** and right **(B)** construct from the occiput to C-2.

Craniocervical junction fusions in patients with hindbrain herniation and syringohydromyelia

ALBERT J. FENOY, M.D., ARNOLD H. MENEZES, M.D., AND KATHLEEN A. FENOY, A.R.N.P.

Department of Neurosurgery, University of Iowa Hospitals and Clinics, Iowa City, Iowa

234 patients
Ages ranged from
2,5 to 86 years
33% (77 <16years)

TABLE 3
*Indications for craniocervical fusion in patients with hindbrain herniation and/or syringomyelia treated between 1996 and 2005**

Group	Description	Examples	Associated Radiographic Findings†	Treatment	No. of Patients (%)‡
I	congenital or acquired cranio-cervical abnormalities w/ reducible bone compression	1) atlas assimilation & other occipital bone atypia; 2) basilar invagination; 3) Klippel–Feil anomaly	dynamic MRI: ventral compression of brainstem in flexion, relieved w/ extension; 3D CT: demonstration of ventral bone abnormality	PFD &/or FMD & dorsal cranio-cervical fusion	59 (25)
II	occurs after previous anterior decompression of lower brain-stem/upper cervical cord			all require dorsal craniocervical fusion	103 (44)
III	unstable CCJ w/o bone abnormalities	1) pathological states such as Noonan syndrome	dynamic MRI: ventral compression of brainstem in flexion, relieved w/ extension; CT: normal osseous anatomical configuration	PFD &/or FMD & dorsal cranio-cervical fusion	60 (26)
IV	musculoligamentous instability	1) pathological states, such as Ehlers–Danlos or Down syndrome; 2) neurogenic – upper cervical cord syrinx causing poor muscle control & instability; 3) after repeated PFDs	craniocervical MRI: evidence of ligamentous laxity/disruption on STIR sequencing, dehiscence of muscle approximation, or hypertrophied fibrotic scar; presence of cervical syrinx	dorsal craniocervical fusion	33 (14)

* STIR = short tau inversion recovery.
† Radiographic findings in addition to the presence of hindbrain herniation or syringomyelia.
‡ Some patients may fall into > 1 category.

“In any of these cases,it is imperative that the treating physician obtains a comprehensive preoperative imaging survey of the CVJ ,including dynamic MR cervical extension and flexion image to be able to formulate the best treatment”

Craniovertebral junction abnormalities with hindbrain herniation and syringomyelia: regression of syringomyelia after removal of ventral craniovertebral junction compression

Clinical article

ARNOLD H. MENEZES, M.D.

Department of Neurosurgery, University of Iowa Hospitals and Clinics, Iowa City, Iowa

A. H. Menezes

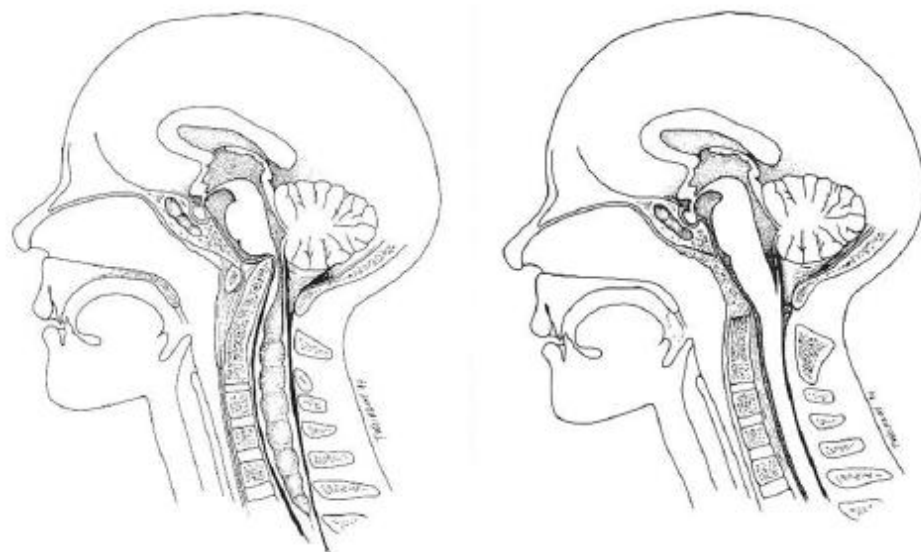


FIG. 7. Artist's illustrations. Left: Atlas assimilation with basilar invagination into the midmedulla and tonsillar impaction. A holocord syrinx is depicted. Right: Result of ventral decompression of the basilar invagination, showing a striking change in the posterior fossa volume and regression of the syrinx.



Chiari I malformation with and without basilar invagination: a comparative study

Jörg Klekamp, MD

Department of Neurosurgery, Christliches Krankenhaus, Quakenbrück, Germany

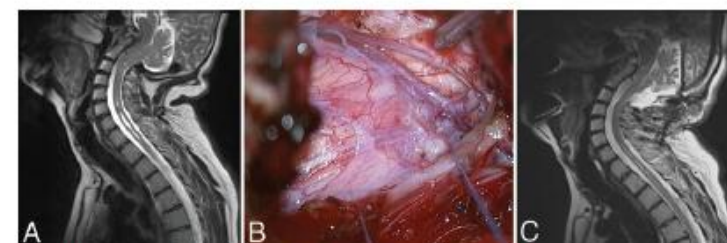


FIG. 2. A: Sagittal MRI scan of a 26-year-old man with a 4-year history of occipital pain and gait ataxia demonstrates a Chiari I malformation with syringomyelia and a basilar invagination with ventral compression of the medulla. B: Intraoperative photograph. After dura and arachnoid opening, adhesions encasing both posterior inferior cerebellar arteries and obstructing the foramen of Magendie become apparent. C: Postoperative sagittal MRI obtained after 3 months shows the realignment achieved by extension and fixation of C0-C3 and a decrease of the syrinx.

