Neurosurgical issues for patients with VACTERL syndrome

Jeffrey W. Campbell, MD
Chief, Neurosurgery

Nemours
Alfred I. duPont Hospital for Children
Nemours Children's Clinic

Introduction

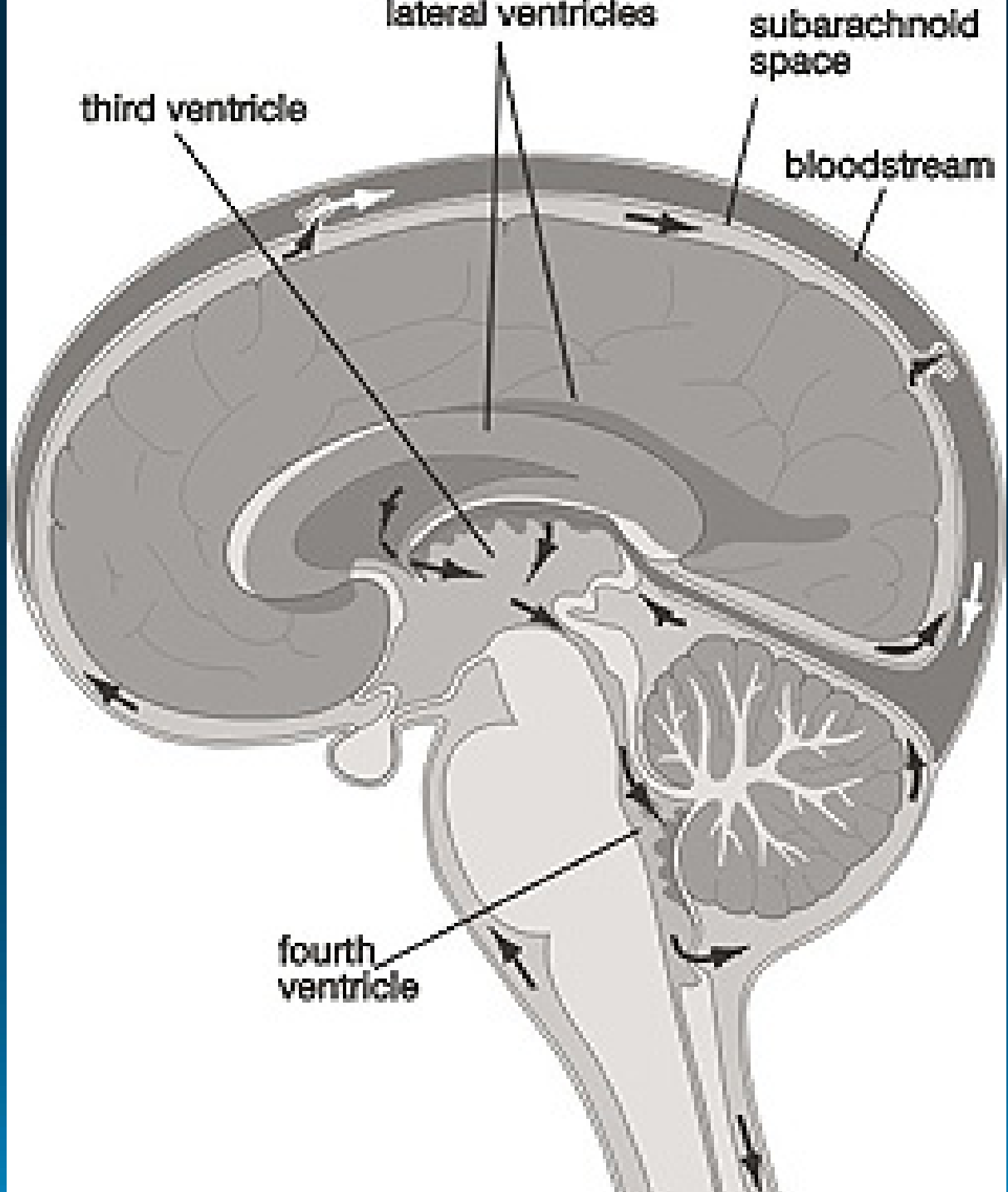
- Hydrocephalus
  - Predominately in VACTERL-H
- Chiari Malformation
  - Predominately in VACTERL-H
- Tethered cord
  - Common in all forms of anorectal malformations

VACTERL-H

- Addition of hydrocephalus to other typical findings of VACTERL syndrome
- Mode of inheritance usually X-linked
  - Affecting only males in the family
  - Couple families with autosomal recessive inheritance
- Most hydrocephalus due to aqueductal stenosis
- Couple cases associated with Chiari malformation

Hydrocephalus

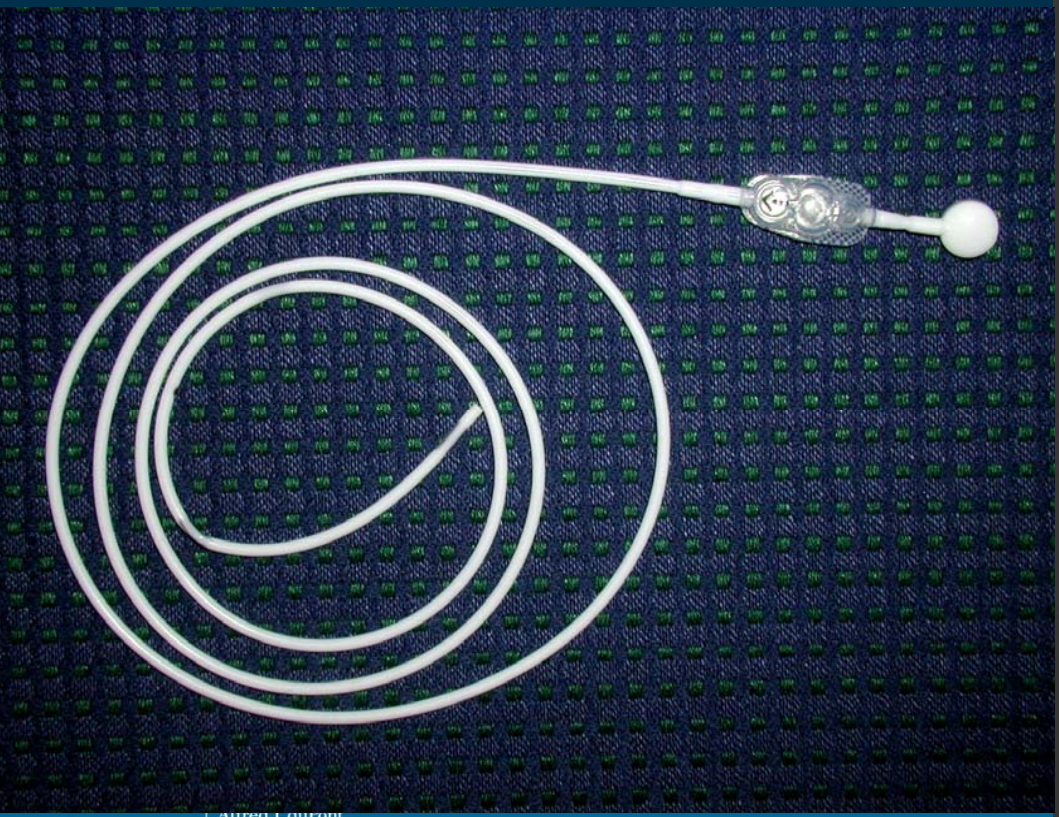
- Spinal fluid produced in spaces inside brain (ventricles)
  - Make about 500 cc/ day
- Fluid has to leave ventricles and travel into spaces between the brain and skull to be reabsorbed
- Blockages of these spinal fluid passageways will cause fluid to build up inside the brain under pressure

