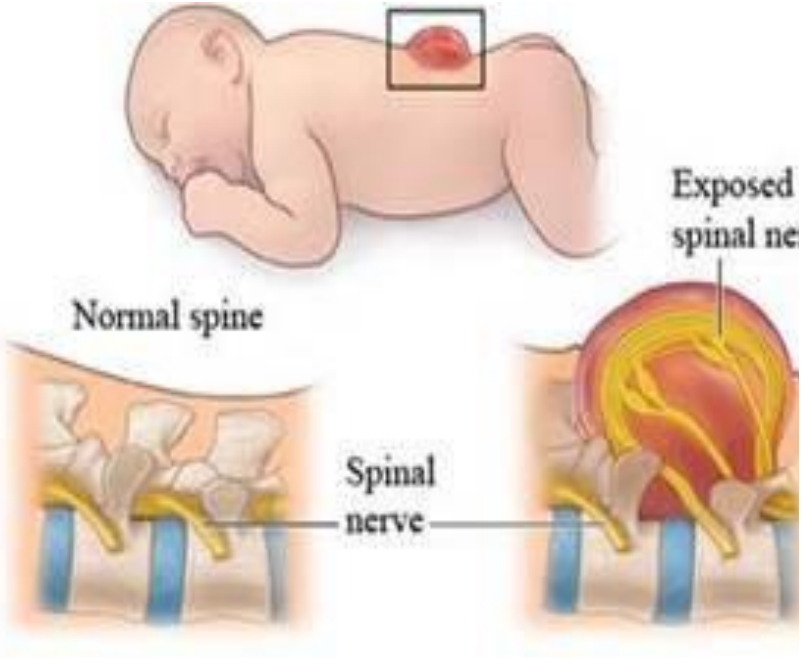
Congenital Compression of Spinal Cord

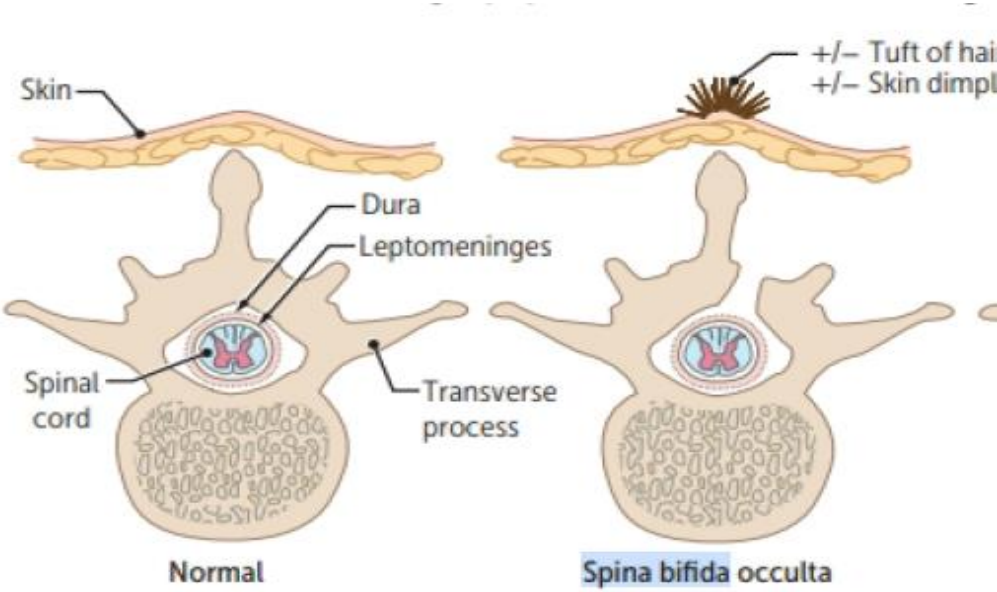
Contents

SPINA BIFIDA

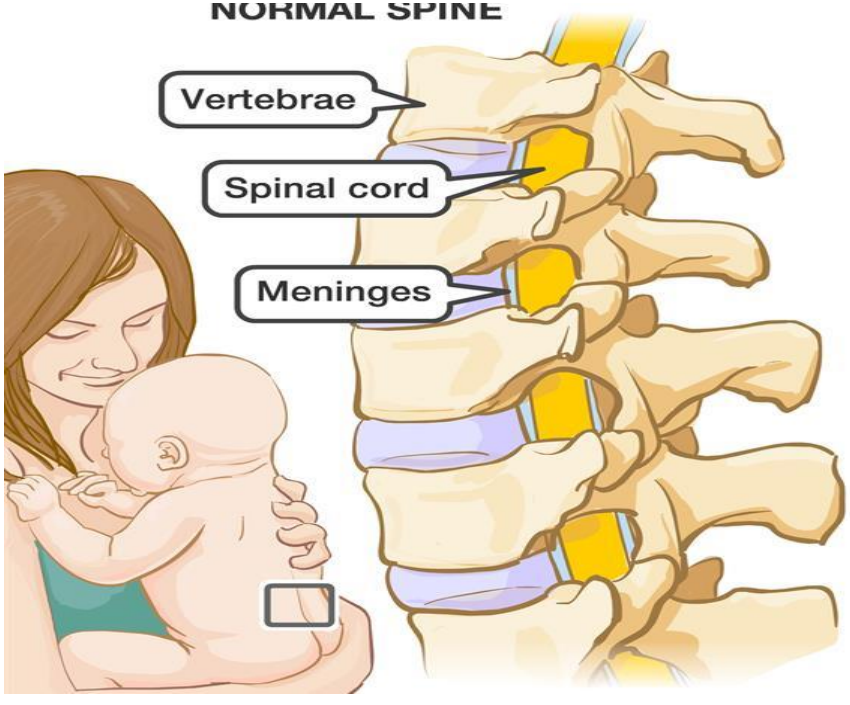
Spina Bifida

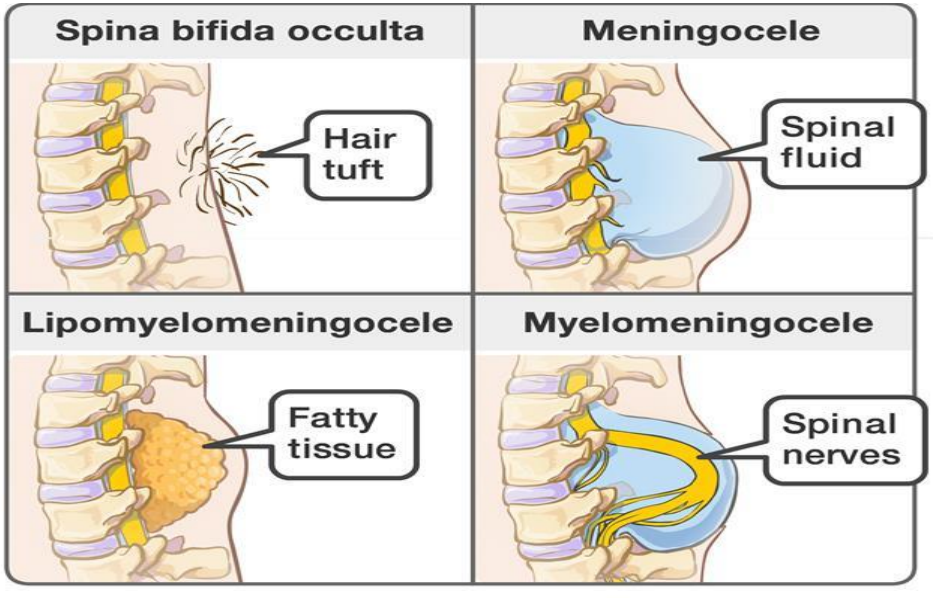
- A developmental disorder due to incomplete closure of embryonic neural tube.
- Protrusion of spinal cord due to formation of an opening.
- Most common type is myelomeningocoele.
- Most likely to appear in females.
- Failure of neural tube closure during the first month of embryonic life.



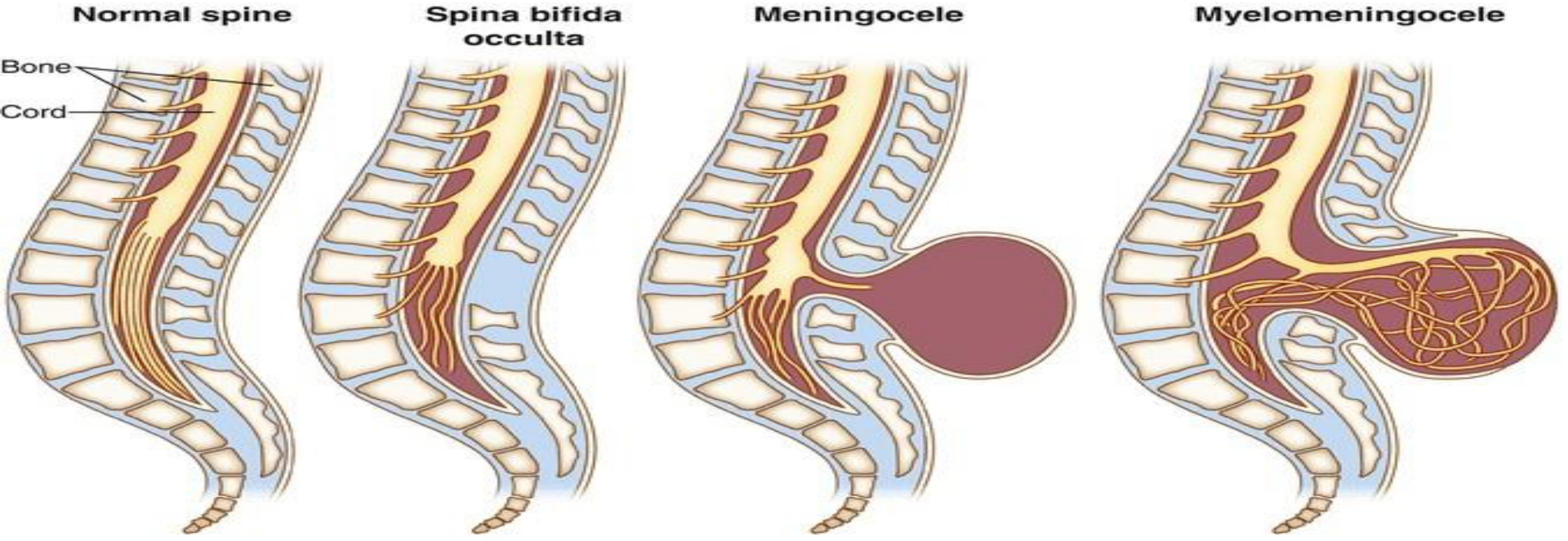


- Prevalence is 1-10 children per 1000 children worldwide.
- Incomplete closure of the neural tube approximately 28th day of gestation.
- 90-95% children born with spina bifida don’t have parental history.
- Most common factors include decrease in dietary folate intake, hyperthermia, pre-gestational maternal diabetes mellitus, etc.
- Diagnosis is made by the pre-ntatal measurement of alpha fetal protein in maternal serum at 16 weeks of gestation or by ultrasound of the fetus at 18-20 weeks of gestation.





- Most common locations are lumbar and sacral region.
- Spina bifida occulta being the mildest form occuring in 10% population.
- Meningocele least common form with posterior meningocele, meninges forced into gap.
- Associated abnormality of the brain called Arnold Chiari II malformations.
- Myelomeningocele most severe from, meninges cover the spinal sac enclosing spinal elements.
- Major complications include higher mental function abnormalities especially learning disabilities and hydrocephalus.