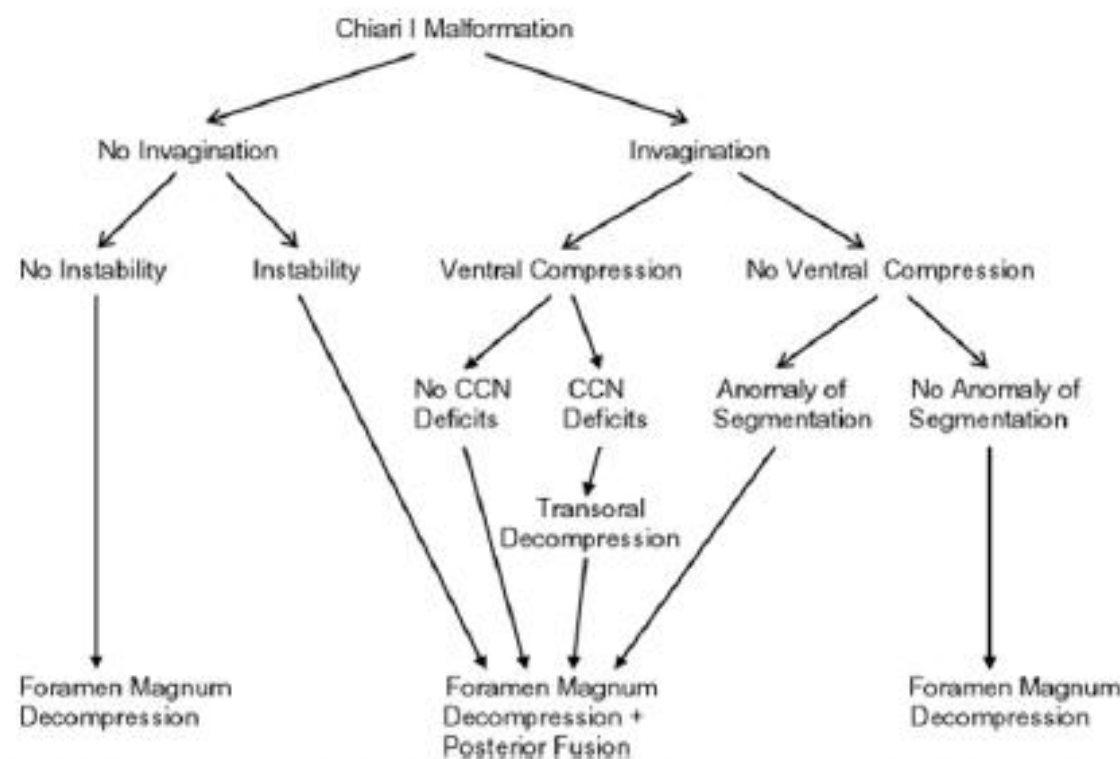
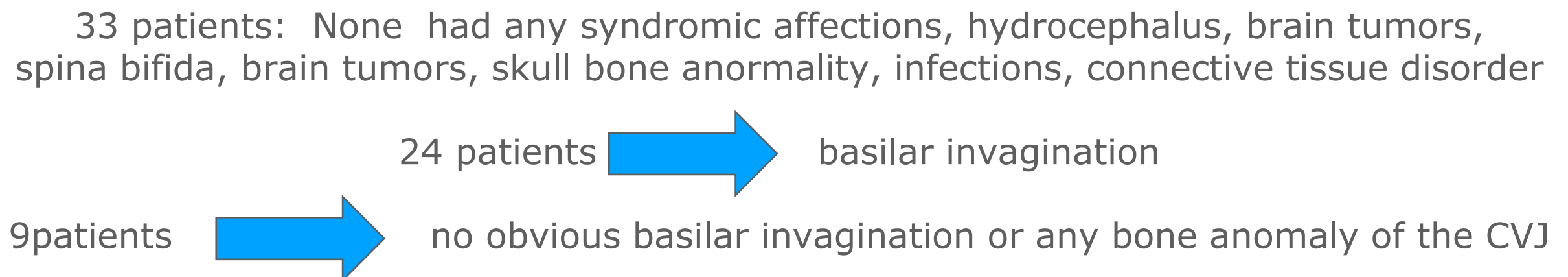


FIG. 9. Algorithm for surgical management of Chiari I malformation in patients with and without additional basilar invagination. CCN = caudal cranial nerves.



Atlantoaxial Fixation for Chiari 1 Formation in Pediatric Age-Group Patients: Report of Treatment in 33 Patients

Atul Goel^{1,2}, Sandeep Gore¹, Abhidha Shah¹, Pralhad Dharurkar¹, Ravikiran Vutha¹, Abhinandan Patil¹



Regardless of the radiologic observations, the presence of Chiari formation by itself was considered a reliable indicator of atlantoaxial instability meriting the need for atlantoaxial stabilization.¹

ORIGINAL ARTICLE



Atlantoaxial Fixation for Chiari 1 Formation in Pediatric Age-Group Patients: Report of Treatment in 33 Patients

Atul Goel^{1,2}, Sandeep Gore¹, Abhidha Shah¹, Pralhad Dharurkar¹, Ravikiran Vutha¹, Abhinandan Patil¹

CM1 secondary to atlantoaxial Dislocation

Atlanto axial stabilization forms the mode of surgical treatment

FM decompression is not necessary and in the long run could be a counterproductive operation



Chiari malformation type I and basilar invagination originating from atlantoaxial instability: a literature review and critical analysis

Arthur Wagner¹  · Lukas Grassner² · Nikolaus Kögl² · Sebastian Hartmann² · Claudius Thomé² · Maria Wostrack¹ · Bernhard Meyer¹

Received: 14 April 2020 / Accepted: 22 May 2020 / Published online: 6 June 2020
© The Author(s) 2020

At present, there is insufficient evidence supporting the hypothesis of atlantoaxial instability being the common progenitor for CVJ abnormalities.

S. A.
Female
11 yrs

Sudden occurrence of paraparesis and bladder globus.

Steroid treatment followed by recovery of normal neurological status within 24 hours.

Brain & Spine MRI: basilar impression, platybasia, medullary ectopia lower than 5 mms, signal abnormality of the spinal cord at C1–C2 level.