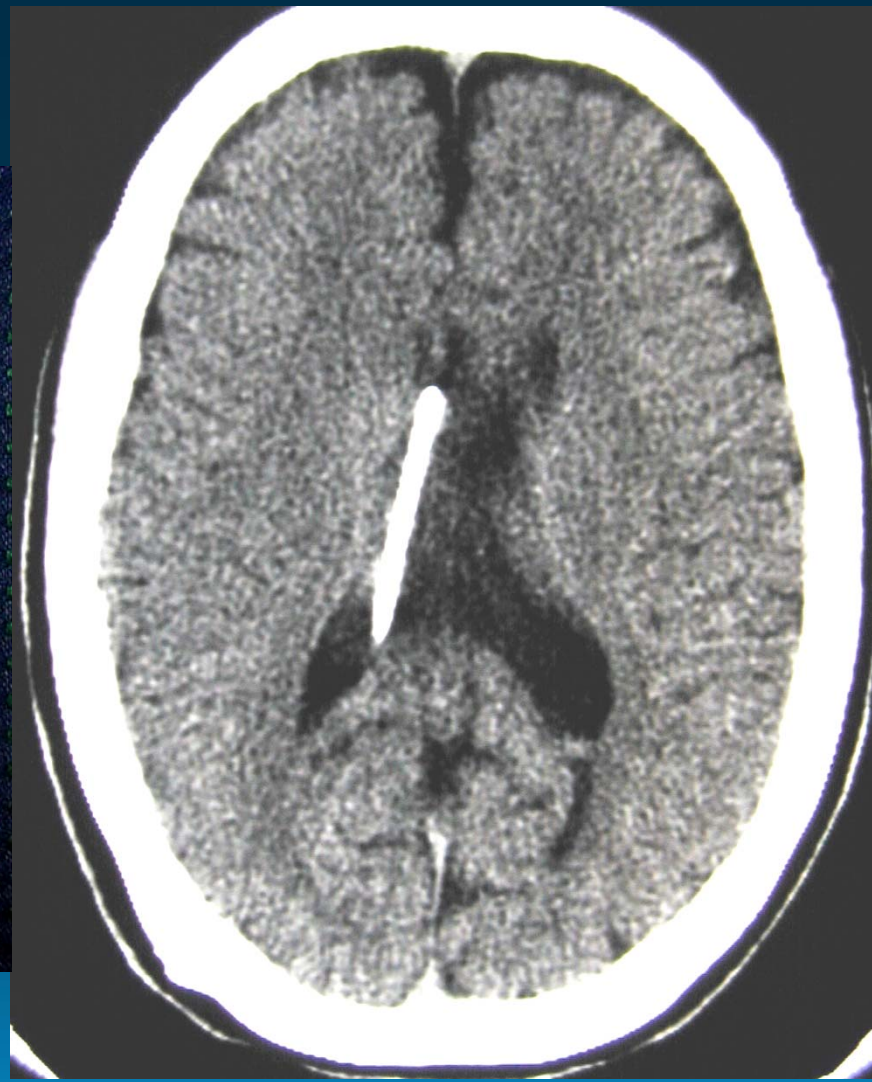


Indications for Treatment of Hydrocephalus

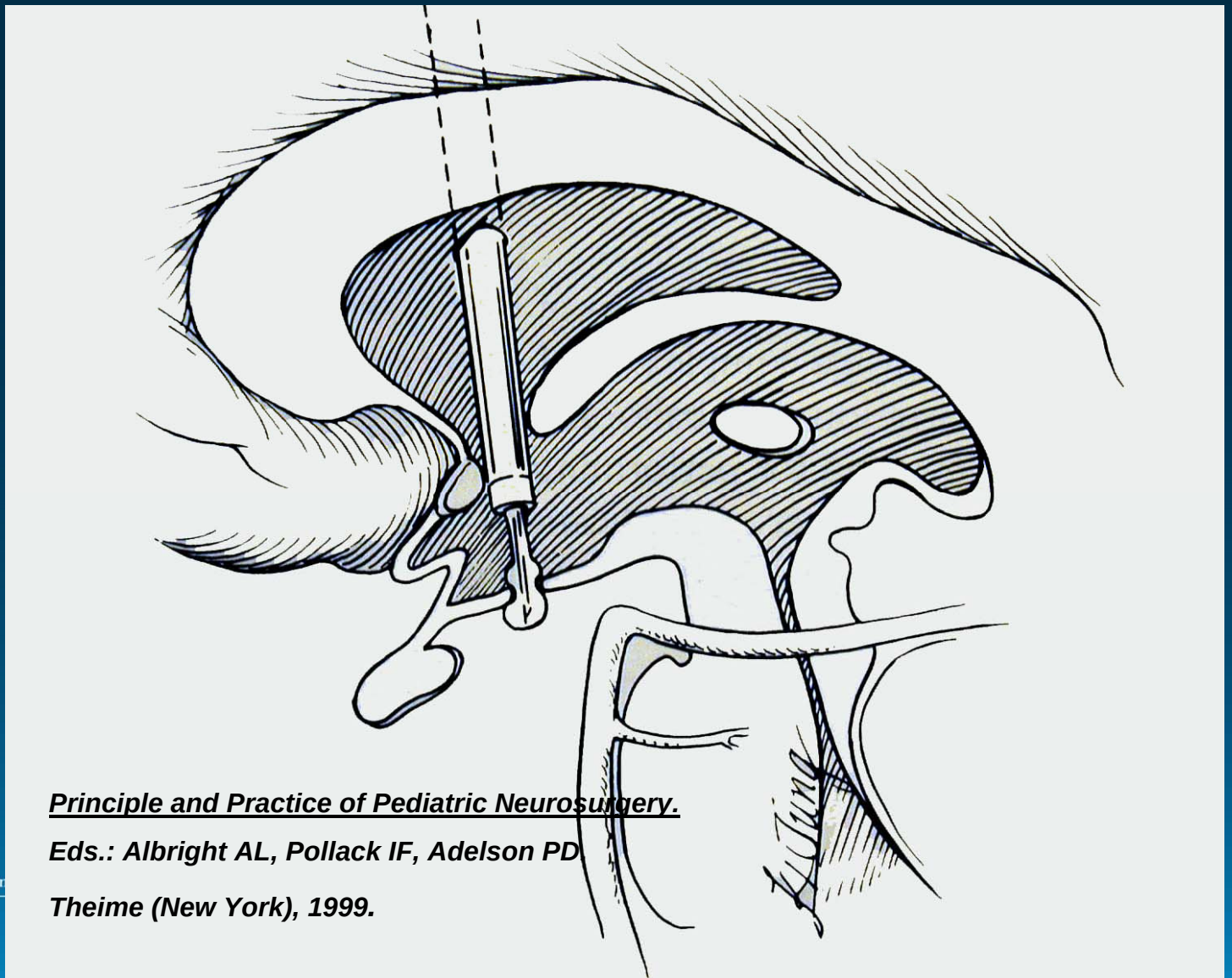
- Progressive macrocephaly
- Progressive ventriculomegaly
- Increased intracranial pressure
  - Full anterior fontanel
  - Usual signs/symptoms
- Developmental delay

Placement of VP Shunt





Endoscopic IIIrd Ventriculostomy



Principle and Practice of Pediatric Neurosurgery.

Eds.: Albright AL, Pollack IF, Adelson PD

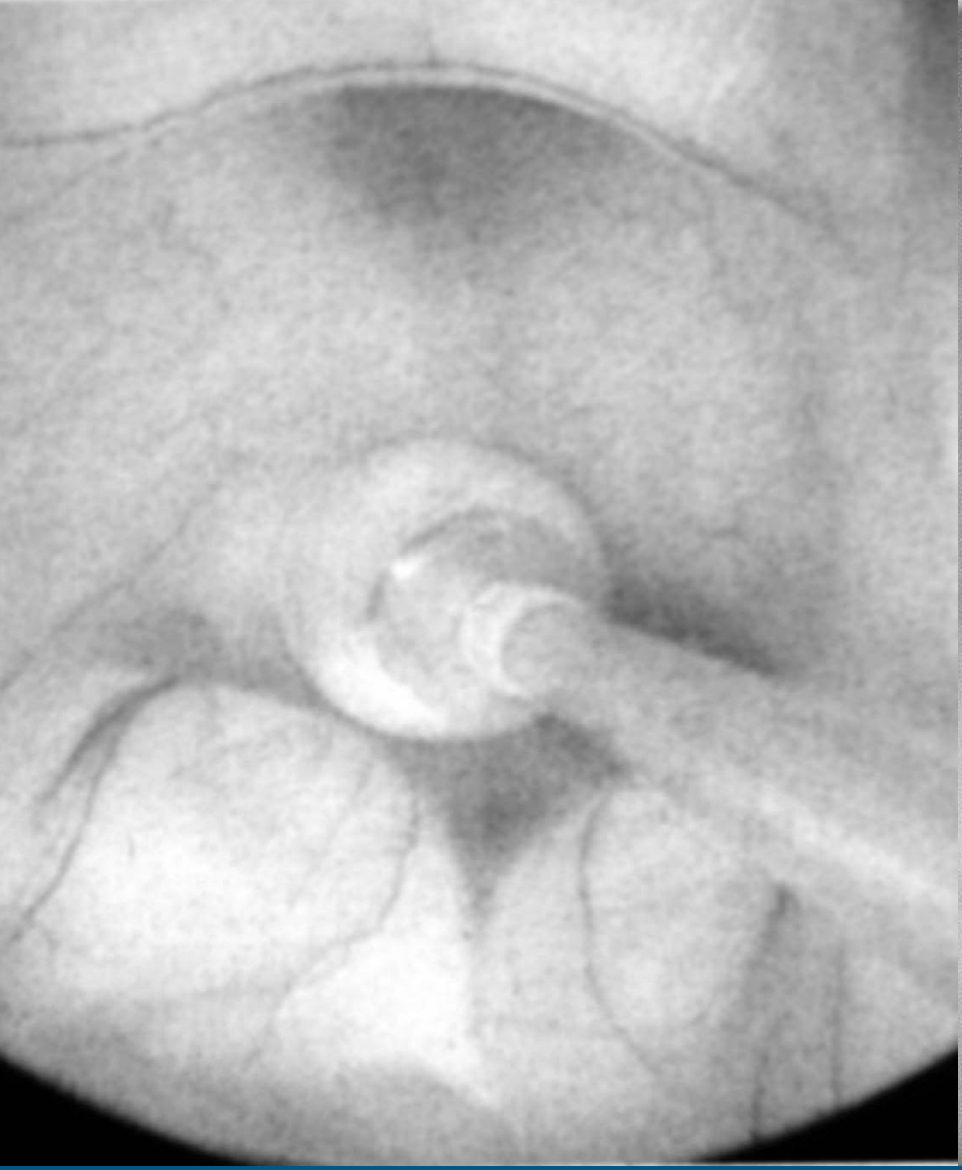
Theime (New York), 1999.

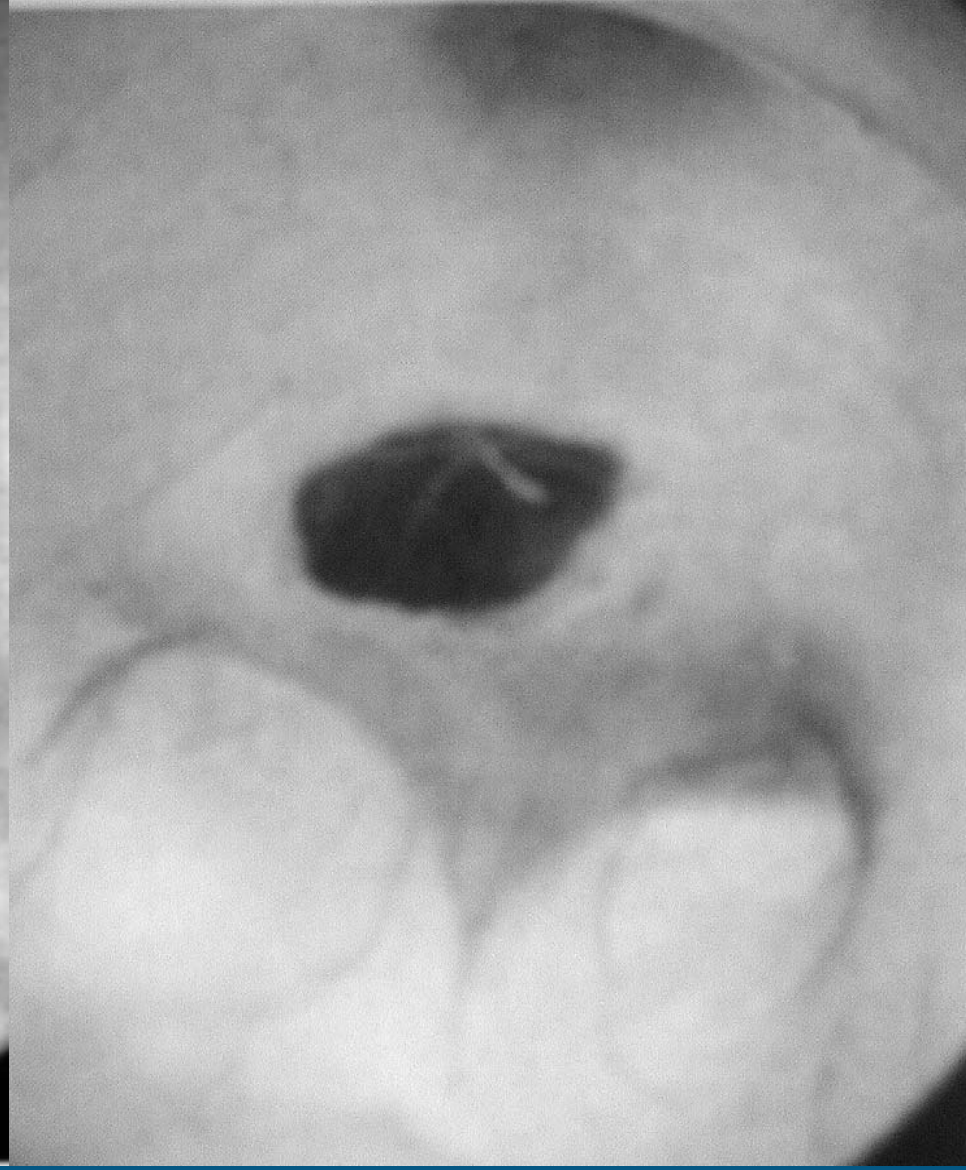
Endoscopic IIIrd Ventriculostomy

- Advantages:
  - Lowers risk of delayed malfunction
  - More physiologic than shunt
  - Low infection rate
- Disadvantages
  - Not always successful

ETV with Choroid Plexus Cauterization

- Indicated in children under a year of age
- Good success with
  - Myelomeningocele
  - Aqueductal stenosis
  - Meningitis
- May be effective with
  - IVH of prematurity
- Probably not effective with
  - Pure communicating hydrocephalus