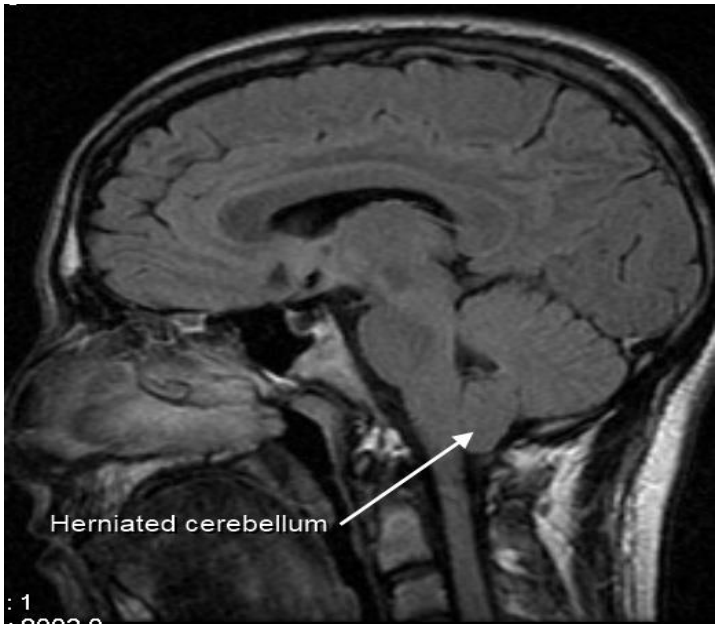
PT Treatment

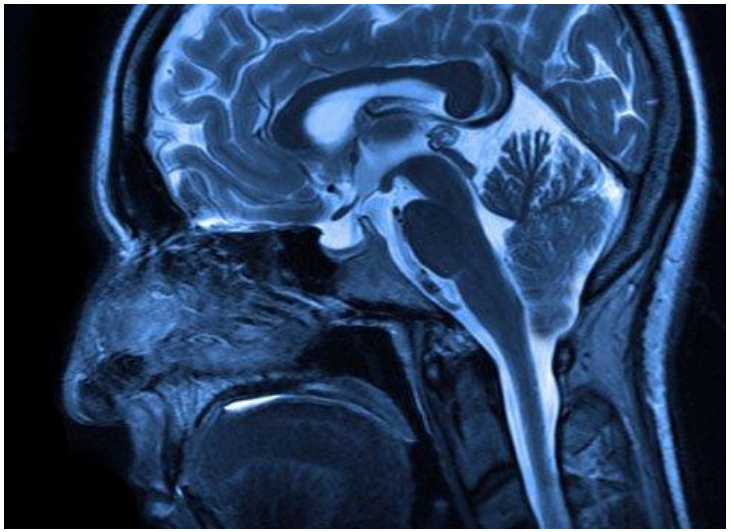
- Passive mobilization of hip and knee joints.
- Ambulation initiated with assistance.
- Upper extremity strength training with manual resistances and thera-bands.
- Progressive resistance exercises (PRE) training for strength and power- 45-50 minutes.
- Cycle ergometry and graded treadmill.
- Aerobic exercises through virtual reality.
- Use of wheelchairs, braces and crutches.

CHIARI MALFORMATION

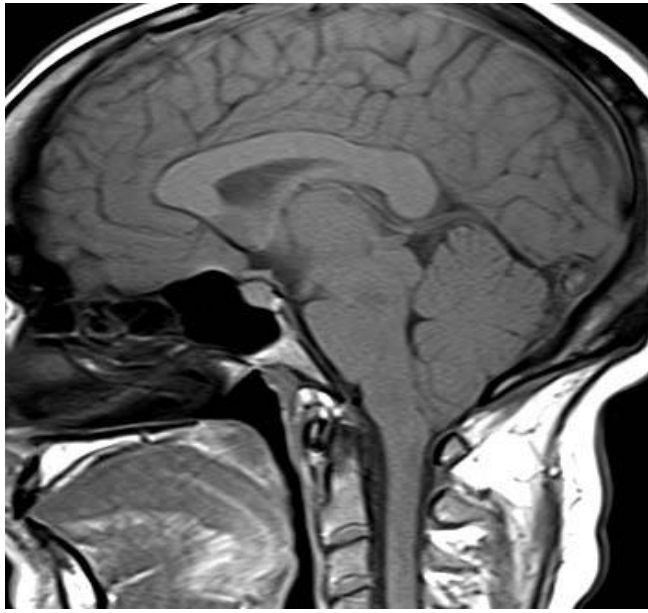
Chiari Malformations

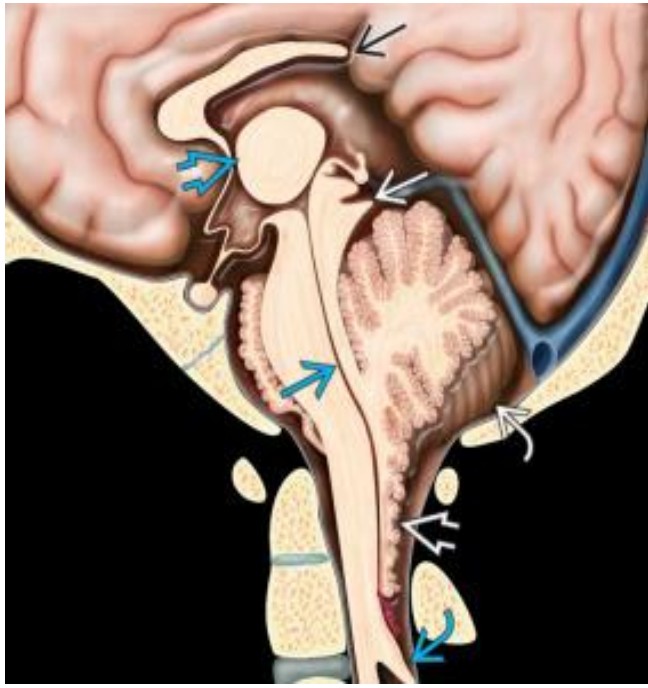
- Pathological herniation of hindbrain through foramen magnum intruding into cervical spinal column.
- Tongue like projections into cervical spine cause compression of the cord.
- Majorly congenital.
- Due to anomaly in chromosome 9 and 15.
- There are basic four subtypes: Chiari I, II, III & IV
- Chiari O is recognized by no herniation and posterior fossa decompression.