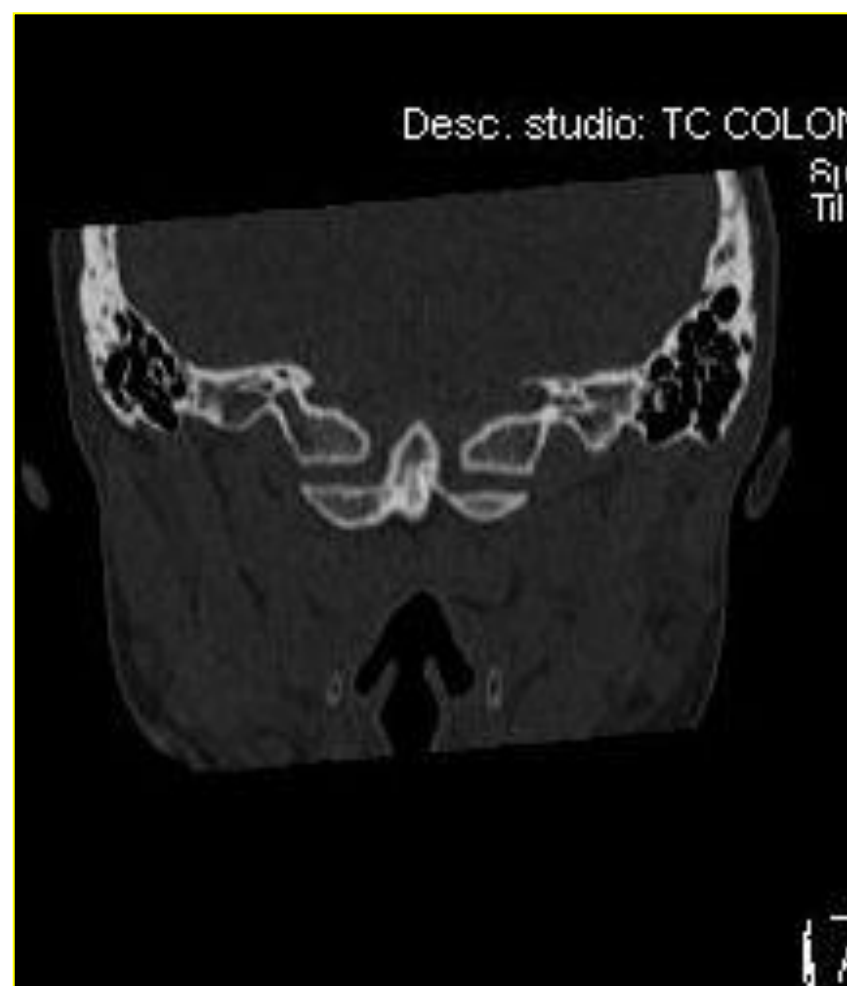


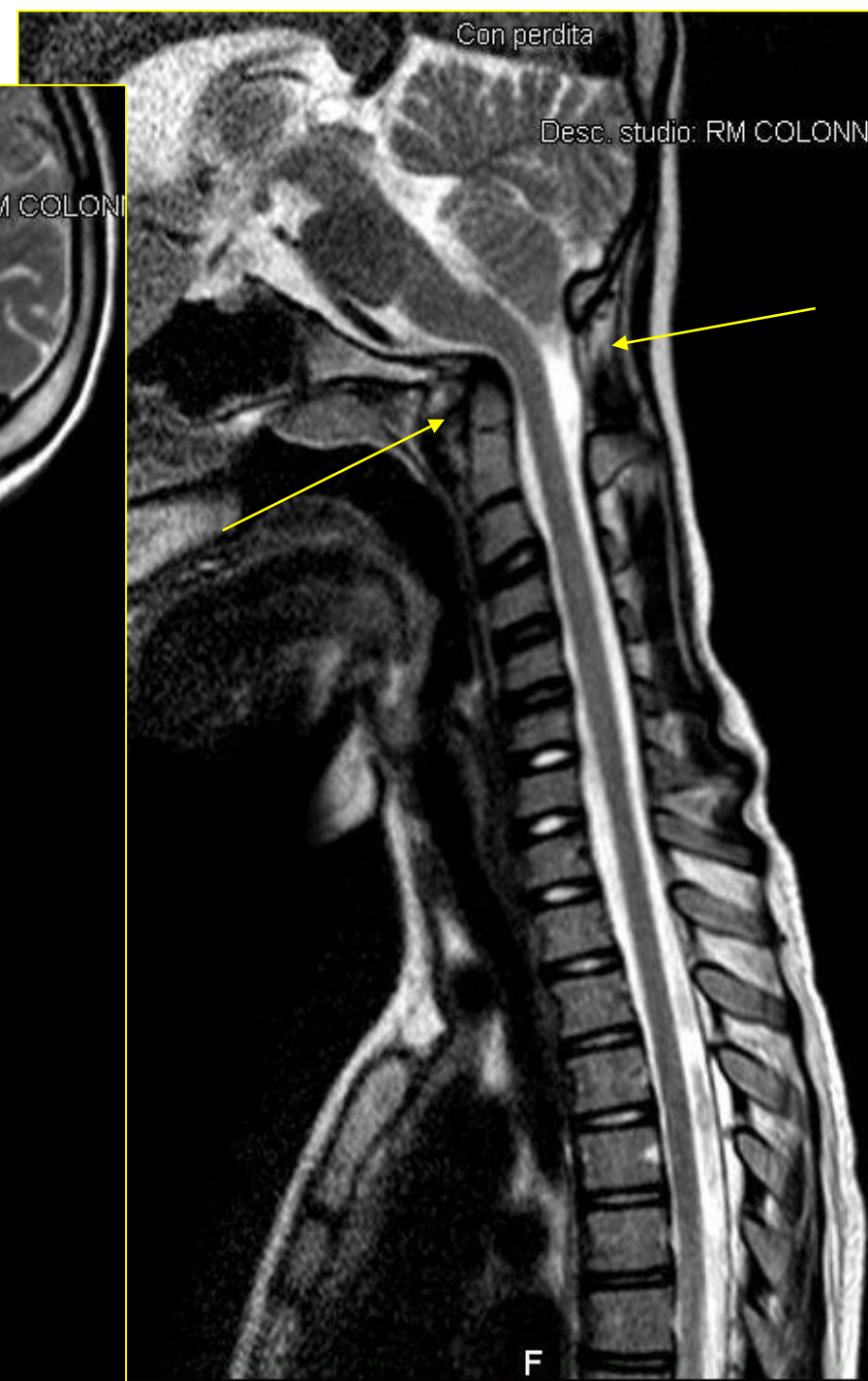
On admittance:
normal neurological status

Neuroradiological assessment:

- Platybasia
- Basilar impression
- Atlanto-occipital fusion and fusion of C2–C3 vertebral bodies and laminae
- Absent C1 posterior arc
- Chiari 1
- Narrowing of the Foramen Magnum









“Treatment of basilar invagination associated with Chiari 1 malformation in the pediatric population: cervical reduction and posterior occipito-cervical fusion.”

Rekate, Sontag.