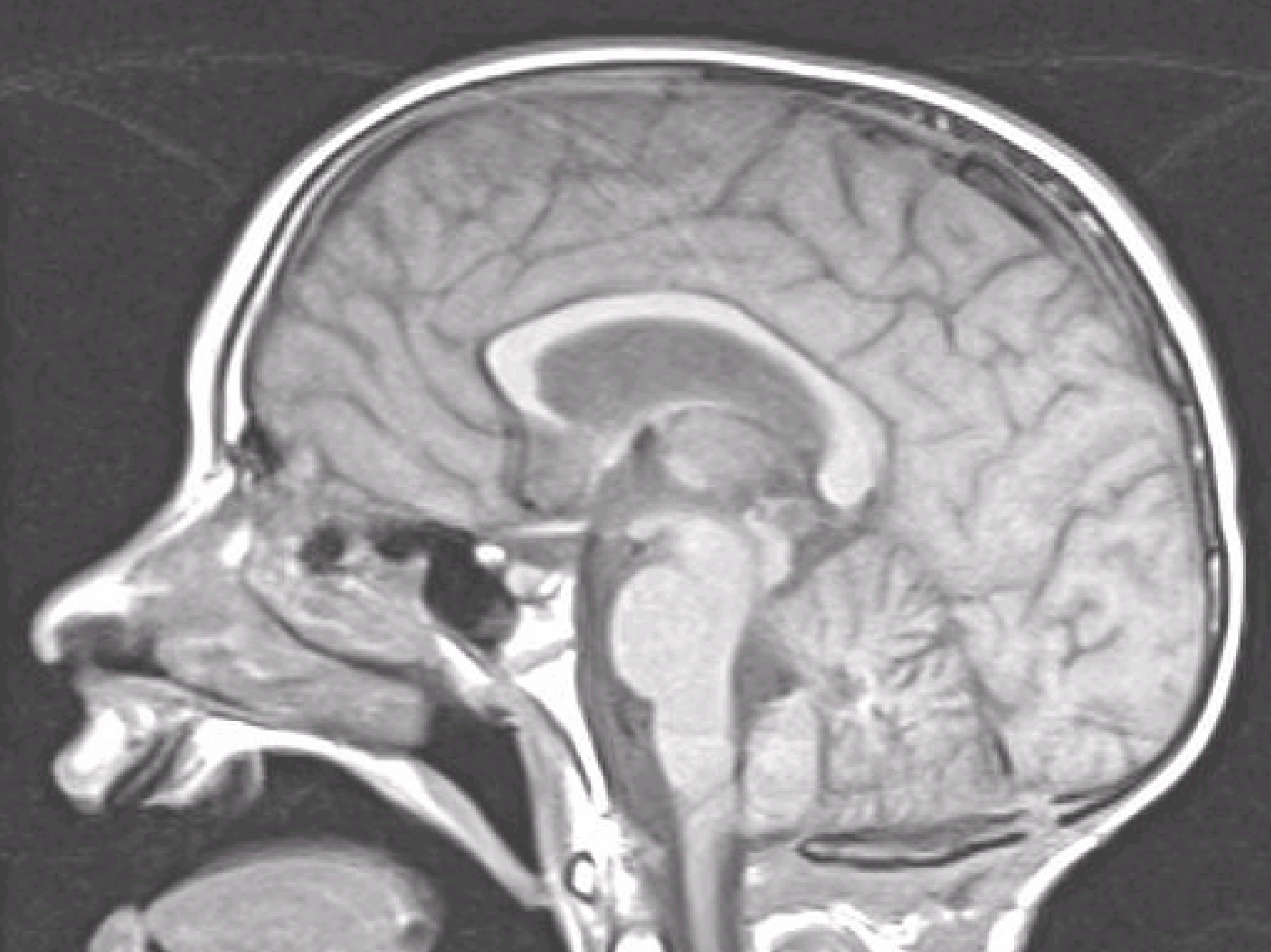


Nemours | Alfred I. duPont Hospital for Children
Nemours Children's Clinic

Principle and Practice of Pediatric Neurosurgery.

Eds.: Albright AL, Pollack IF, Adelson PD.

Theime (New York), 1999.

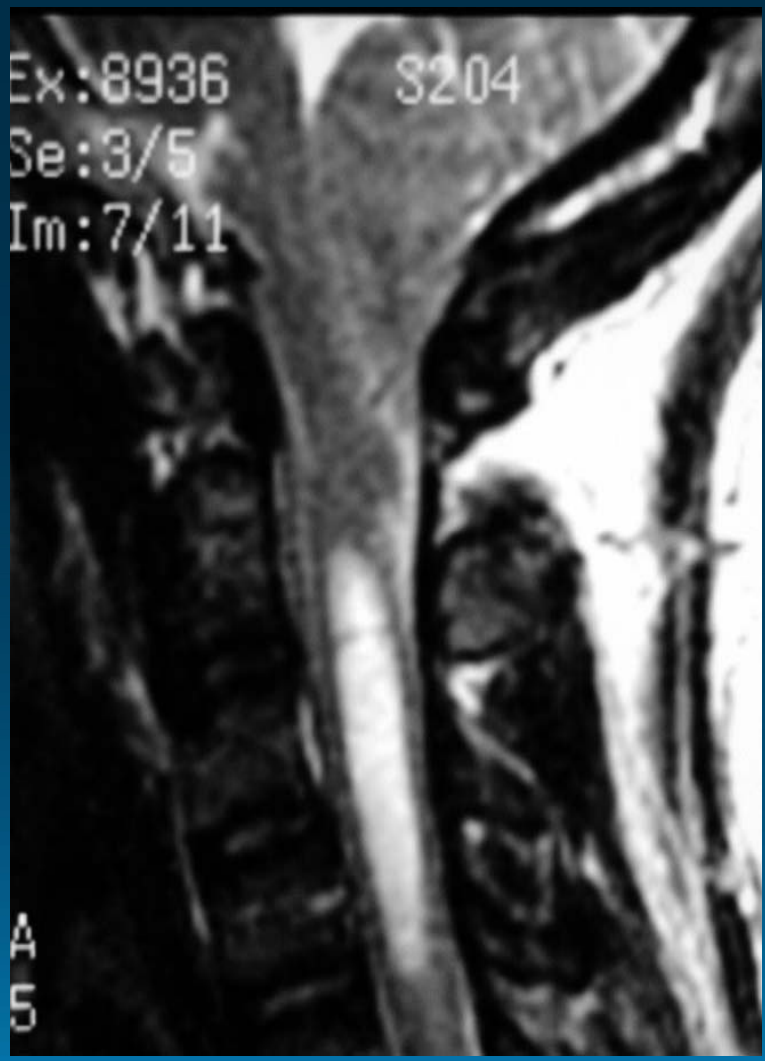


Chiari Malformation

- Congenital anomaly of cerebellum and brain stem
  - Types I to IV
- 3 of 4 types involve hindbrain herniation
- Types I and II most common

Chiari I Malformation

- Tonsilar herniation >5 mm below foramen magnum
- Brainstem in normal configuration
- High incidence of associated syrinx (50-70%)



Chiari I Malformation

- Average age of presentation is 5
  - Usually presents in childhood
- Etiology is unclear
  - Smaller posterior fossa
- Incidence unknown
- Symptoms result from brainstem/spinal cord compression and syrinx

Symptoms in Children

- Occipital headaches
- Back pain
- Limb pain
- Ataxia
- Motor or sensory loss
- Progressive scoliosis
- Bowel or bladder changes
- Hiccups
- Strabismus

Evaluation

- MRI
  - Cine MR to look for CSF flow through posterior foramen magnum
- Swallowing study
- Direct observation of vocal cords
- Sleep study
- SSEP

Treatment

- Treat hydrocephalus
  - If patient has shunt, make sure it is working
- Chiari decompression
  - Enlarge foramen magnum
  - Remove necessary cervical lamina
  - *Duroplasty*
  - *Ensure free egress of fluid from IV ventricle*

Treatment

- Syrinx usually resolves with Chiari decompression
  - May occasionally need to secondarily shunt syrinx

Conclusion

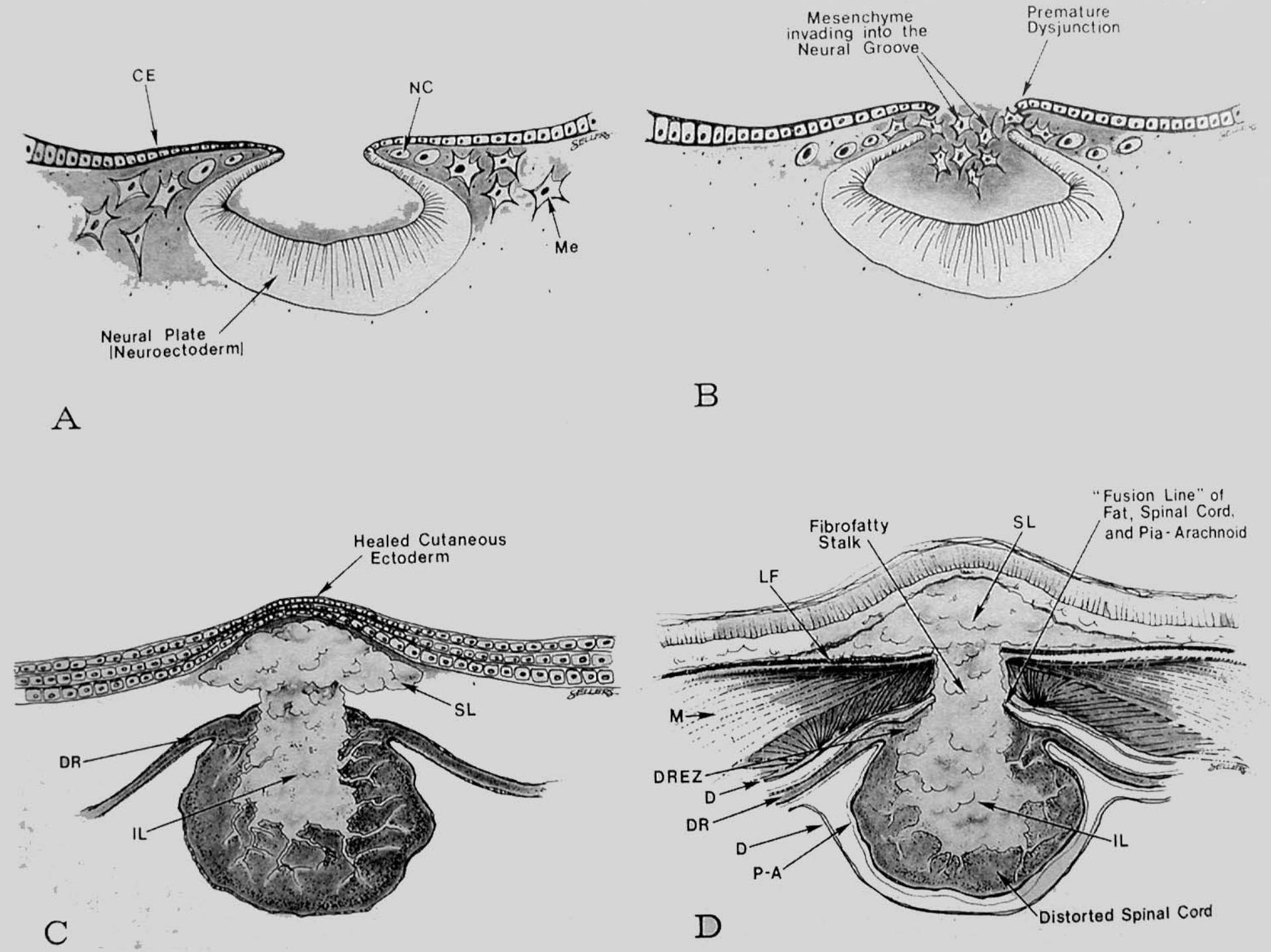
- Chiari malformations may cause progressive neurological deterioration in children
- Early diagnosis and treatment is necessary to minimize morbidity

Tethered Spinal Cord

- Spinal cord normally ends just below the rib cage
- A number of developmental anomalies can pull the spinal cord unusually low in the spinal canal
- High incidence in patients with imperforate anus
  - Roughly 25% regardless of level of lesion
  - Most of these patients have a fatty filum

Tethered cord

- Incidence not well known
  - Similar to myelomeningocele
  - Female predominance
- Embryogenesis unclear
  - Secondary neurulation
  - Early dysjunction of ectoderm


CE
NC
Me
Neural Plate
(Neuroectoderm)
A
Mesenchyme
invading into the
Neural Groove
Premature
Dysjunction
B
Healed Cutaneous
Ectoderm
SL
DR
IL
C
Fibrofatty
Stalk
SL
"Fusion Line" of
Fat, Spinal Cord,
and Pia-Arachnoid
LF
M
DREZ
D
DR
D
P-A
IL
Distorted Spinal Cord
D