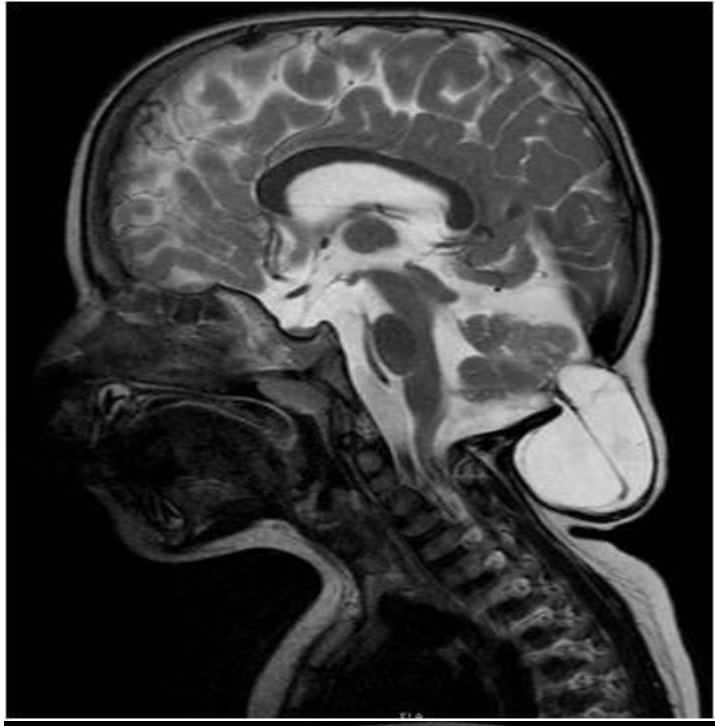


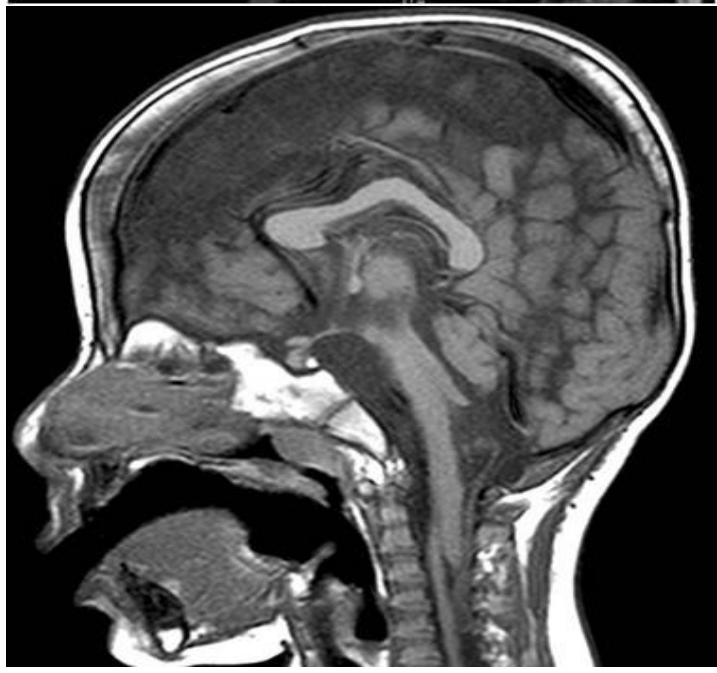
- Named after Hans Chiari.
- Chiari I, II and III described in 1891 and Chiari IV in 1896.
- Tonsilar herniation below the level of foramen magnum is called Chiari Type I.
- Caudal herniation of brainstem, cerebellar vermis and fourth ventricle is termed as Chiari type II also called Arnold Chiari Malformation.
- Hydrocephalus and Syringohydromyelia are common in type II.
- Chiari type III seen with occipital encephalocele with same anomaly as type II.
- Chiari type IV recognized as hypoplasia/ aplasia of cerebellum with no hindbrain herniation with spina bifida.





- In Chiari I, symptoms often present in adulthood, hydrocephalus seen in 10-20% cases.
- Tonsillar descent of 5mm is termed as Chiari Malformation I.
- Chiari II comes always with neural tube defect like myelomeningocele and encephalocele and hydrocephalus in 90% cases with common syrinx.
- Chiari III is rare and extreme case with prevalence of 1%, hydrocephalus with neurodevelopmental problems.
- Chiari type O is recognized as presence of syringomyelia with no evidence of hindbrain herniation.
- Chairi type 1.5 is described as tonsillar herniation along with elongation of brainstem and 4th ventricle.



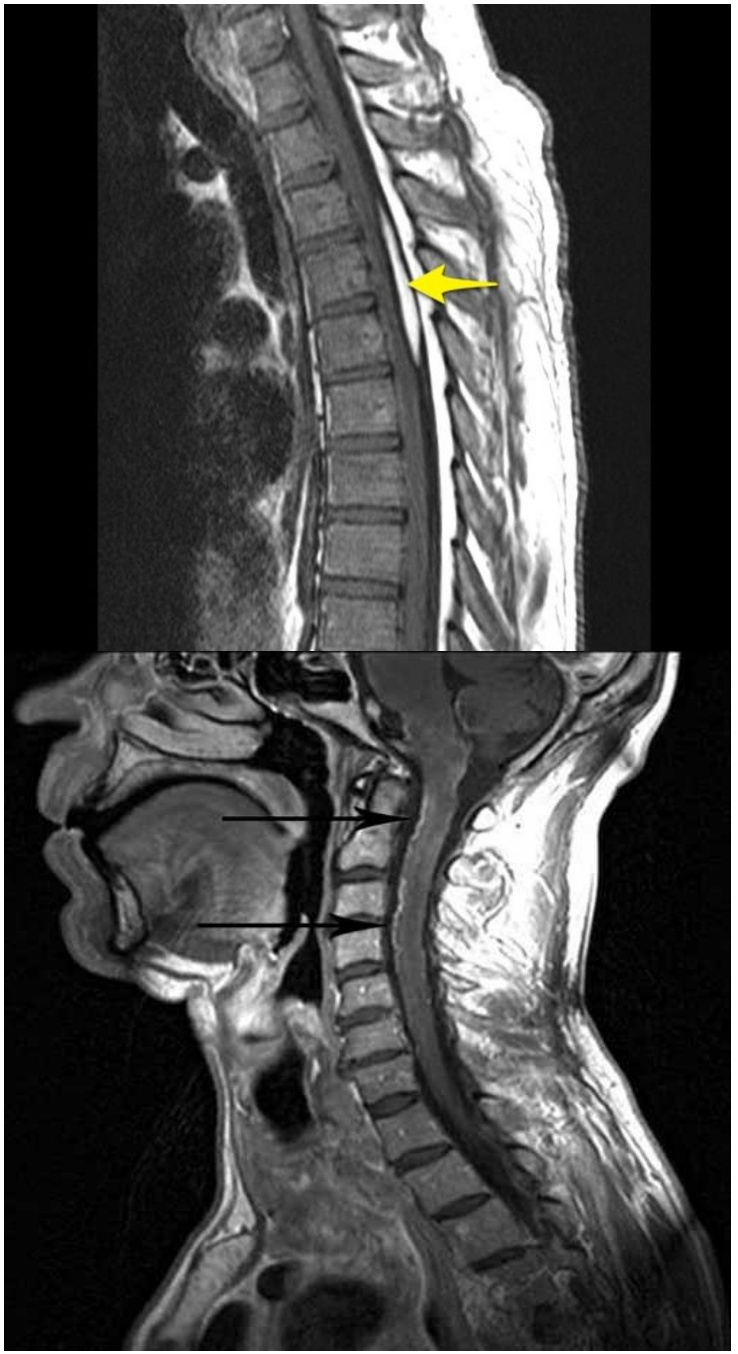


Sign and Symptoms

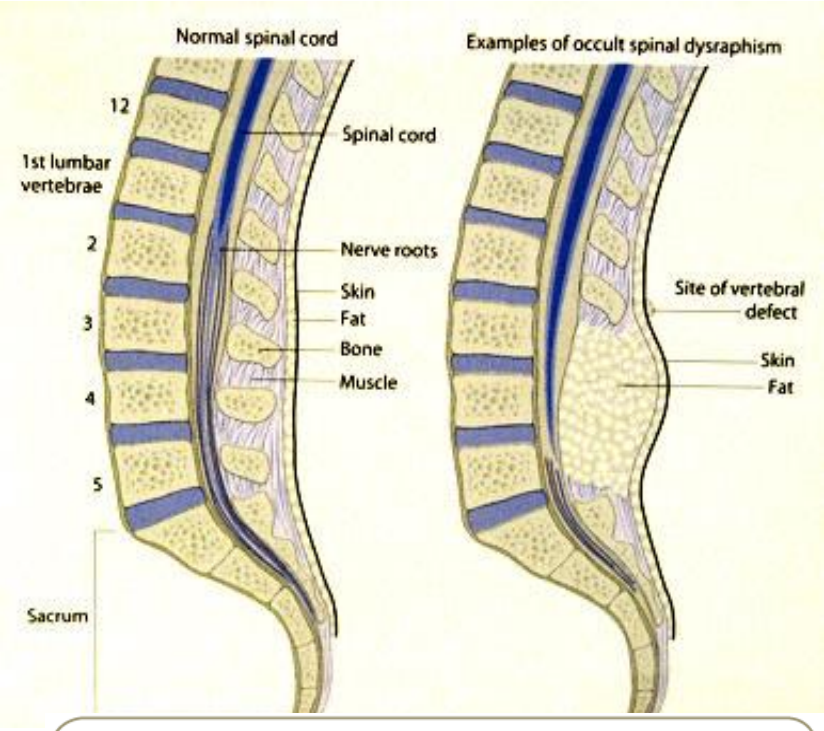
Region	Chiari Malformation I	Chiari Malformation II
Brainstem	Neck pain, headache, hoarse voice, dysphagia, hiccups, severe snoring, facial numbness, palatal dysfunction, dysarthria, respiratory dysrhythmia, tongue atrophy, etc.	Dysphagia, palatal weakness, pneumonia, tongue atrophy, central apnea, poor or weak cry, inspiratory wheezing, nerve IV palsy, facial weakness, decreased or absent gag response, prolonged hiccups.
Spinal Cord	Scoliosis, dissociated sensory loss, wasting of hands or arms, spasticity of legs, urinary incontinence, etc.	Upper extremity spasticity, suspended dissociated sensory loss, huand muscle weakness or wasting, scoliosis.
Cerebellum	Ataxia, nystagmus.	Truncal Ataxia, Nystagmus