J.Neurosurg 2004 Nov; 101(2 suppl): 189-95





“In selected cases, manual cervical distraction and extension, posterior fixation and fusion appear to provide a safe, effective standalone treatment for basilar invagination associated with Chiari malformations in children”



Chiari malformation and atlantoaxial instability: problems of co-existence

Sandip Chatterjee¹  • Pankaj Shivhare² • Shyam Gopal Verma²

Received: 19 June 2019 / Accepted: 26 June 2019 / Published online: 13 July 2019
© Springer-Verlag GmbH Germany, part of Springer Nature 2019



Fig. 1 Worsening of spastic quadriparesis in a child due to missed AAD and operated Chiari I malformation

Gaslini CM1 series

556 CM1 297 surgery

7 patients (2,3 %) underwent occipito cervical fixation (everyone had a cvj malformation , see basilar invagination, ossiculo terminalis,Hypoplasia of odontoid components)

1 patient developed postoperative Kyphosis completely resolved with orthosis

Finally in our experience a very low number of cases needed occiput cervical arthrodesis

CM 1fixation

- 6 patients decompression+ fixation at the same time
- 1patients with basilar impression only occipito-cervical arthrodesis without decompression
- In 6 cases an occipital plate was employed, in 1 case an occipital hooks
- In 4 children we used hooks for the cervical spine in 2 patients screws in the lateral mass, in 1 translaminar screws in C2
- Mean age at surgery 8 Years (range 7-11 years)
- FU : range 1-8 years
- No clinical complications
- No dislocation of instrumentation

Type of Fixation

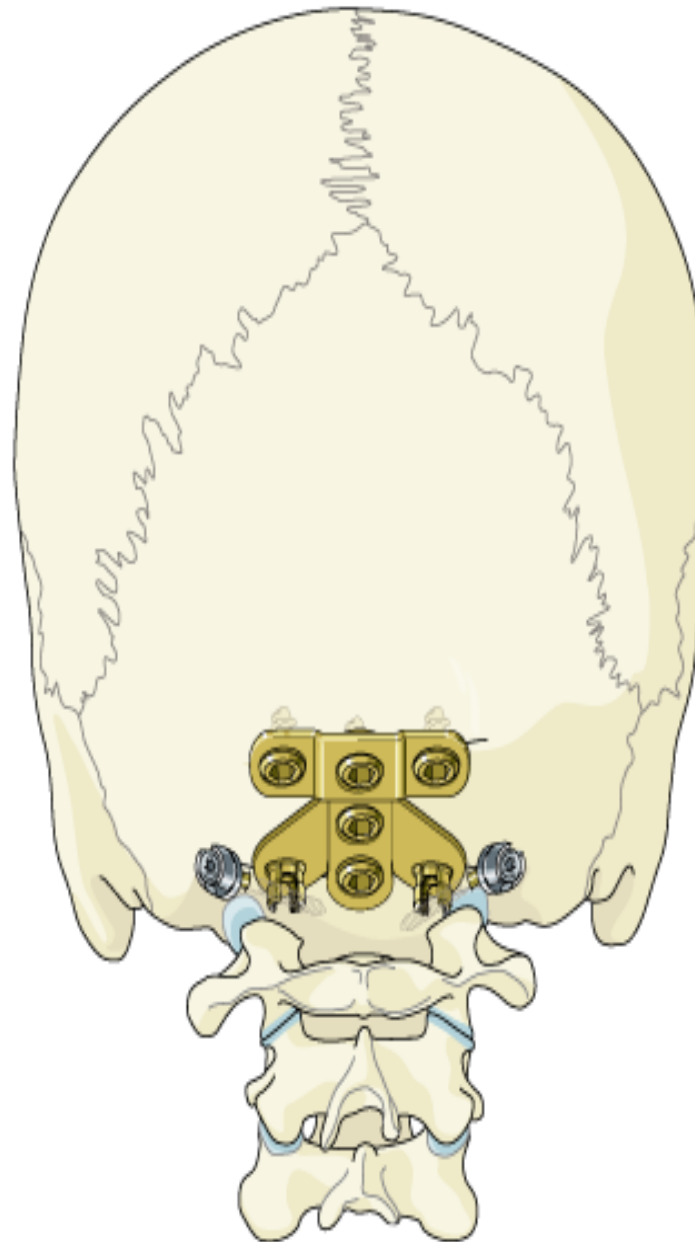
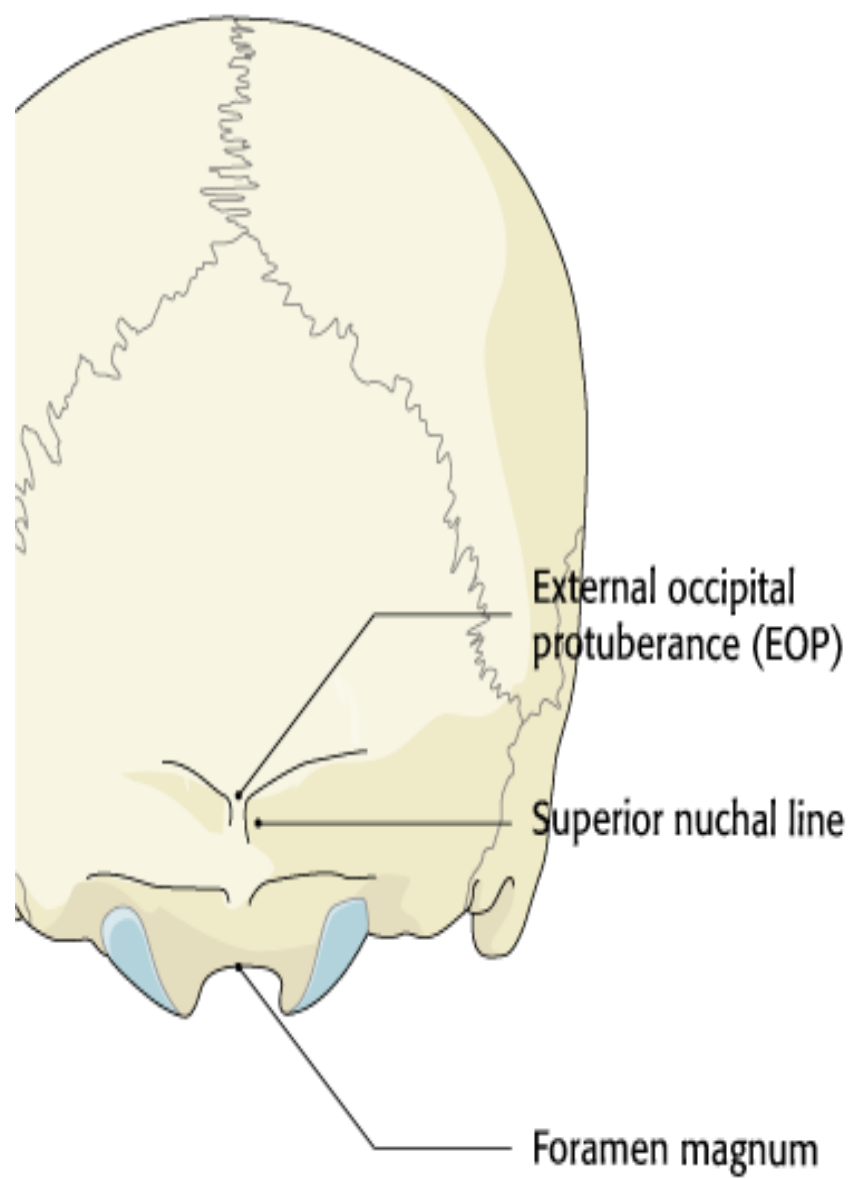
OCCIPUT: Occipital plate/hooks
Occipital condyle screw(unusual)