Occult Spinal Dysraphism

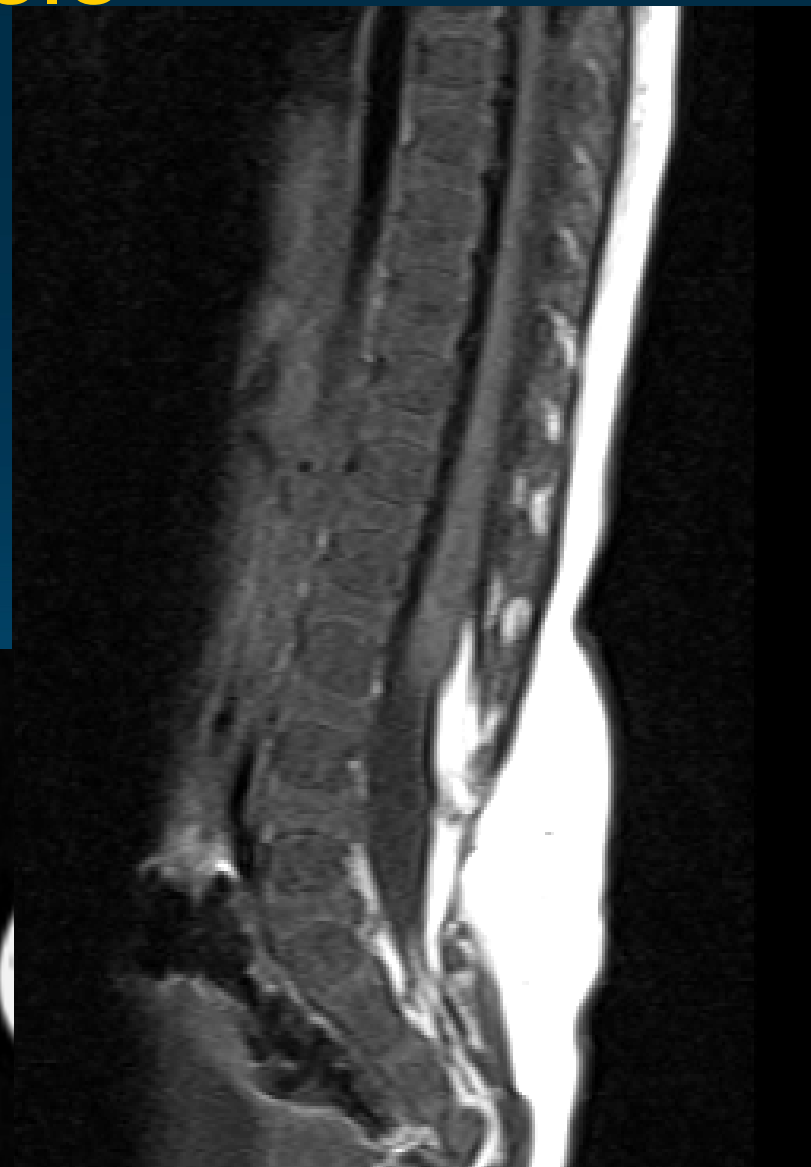
- Tight and fatty filum terminale
- Lipomyelomeningocele
- Split cord malformation
- Dermoid cyst

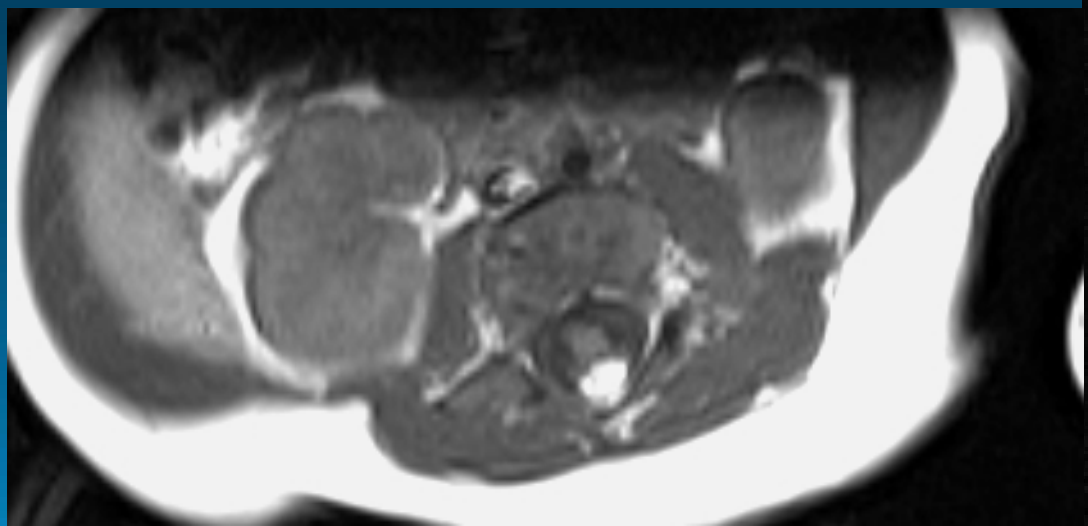
Occult Spinal Dysraphism Cutaneous Stigmata

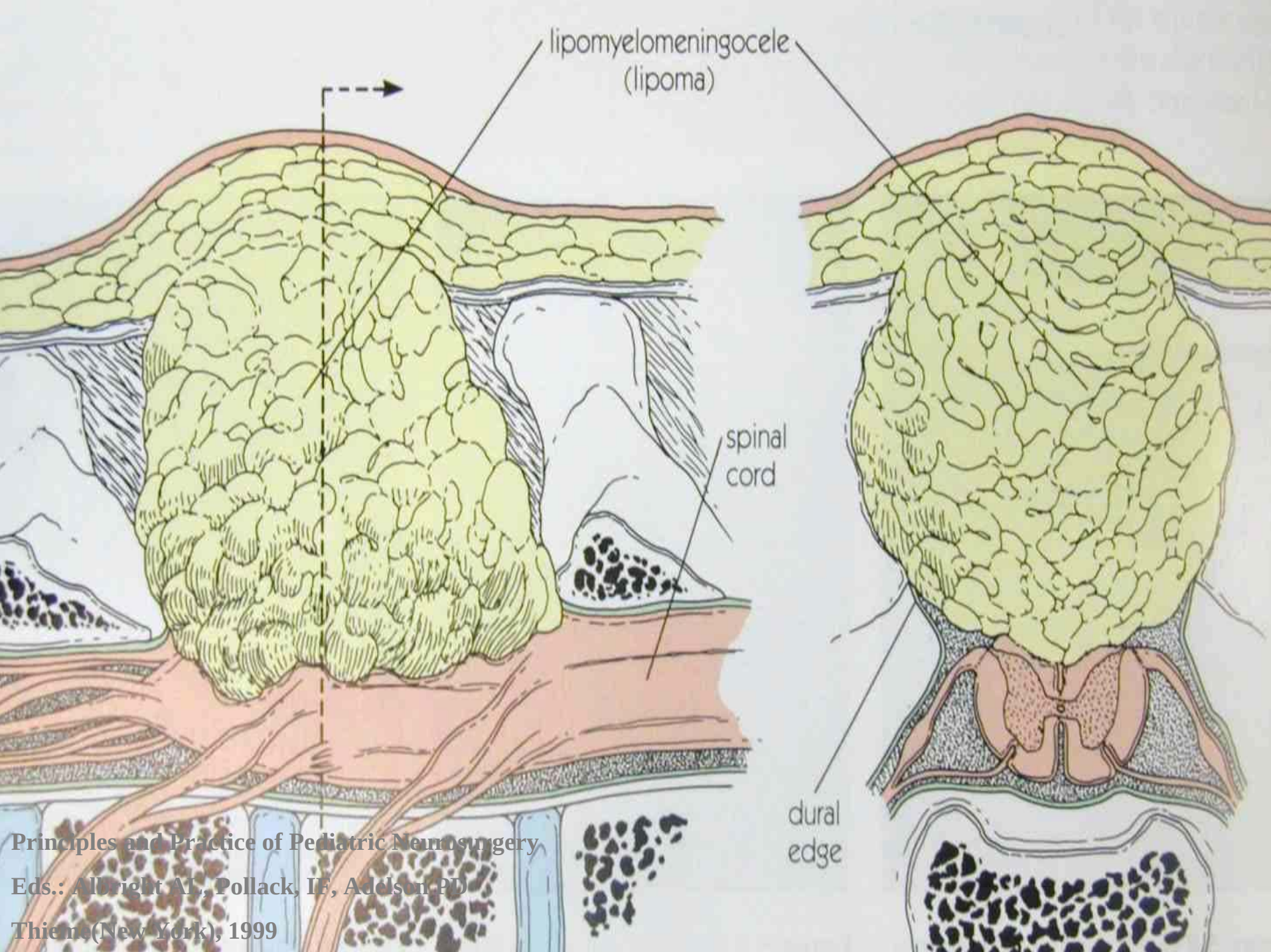
- Hairy patch
  - Associated with split cord malformations
- Birthmark
  - Associated with fatty filum or lipomyelomeningocele
- Dimple
  - Associated with dermoid cyst
- Forking or deviation of gluteal cleft

Lipomyelomeningocele

- Low and dorsal conus
- Fat attached to dorsal aspect of spinal cord
  - Extends through fascial defect to subcutaneous fat