SPINAL LIPOMA

- Spinal lipomas are intradural.
- They are typically intradural or juxtamedullary lesions or occasionally intramedullary.
- 84% occur as lipomyelocele/ lipomyelomeningocele, 12% lipoma of filum terminale and 4% intradural lipoma.
- Typically present around second and third decades of life.
- Males and females equally involved.
- Most frequent anomaly seen is numbness or spastic weakness in extremities, with frequent back pains.
- Intradural spinal lipomas consists of normal fat.



- In adults most common site is thoracic region whereas in children it is cervical spine which is most commonly involved.
- Usually occur along the dorsal midline of spinal cord.
- Widening of spinal canal with spreading and thining of pedicles.
- Treatment is obtained by surgical re-sectioning and decompression.



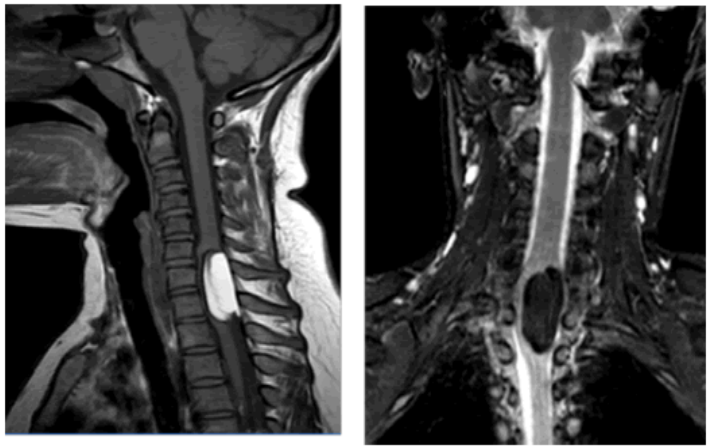
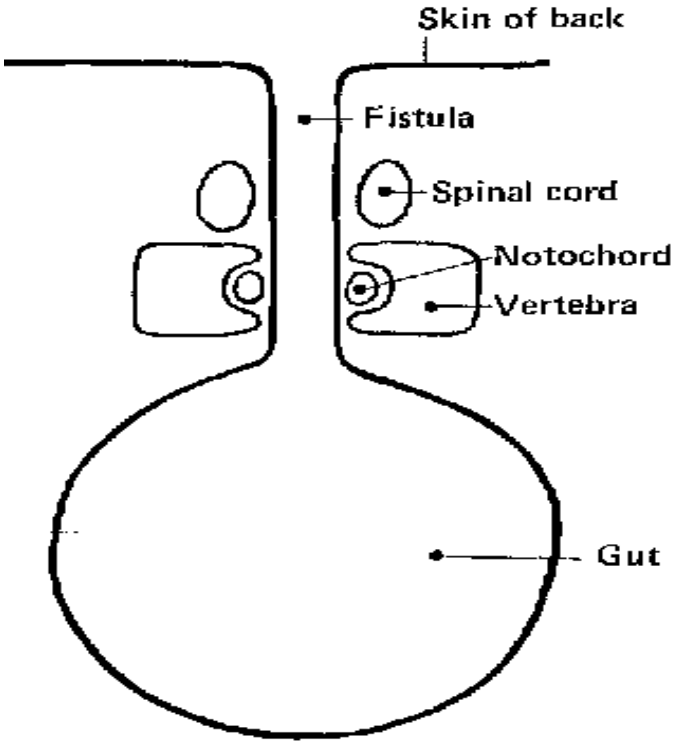
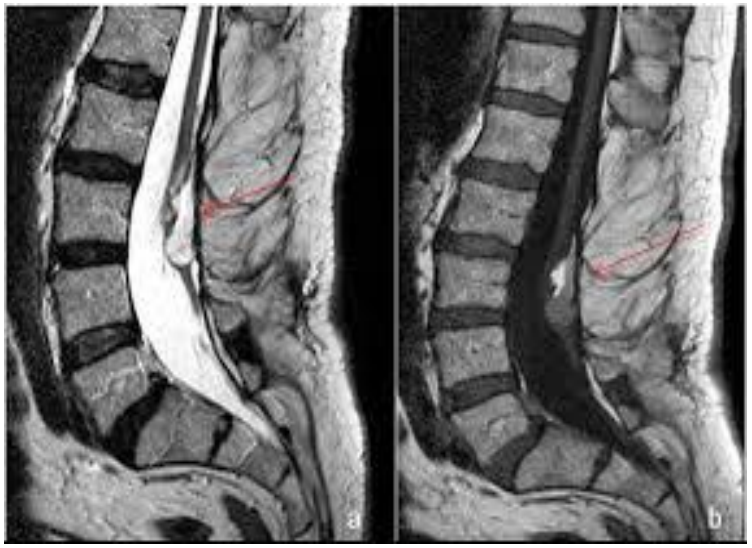


Figure 7A T1 sagittal and T2 coronal fat suppression images of intradural lipoma of cervical cord with cord adherence and intra medullary extension.