C1: Hooks
Lateral Mass Screw

C2: Hooks
Lateral mass screw,
pedicle screw,
pars screw,
interlaminar screw

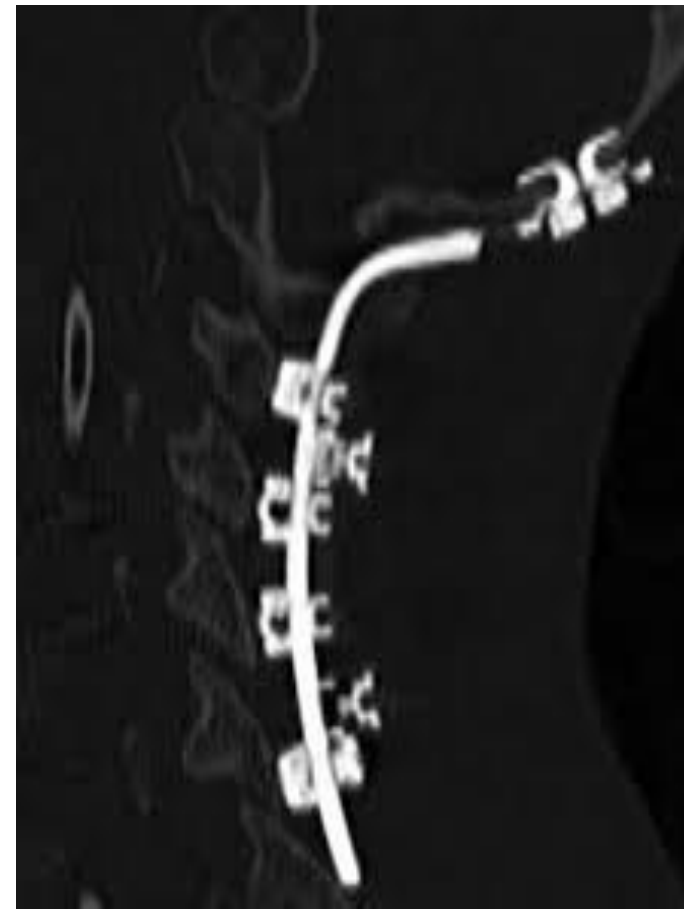
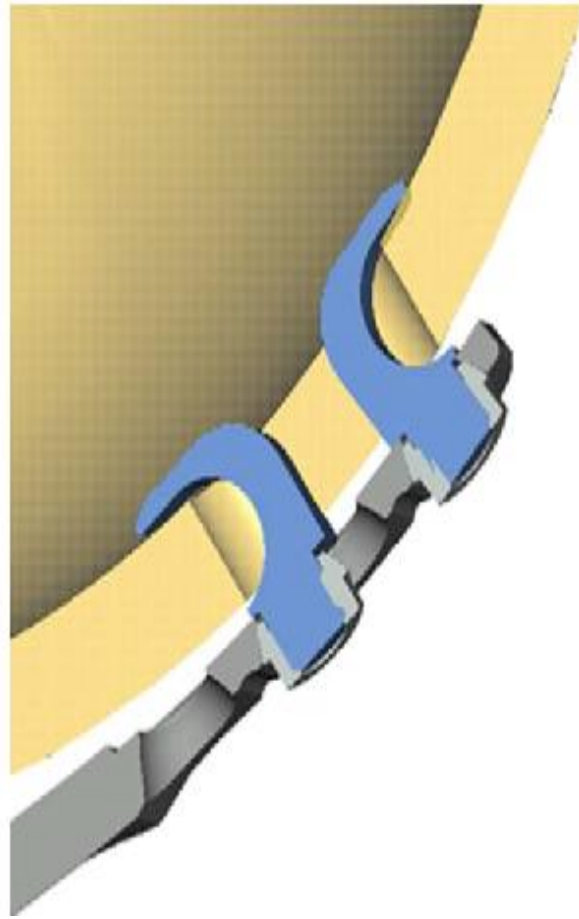
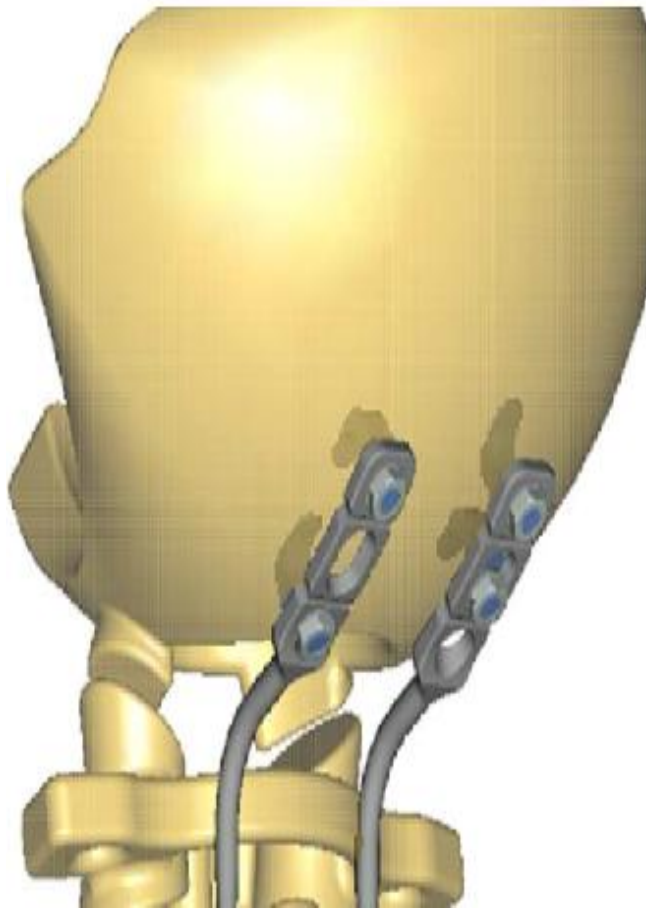
C1-C2:Transarticular screw (Magerle technique)
Subaxial cervical spine:lateral mass screw
Pedicle screw