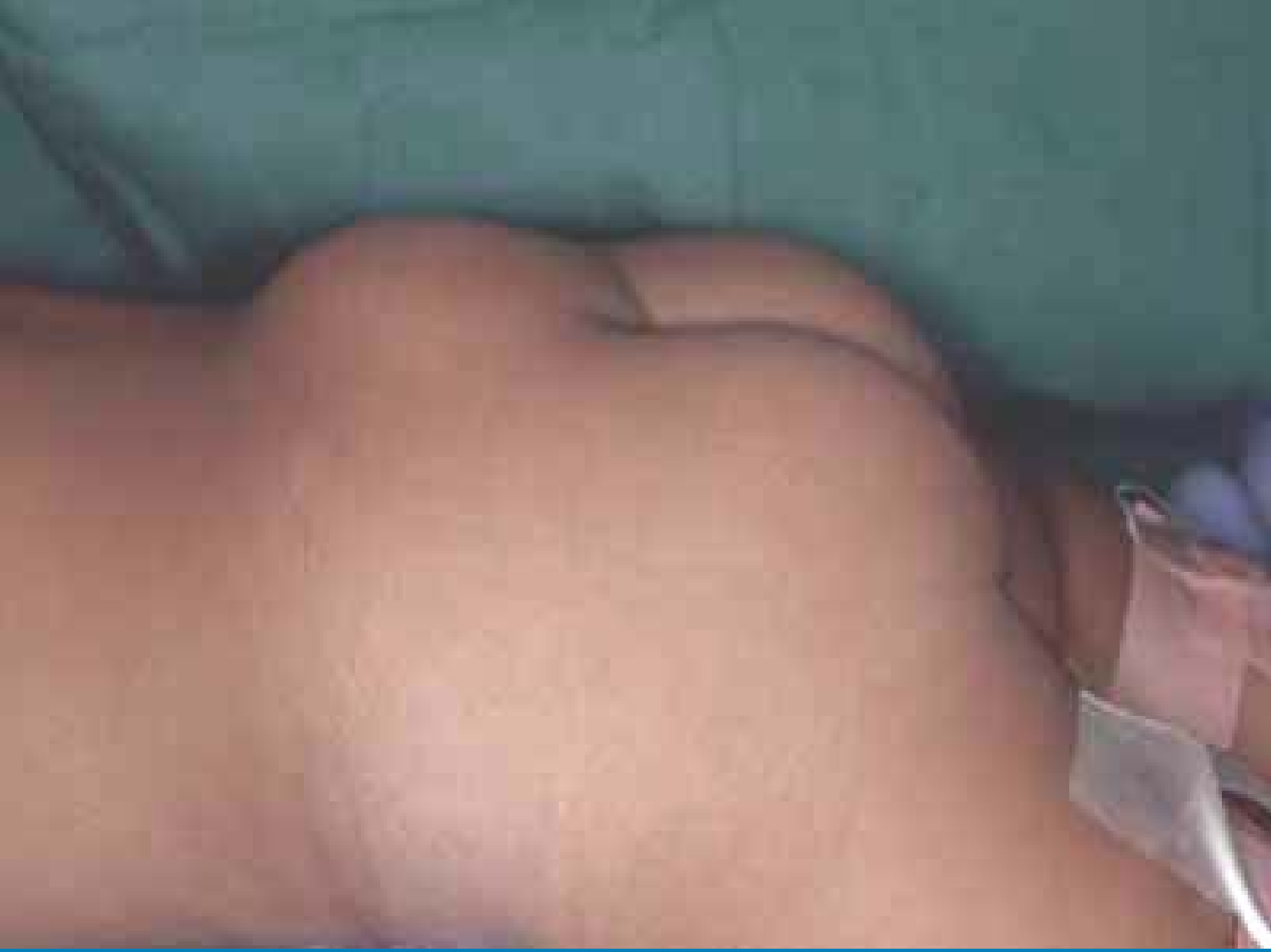


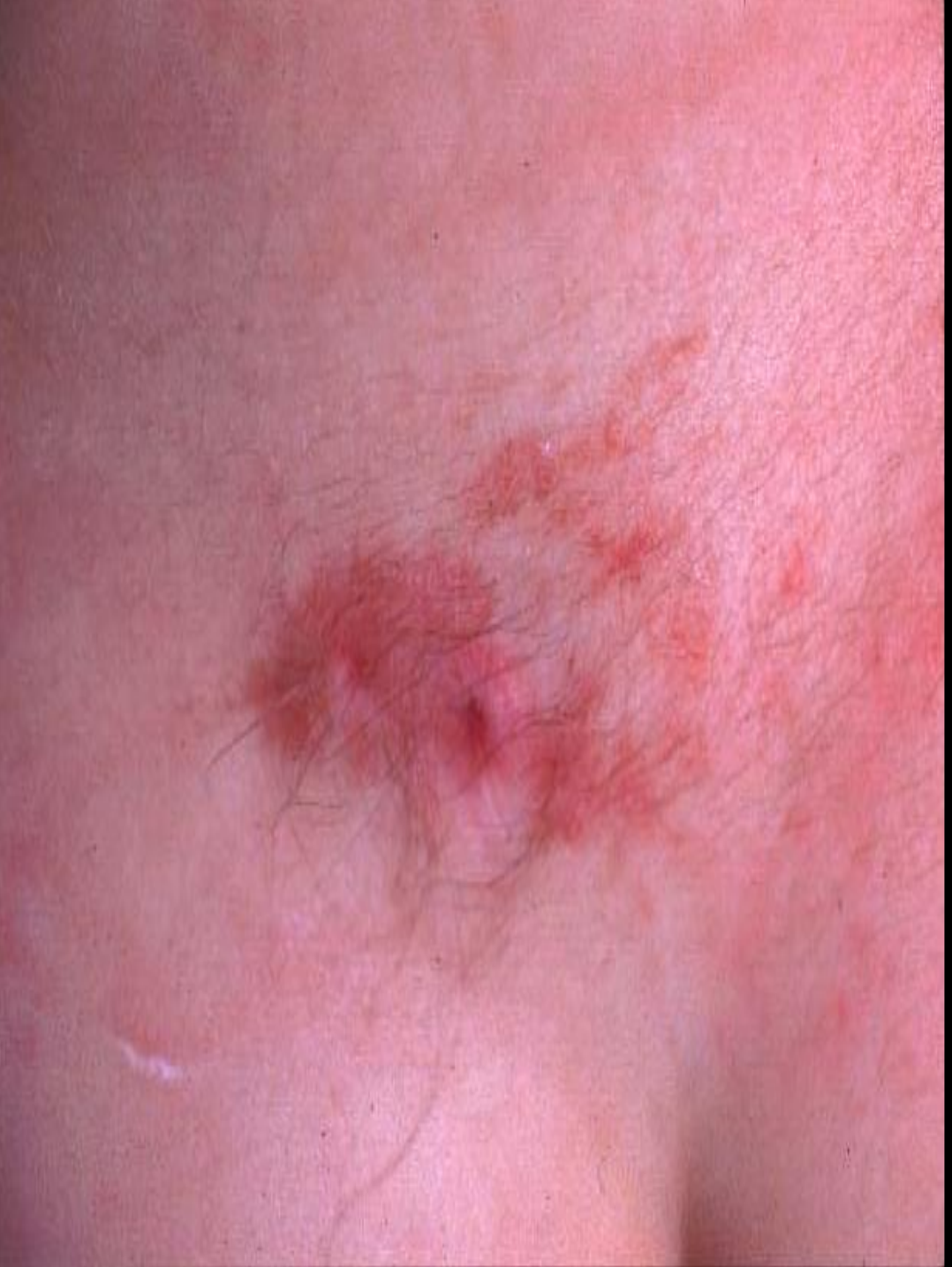


Principles and Practice of Pediatric Neurosurgery

Eds.: Albright AL, Pollack, IE, Adelson PD

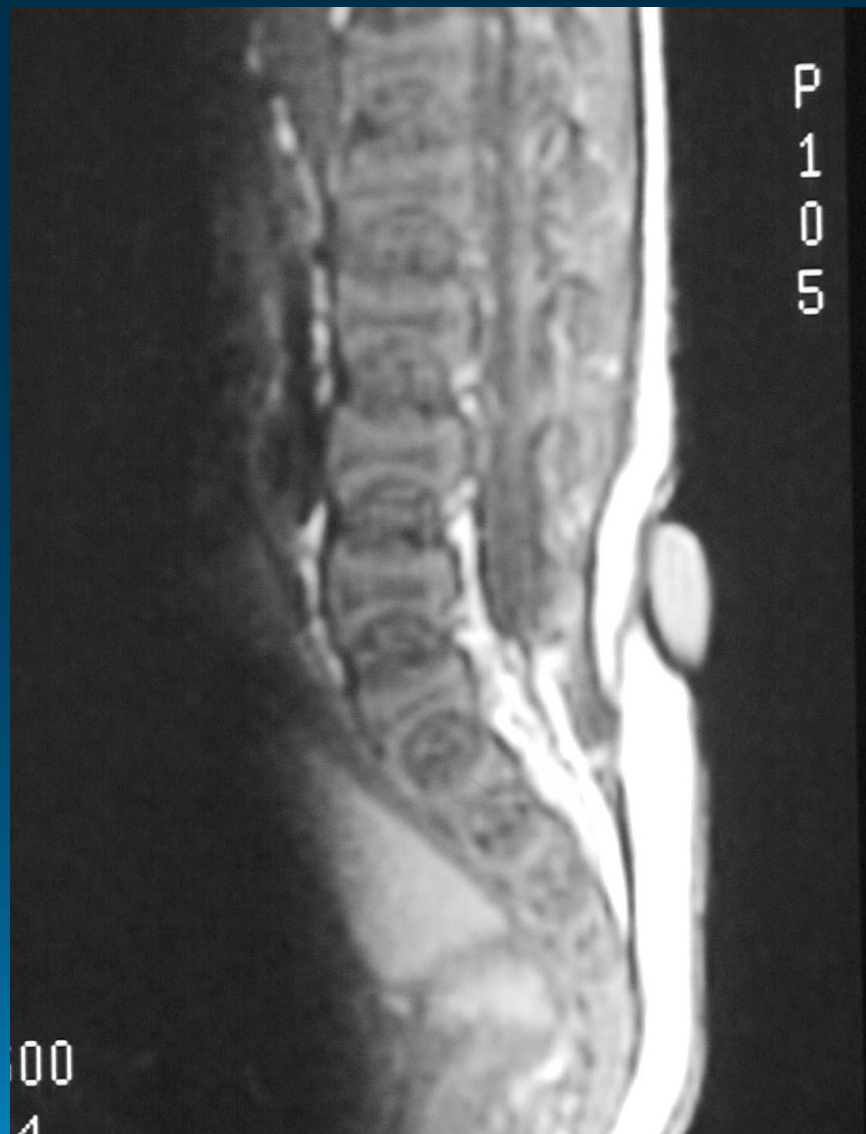
Thieme(New York), 1999

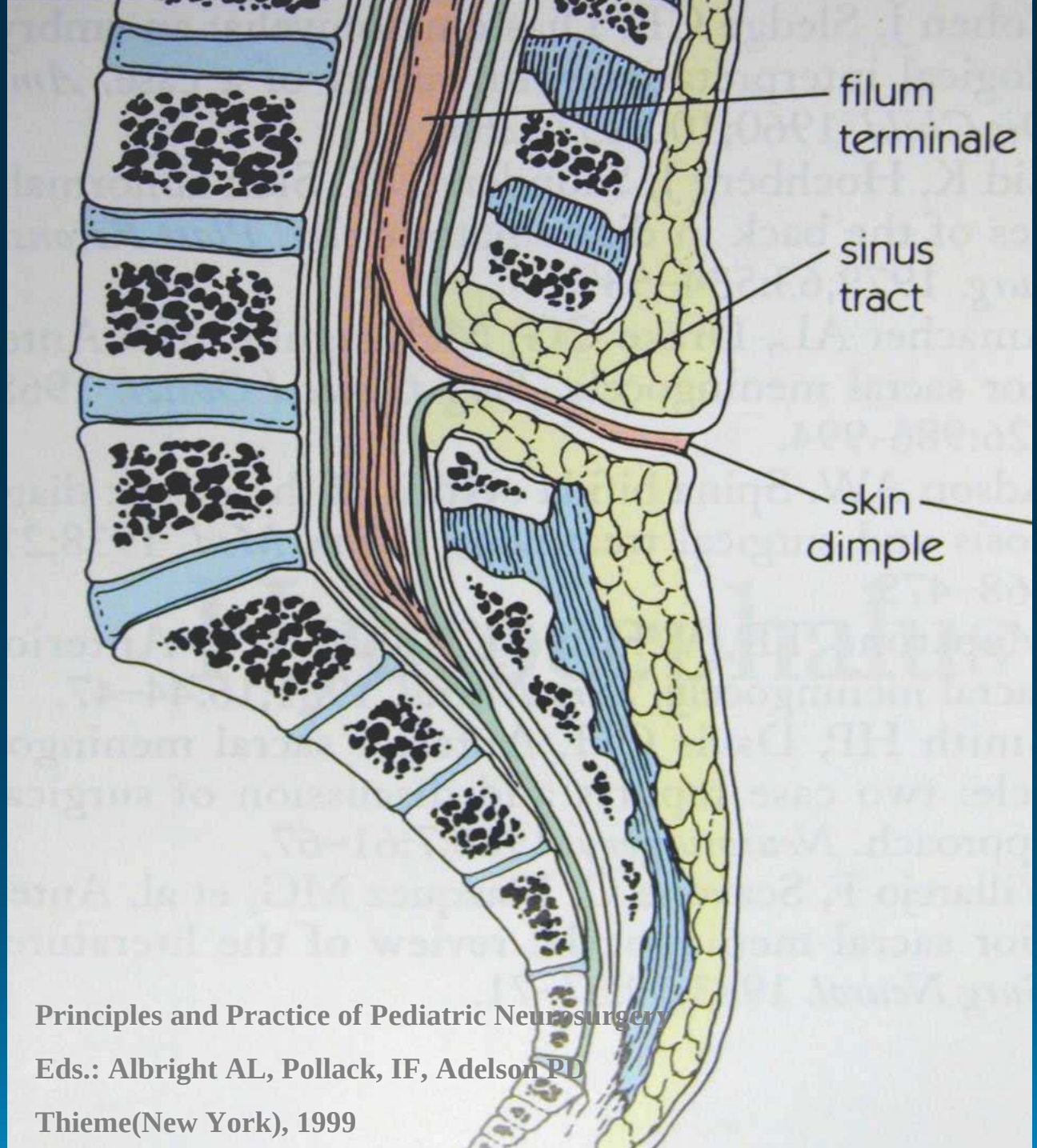





Dermal Sinus

- Dermoid tumor adherent to spinal cord
- Dermal sinus tract extending through fascial defect
- Dimple evident on skin
  - Coccygeal dimples not associated with intraspinal pathology