SPLIT NOTOCHORD SYNDROME

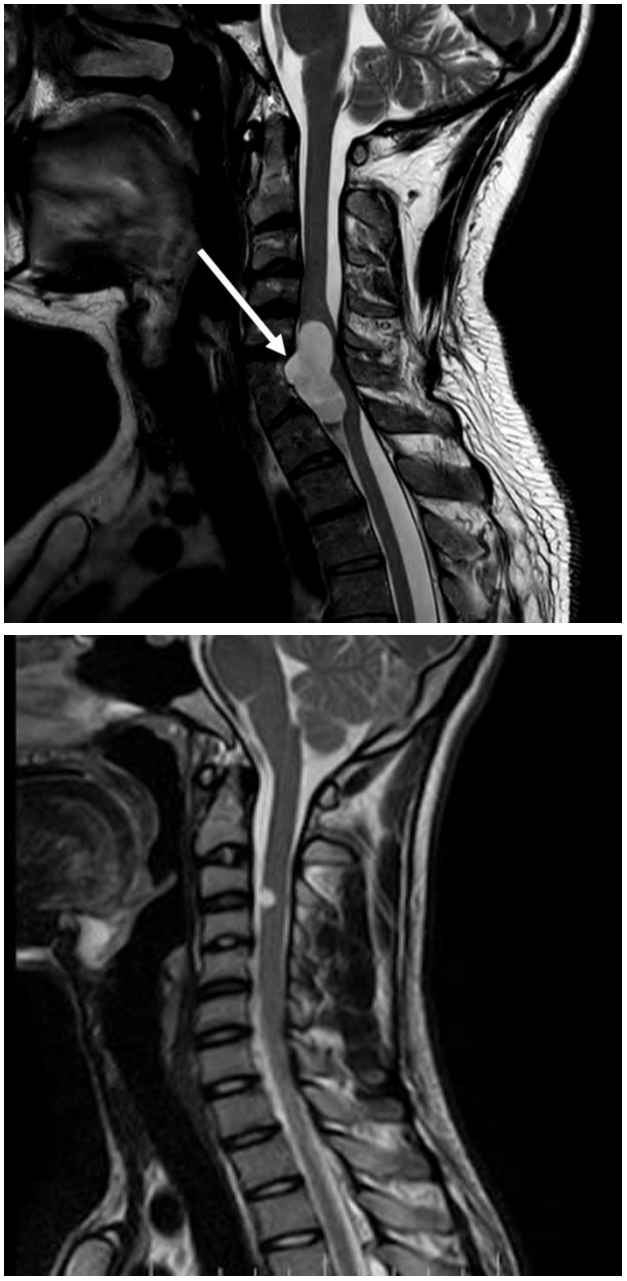
- Extremely rare congenital malformation associated with anomalies of vertebral column, gastro-intestinal tract and central nervous system.
- Persistent communication between the endoderm and ectoderm resulting in splitting or deviation of notochord.
- Cleft in the dorsal midline through which protruding intestinal loops may occur.
- Myelomeningiomas or teratomas may occur at the site.
- Complete midline cleft usually present between the T11 to L5 region.





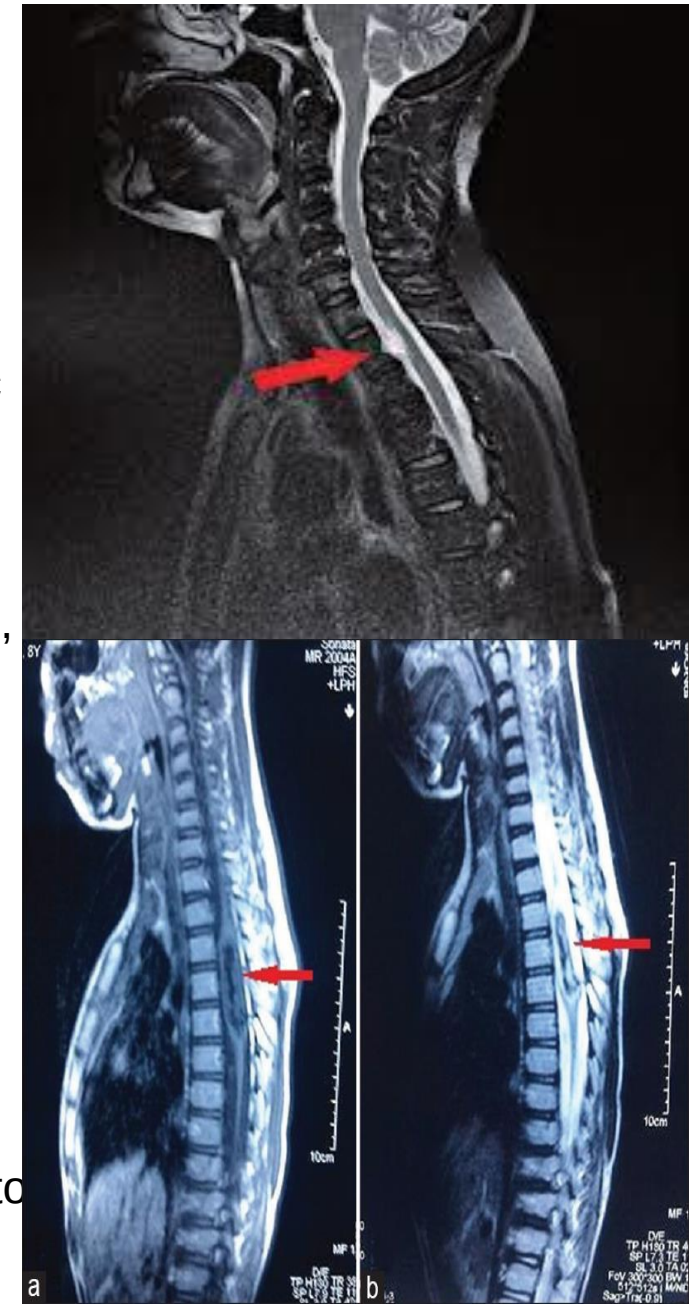
NEUROENTERIC CYSTS

- Rare lesion of spinal axis composed of endodermal tissue.
- Persistence of normally transient neurenteric canal prevents appropriate separation of endoderm and notochord.
- Presence of mucus-secreting epithelium.
- First described by Kubie and Fulton in 1928 and named by Holcomb and Matson in 1954.
- Accounts for 0.7-1.3% of spinal cord tumors.
- 90% ar located in the intramedullary/ extramedullary region.
- Mostly in second and third decade of life.
- Progressive focal pain and fluctuating myelopathy symptoms are the main stay.



Savage JJ, Casey JN, McNeill IT, Sherman JH. Neurenteric cysts of the spine. *J Craniovertebr Junction Spine*. 2010;1(1):58-63. doi:10.4103/0974-8237.65484

- Symptoms often associated with lesions in the cervical and lumbar regions.
- Focal weakness, radicular pain, paresthesia, etc appear later.
- Closely associated with aseptic meningitis, pyogenic meningitis, chronic pyrexia, incontinence and paraplegia.
- 50% of cases are associated with spinal dysraphism, scoliosis, spina bifida, split cord malformation and Kippel Fiel Syndrome.
- Can be easily diagnosed with the help of CT scans and MRI.
- Heterogenous fluid content within the cyst is a rare finding.
- Majority of the neurenteric cysts are located ventral to the spinal cord.