C0 / Occiput



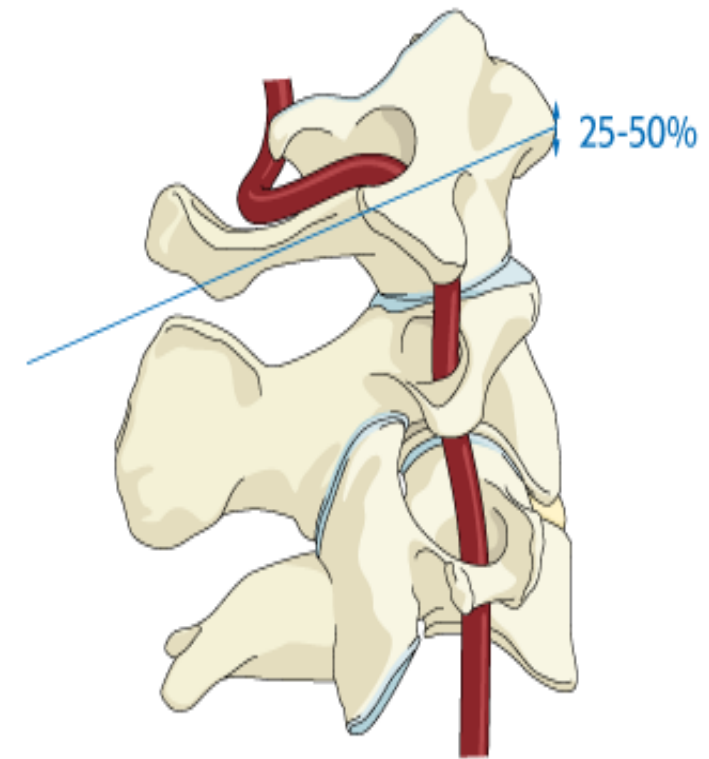
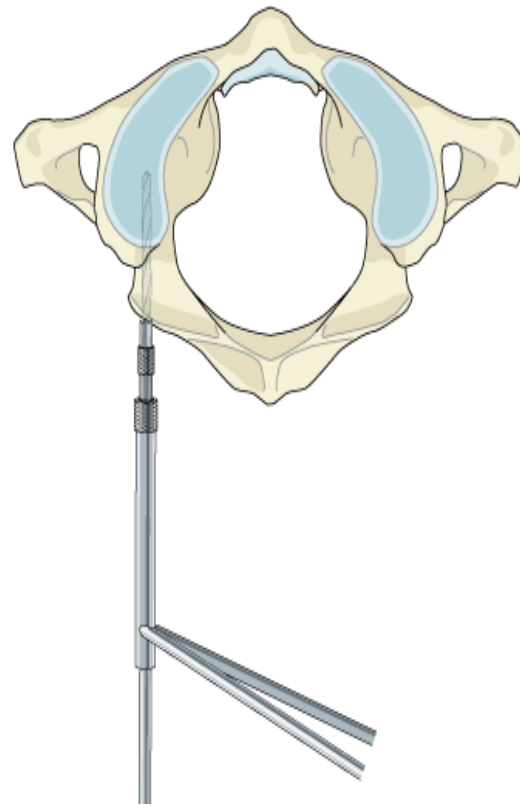
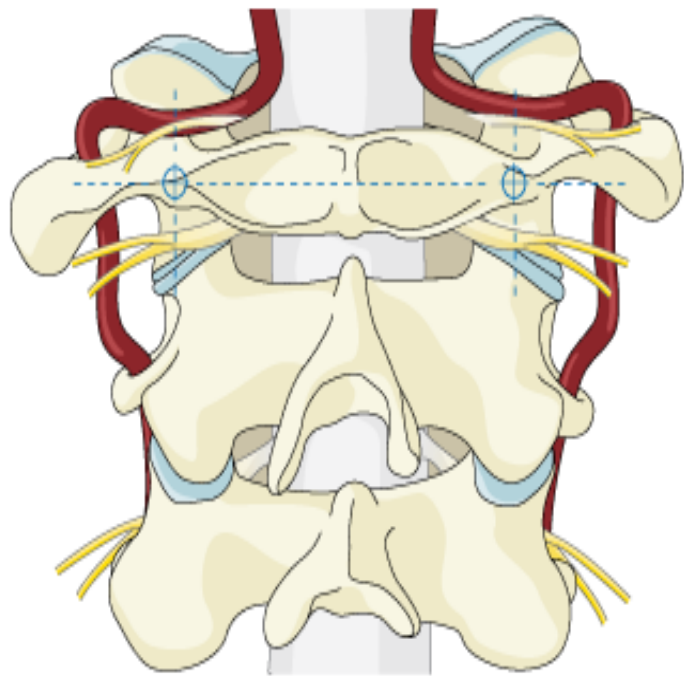
**Occipital
plate**