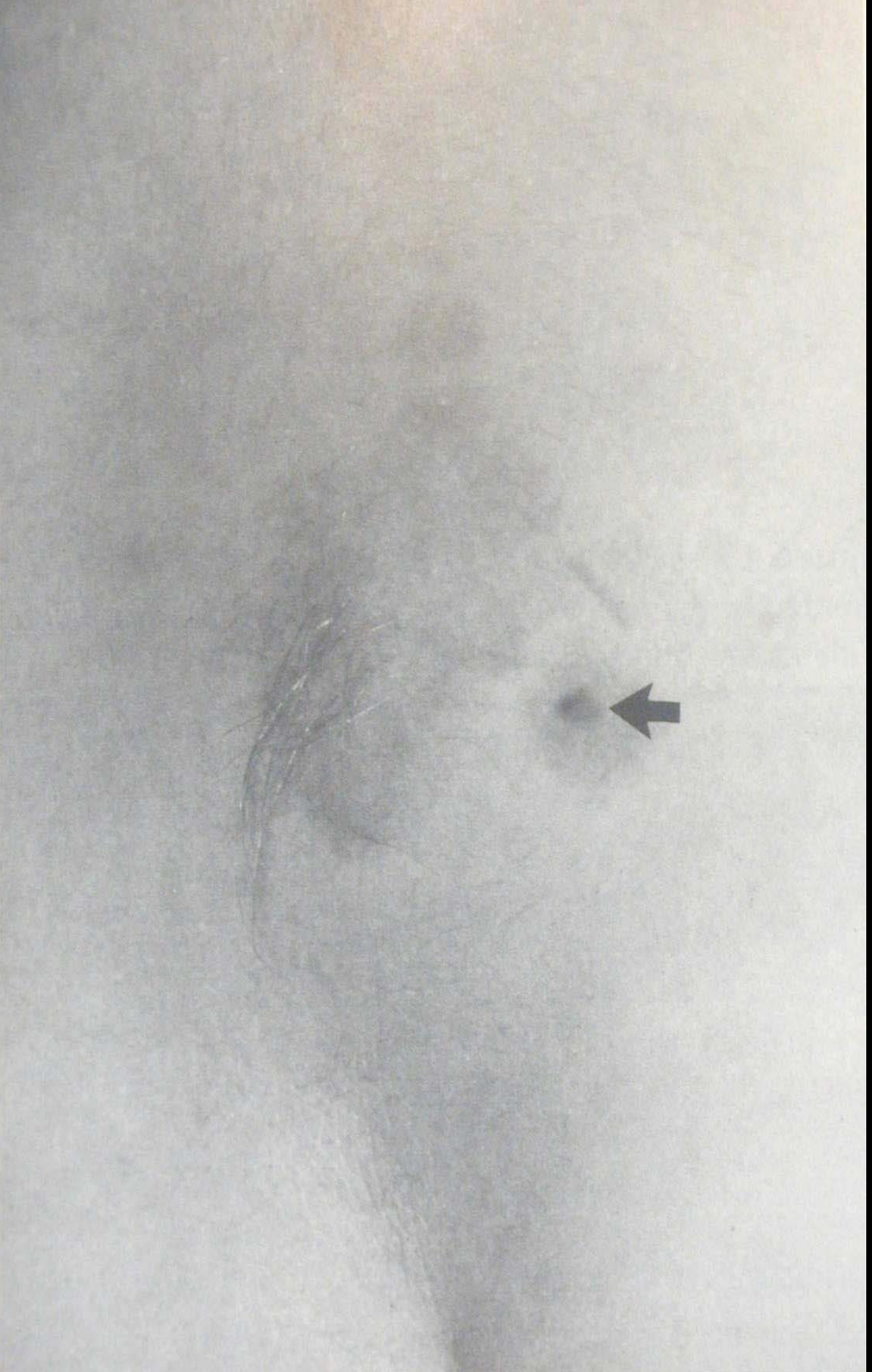


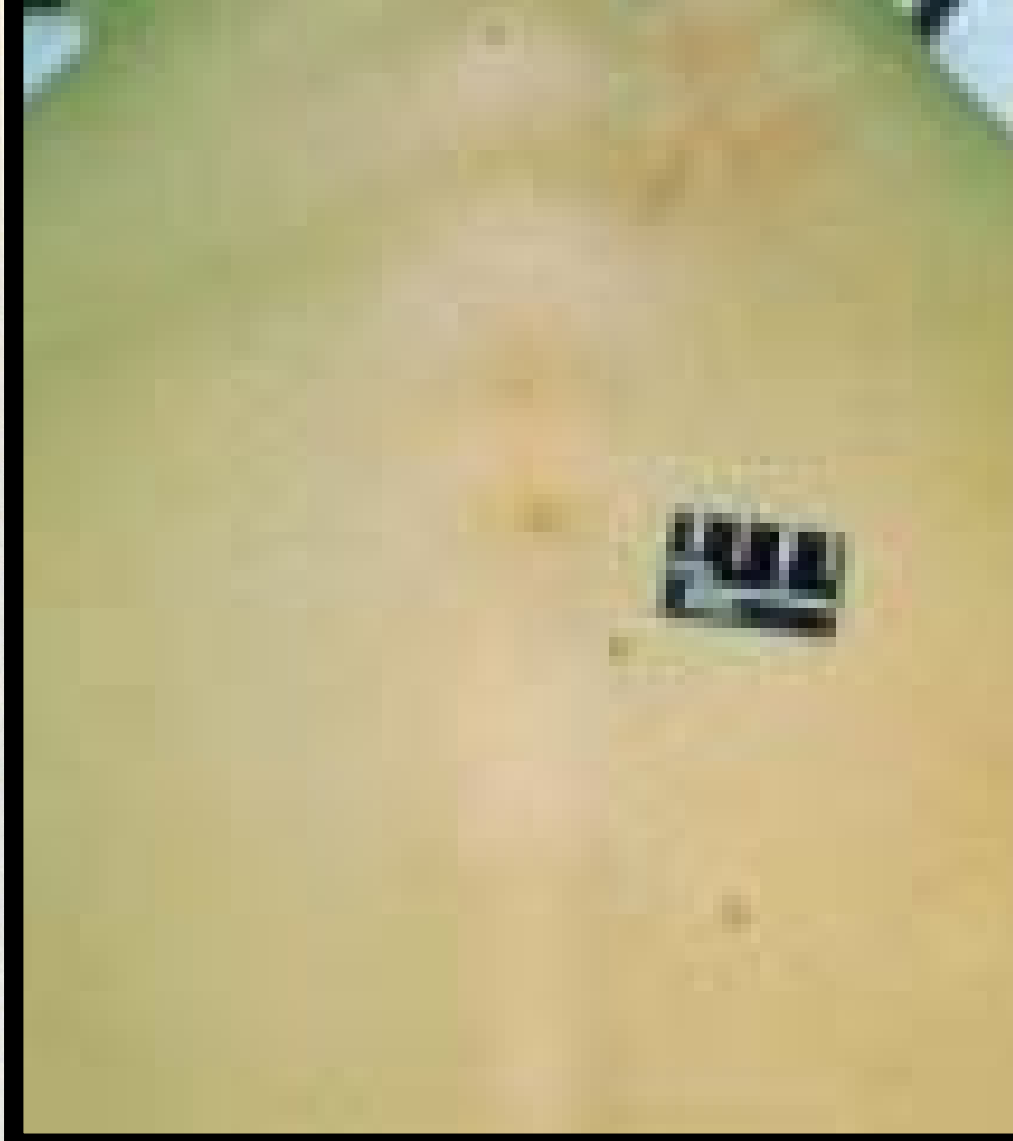


Principles and Practice of Pediatric Neurosurgery

Eds.: Albright AL, Pollack, IF, Adelson PD

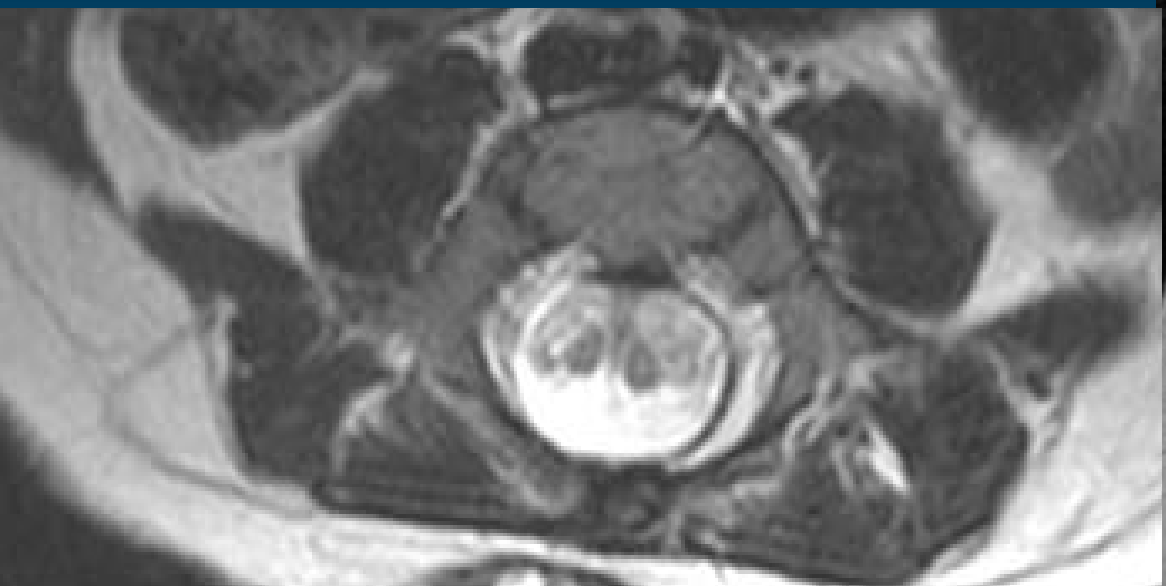
Thieme(New York), 1999

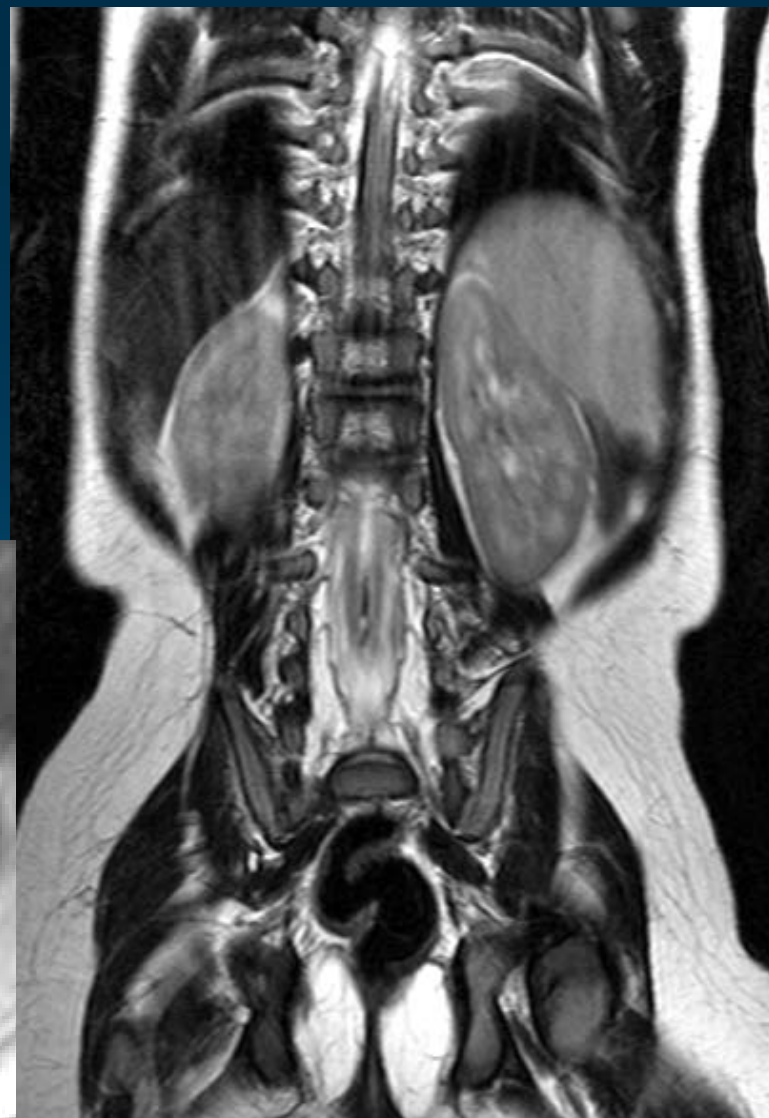


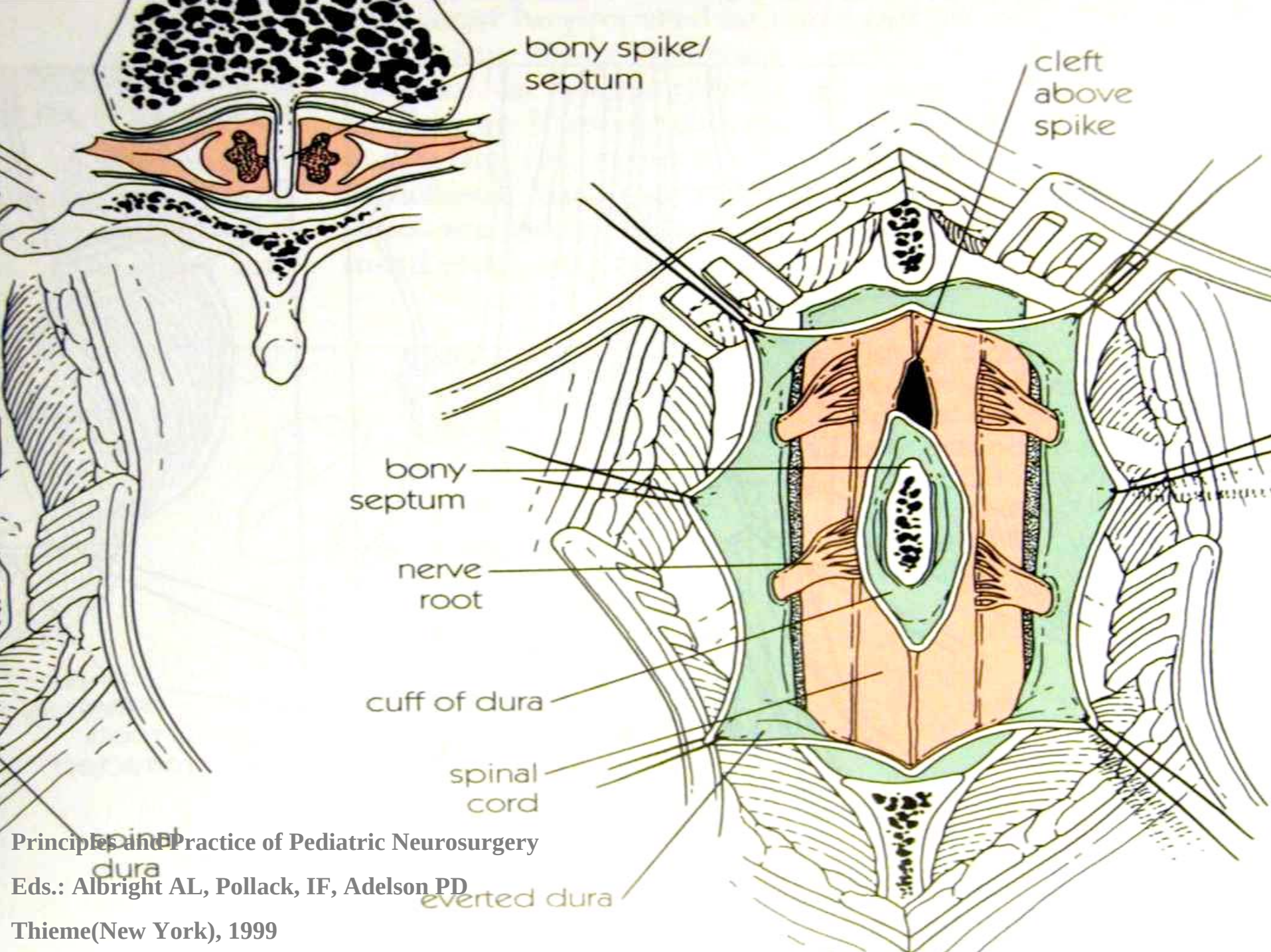


Split Cord Malformation

- Double spinal cord
- Midline septum
  - Bone or fibrous bands
- One or two dural sacs