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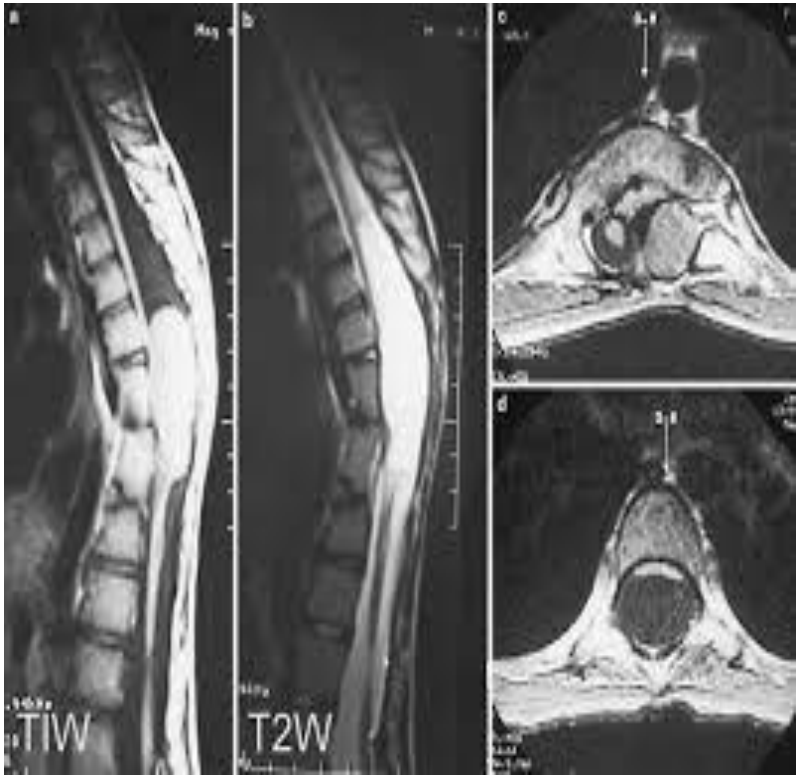
- Surgical resection is the first line of treatment.
- Most often sensory and motor deficits are involved.
- Re-occurence rate of 4% is seen in these cases.
- Compression of spinal cord and associated nerve roots is commonly seen.



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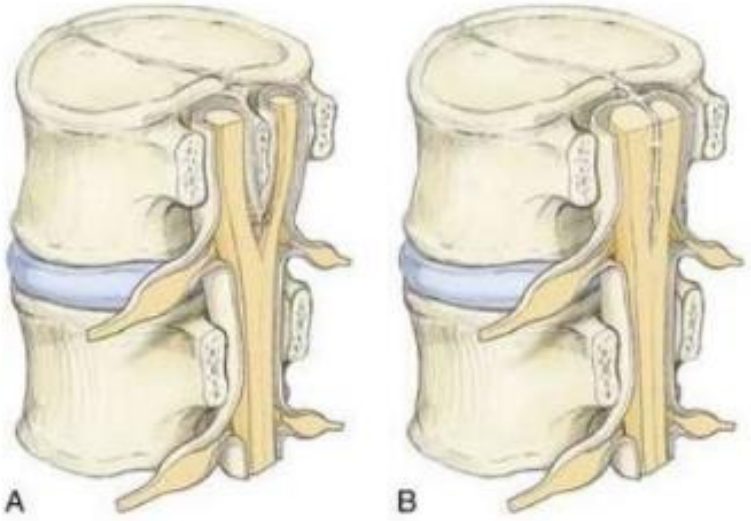
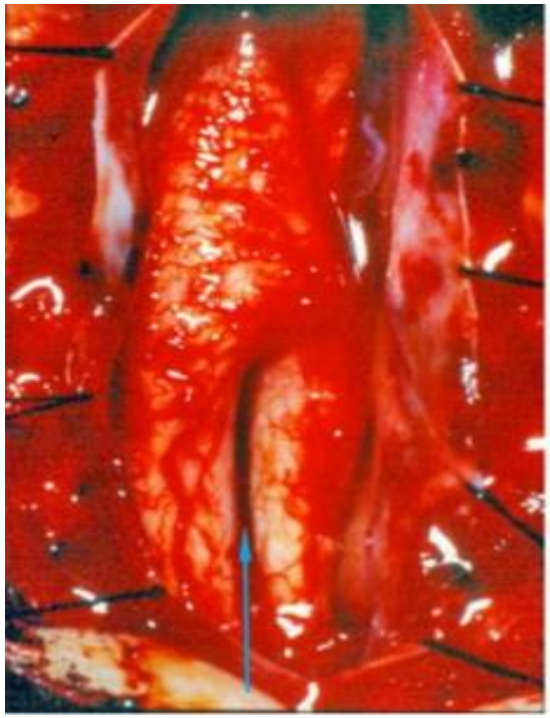
DIASTEMATOMYELIA

- Congenital disorder in which the spinal cord splits usually at the level of upper lumbar region.
- Females are more commonly affected.
- Often termed as a spur and hypertrichosis in association with spina bifida occult.
- Occurs due to presence of osseous, cartilaginous or fibrous septum in the midline of the spinal canal during the 3-4th week of gestation.
- When the split does not unite distally it is termed as diplomyelia.
- Neurological symptoms are non-specific, often related to other spinal cord malformations.
- Course of the disease is progressive.