C0 / Occiput



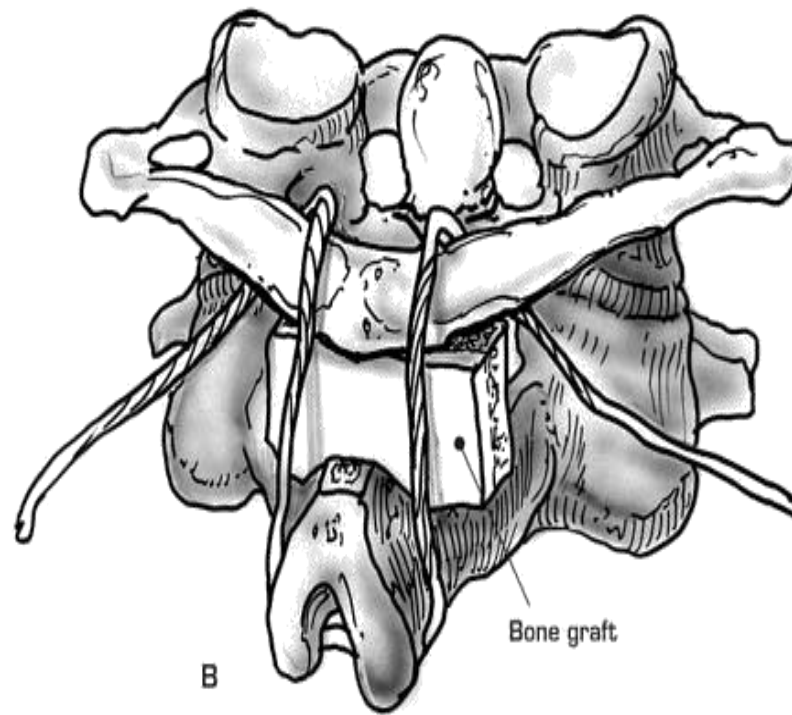
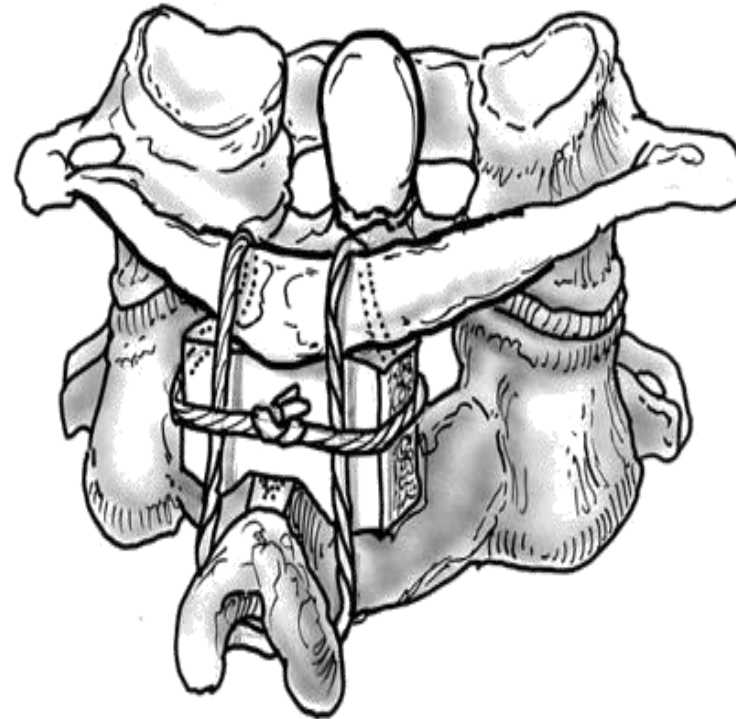
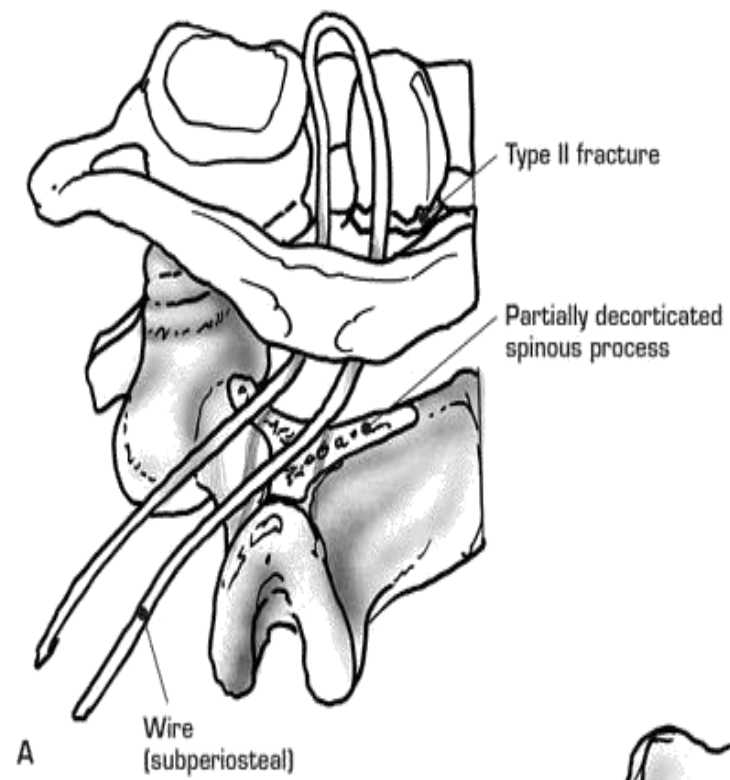
**Occipital and
cervical hooks**