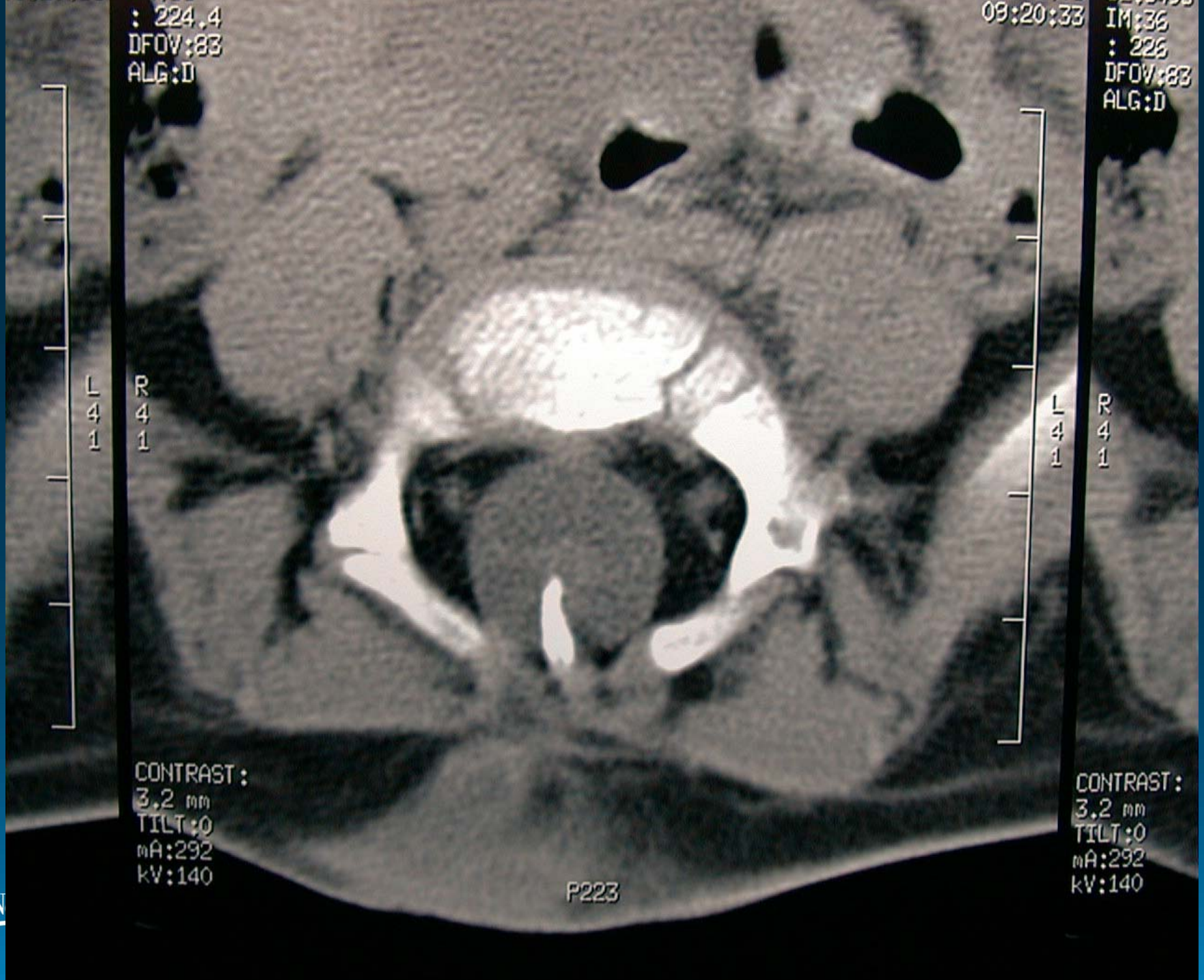


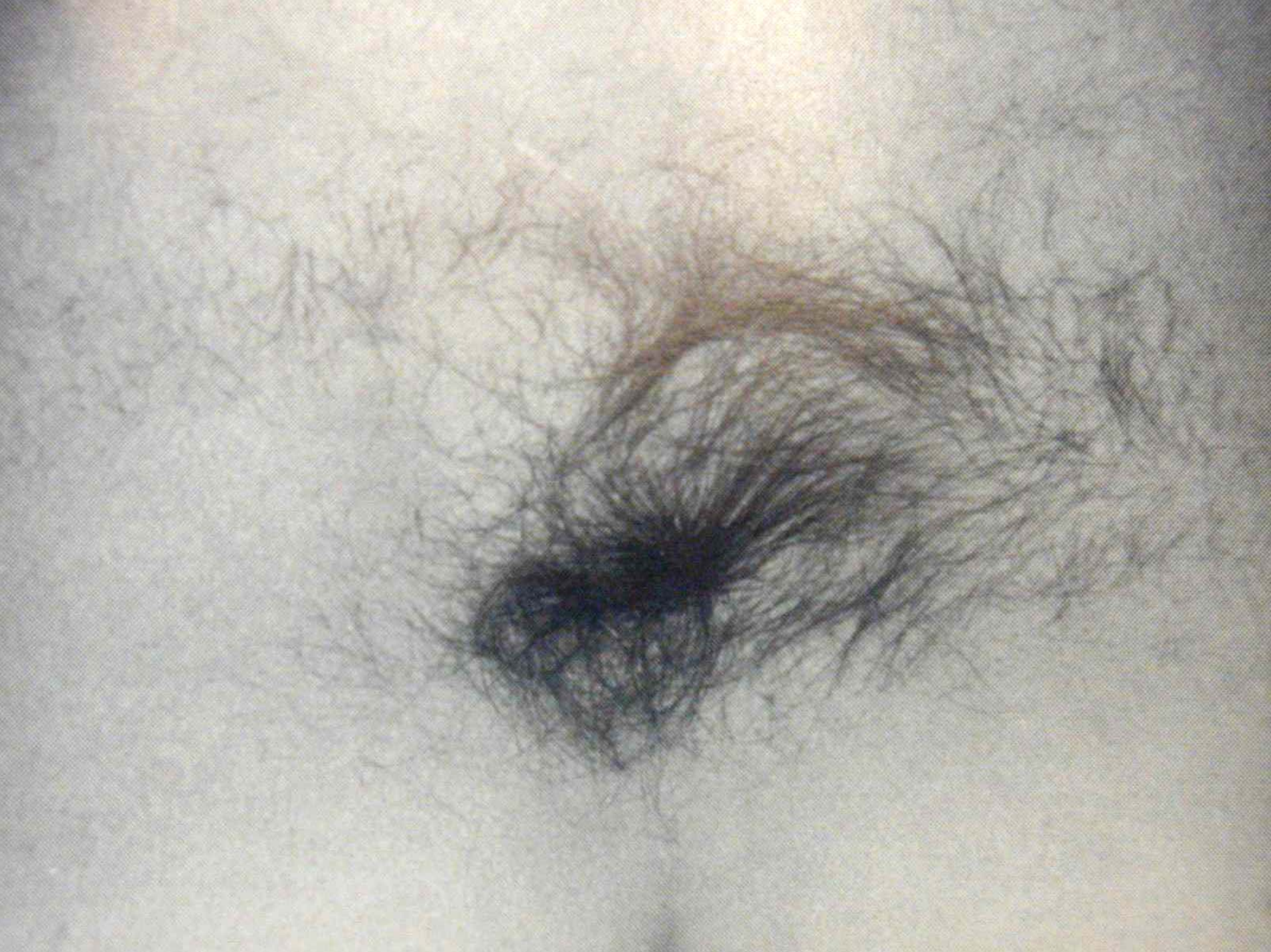


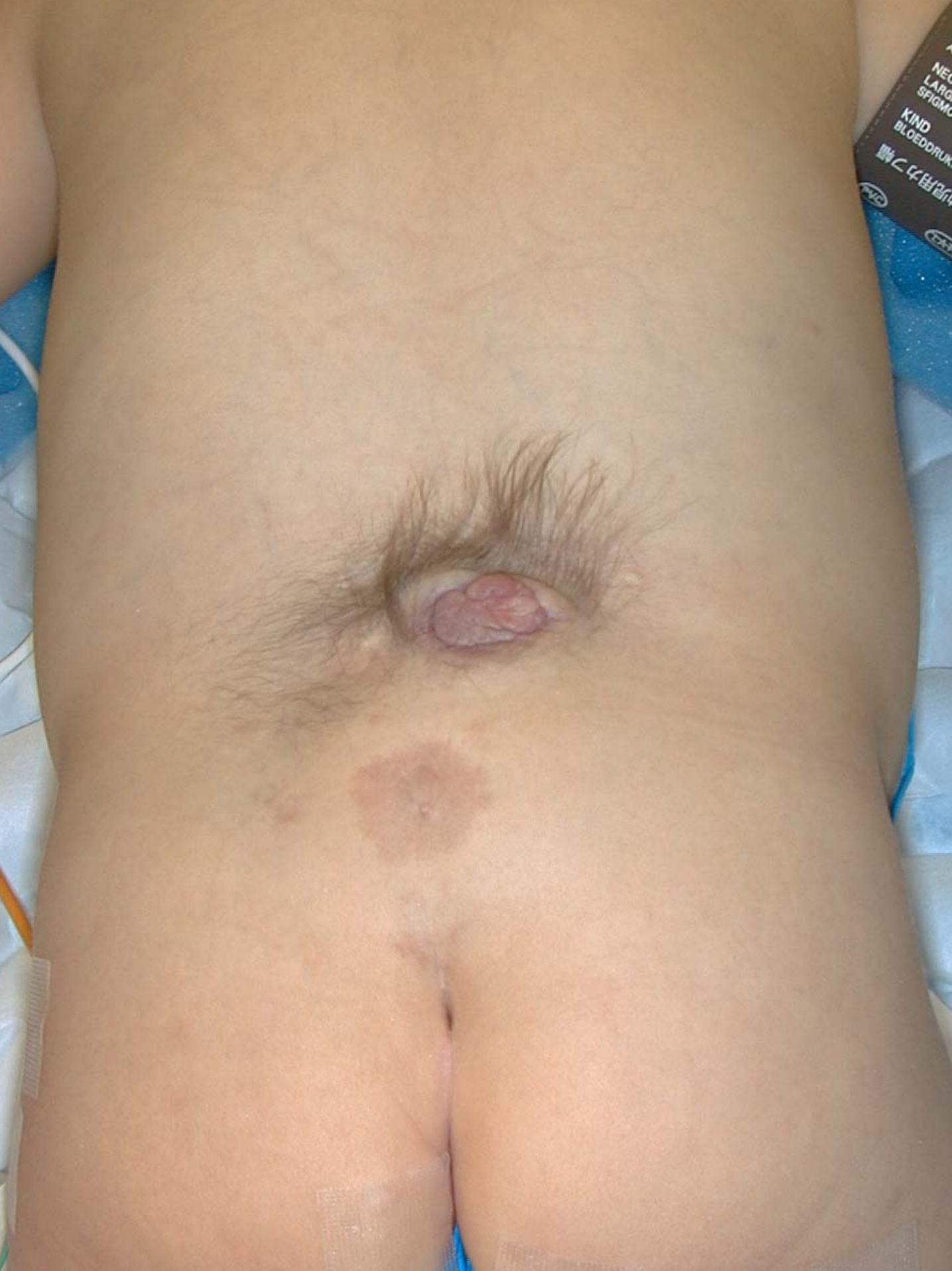
Principles and Practice of Pediatric Neurosurgery

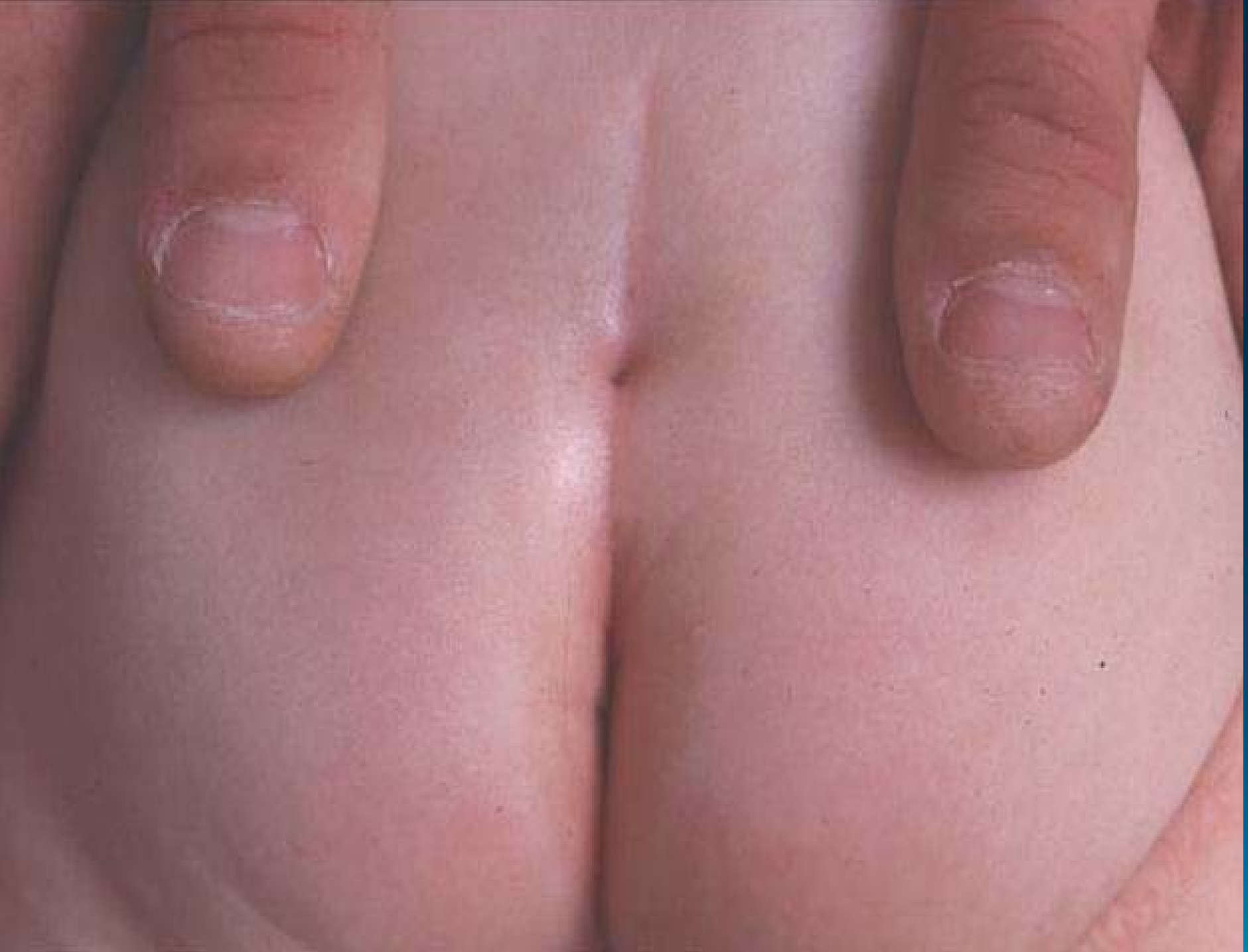
Eds.: Albright AL, Pollack, IF, Adelson PD

Thieme(New York), 1999


: 224.4
DFOV:83
ALG:D
09:20:33
IM:36
: 226
DFOV:83
ALG:D
L
4
1
R
4
1
L
4
1
R
4
1
CONTRAST:
3.2 mm
TILT:0
mA:292
kV:140
P223
CONTRAST:
3.2 mm
TILT:0
mA:292
kV:140



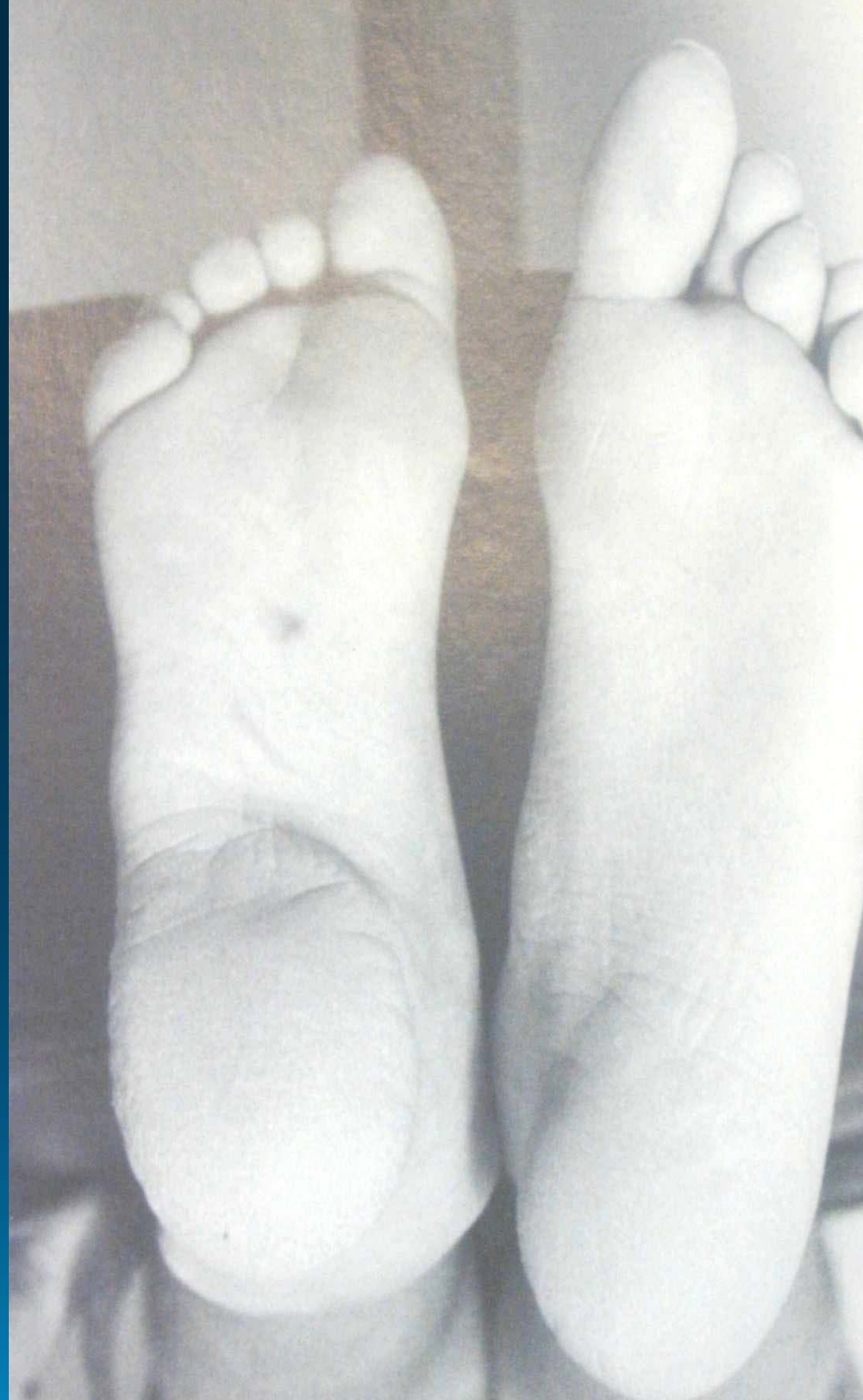

KIND



Occult Spinal Dysraphism Neurological Symptoms

- Leg atrophy or weakness
  - Foot asymmetry
- Gait abnormality or delay in walking
- Motor or sensory dysfunction
- Back or leg pain
- Spasticity or hyperreflexia

Foot Asymmetry



Nemours | Alfred I. duPont Hospital for Children
Nemours Children's Clinic

Occult Spinal Dysraphism
Other Symptoms

- Urological symptoms
  - Urinary tract infections
  - Incontinence
  - Neurogenic bladder
- Orthopedic deformities
  - scoliosis

Evaluation

- MRI
  - Level of conus
  - Fat or tumor
- Ultrasound
  - Level of conus
  - Cord movement
- Urodynamics

Treatment

- Surgical release of tethered cord when diagnosed at early age
  - Likelihood of deterioration
- Timing of surgery depending on etiology
  - Dermal sinus
  - Other
- Surgical release in adults is debated topic

Treatment

- Complexity and morbidity of surgery depends on etiology
  - Tight filum versus lipomyelomeningocele
- Intraoperative electrophysiological monitoring to identify nerve roots

Outcome

- Preoperative deficits will unreliably improve
  - Indication for prophylactic surgery
- Risks include
  - Neurological worsening
  - CSF leak
  - Re-tethering

Conclusion

- Some neurological disorders have cutaneous stigmata that can be recognized
  - Phakomatoses
  - Spinal dysraphism
- Early recognition of disorder can improve treatment of child

Nemours

Alfred I. duPont Hospital for Children

Nemours Children's Clinic