C1 / Atlas



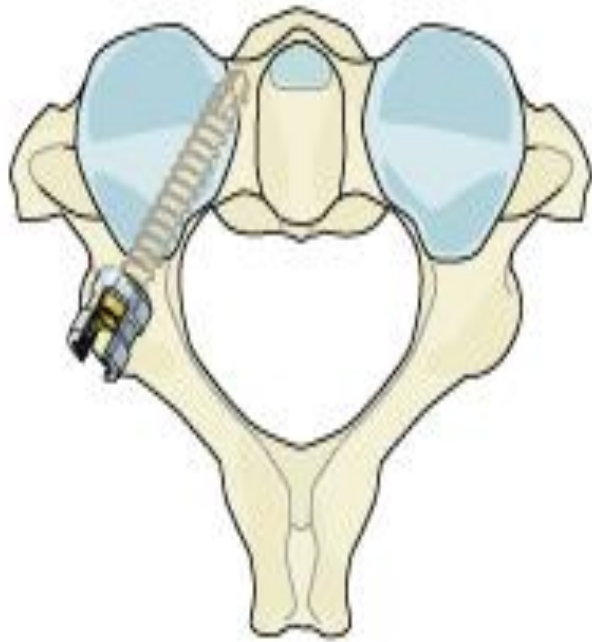
**C1 lateral mass
screw**

C1 / Atlas



C1 posterior wiring

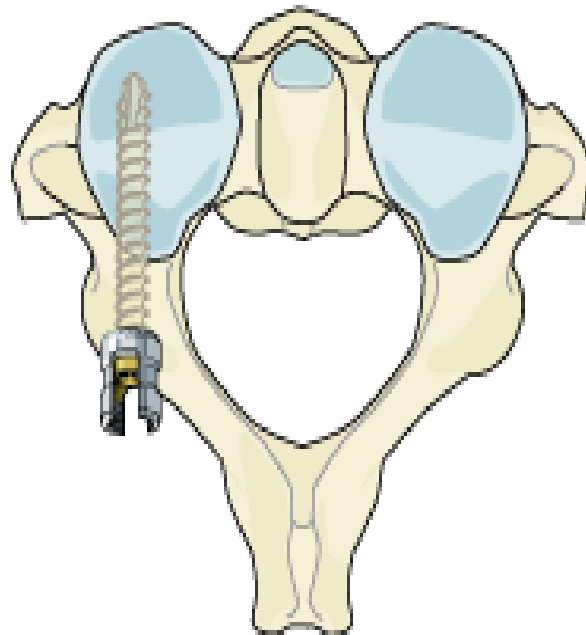
C2 / Axis



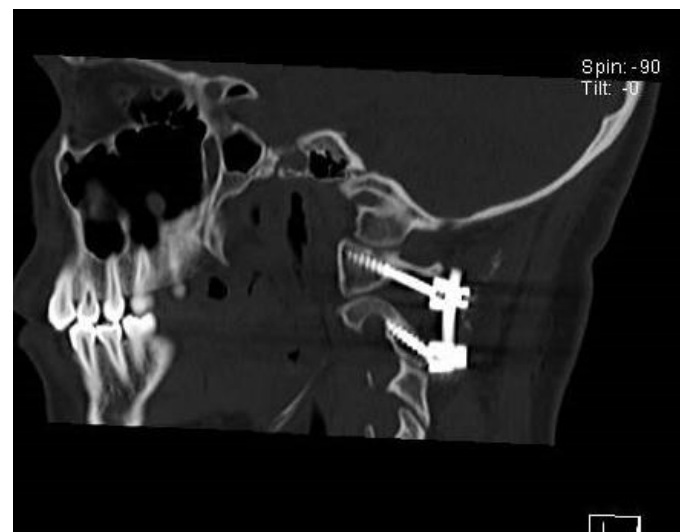
Pedicle screw



Laminar screw

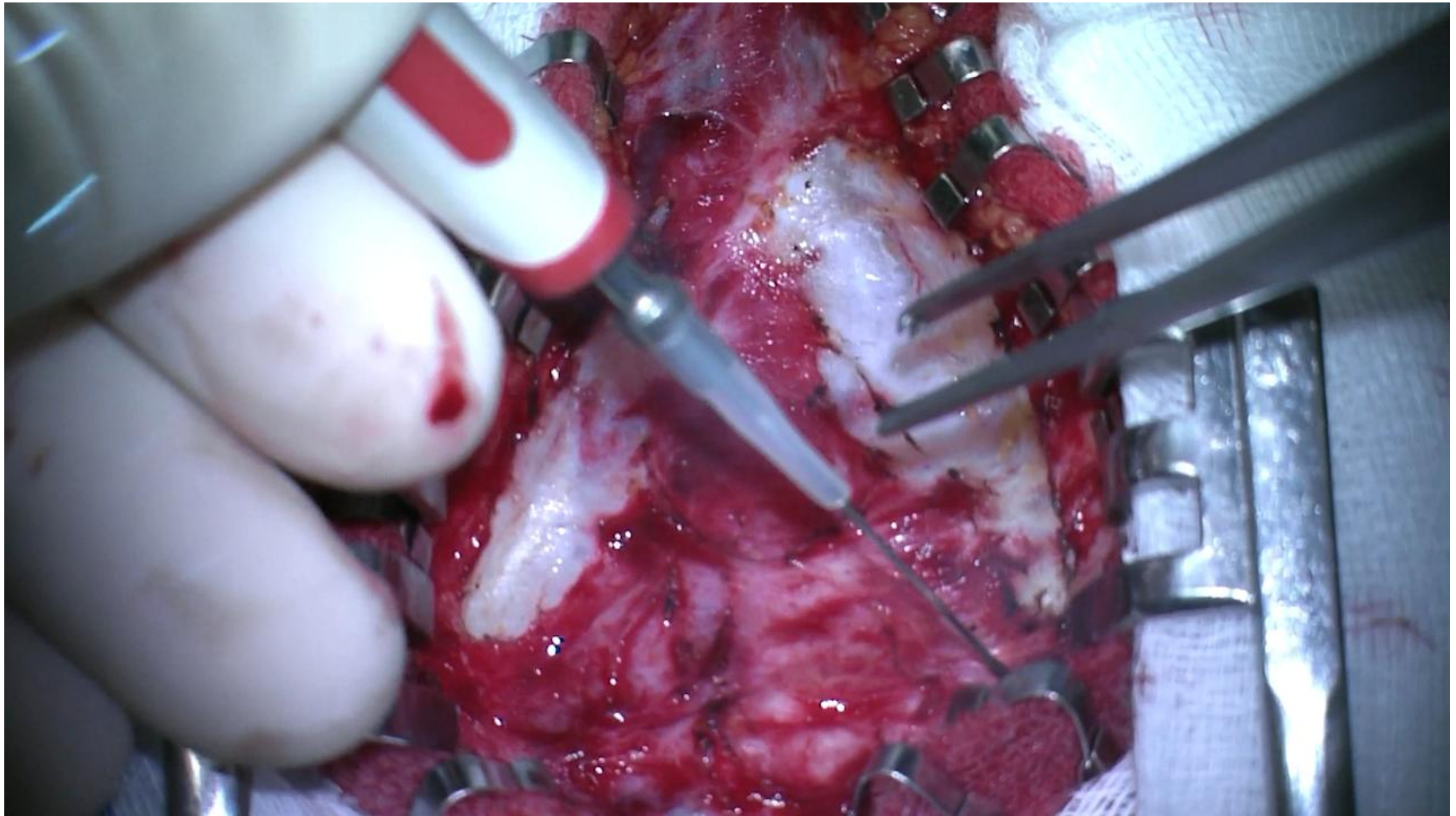


Pars screw









**“The surgical management of craniovertebral junction instability in pediatric patients has unique challenges”
(A.H. Menezes)**

«Indications for internal fixation in children are similar to those of adults

Diminutive osseous and ligamentous structures and anatomical variations associated with syndromic craniovertebral abnormalities frequently complicate the approach and limit the use of internal fixation in children

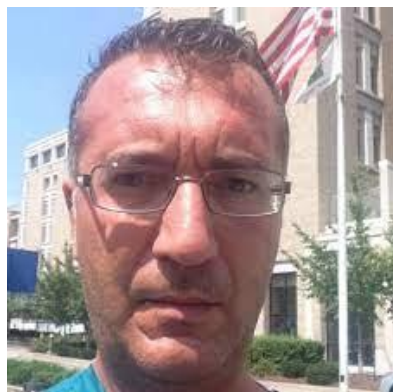
Preoperative planning is important to evaluate the size of the instrumentation and its relation to the patient's anatomy «



Passo dello Stelvio



Importante Valutazione dei soggetti a rischio di instabilità anche se l'impatto sulla storia naturale della malformazione è percentualmente basso, quantomeno nella nostra esperienza



M.Pavanello



M.Pacetti



F.Secci



A.Consales



M.Cataldi



F.Canavese



Thank you!