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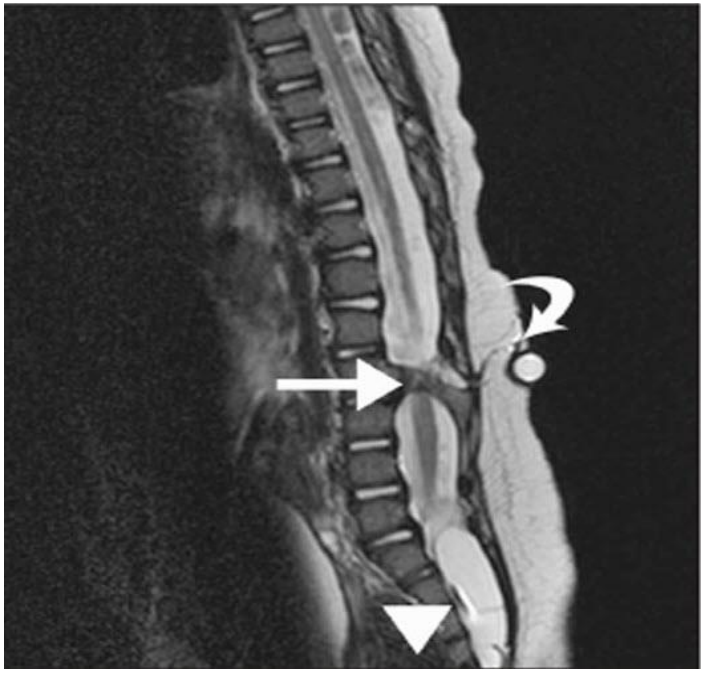
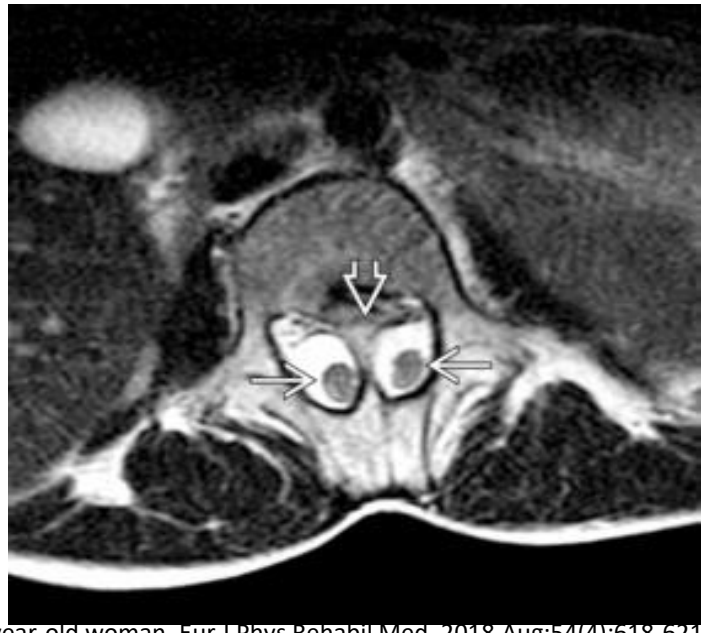
2. Humphreys RP, Hendrick EB, Hoffman HJ. Diastematomyelia. Clin Neurosurg. 1983;30:436-56.
3. Huang SL, He XJ, Wang KZ, Lan BS. Diastematomyelia: a 35-year experience. Spine (Phila Pa 1976). 2013 Mar 15;38(6):E344-9.

- Symptoms appear due to loss of movement at the spinal cord.
- Movements cause abnormal stretching of the spinal canal which leads to tethered cord like symptoms.
- In childhood, symptoms like ankle and foot abnormalities, weakness in legs, incontinence, low back pain and scoliosis are most common.
- In adulthood, progressive sensory and motor abnormalities, loss of bladder and bowel control.
- Delay in presentation of symptom is due to degree of strain placed over the spinal canal.





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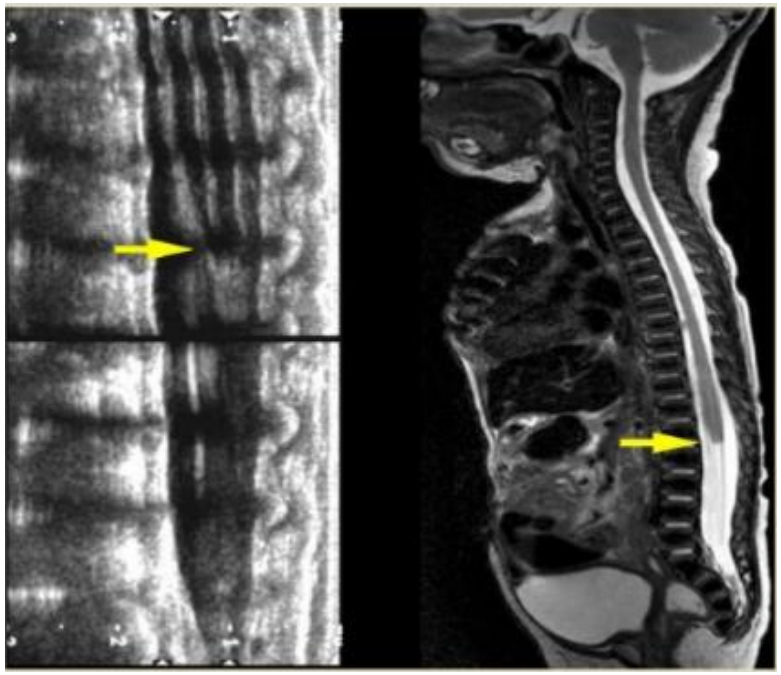
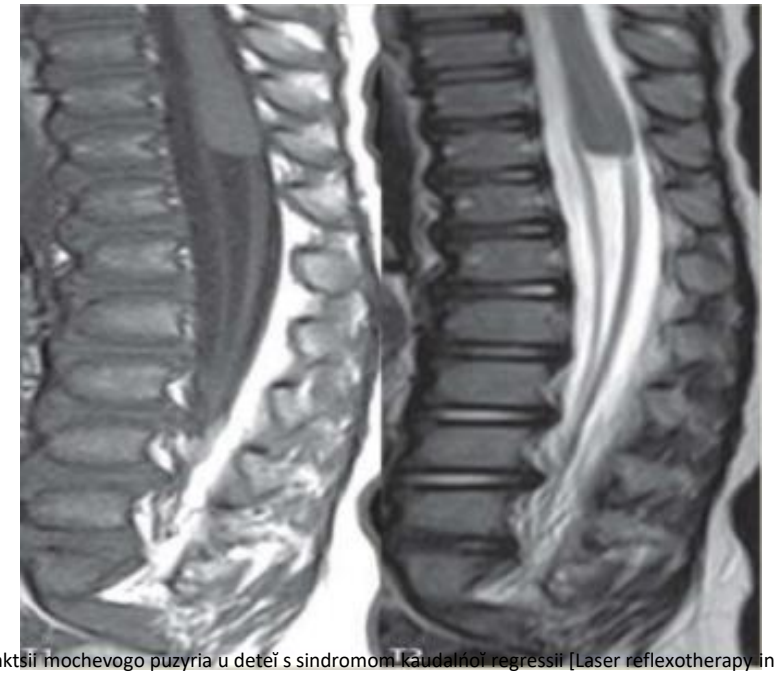
CAUDAL REGRESSION SYNDROME

- Disorder in which there is agenesis of the caudal vertebrae.
- First described by Duhamel in 1961.
- Genetic abnormalities like genitourinal and gastrointestinal problem persist.
- Incidence is 1-5 cases worldwide among every 100,000 childbirths.
- Majorly syndrome is associated with the causal factor maternal diabetes either type I or II.
- Results due to hyperglycemia, infections, toxic and ischemic insults before 4 weeks of pregnancy.
- Due to disturbance of primary neuralation process.
- Primary clinical symptom is neurogenic bladder.





1. Nikolaev SN, Pospelov NV, Prityko AG, Khalpakhchian LKh. Lazerorefleksoterapiia v kompleksnom lechenii neĭrogennoĭ disfunktsii mochevogo puzyria u deteĭ s sindromom kaudalńoĭ regressii [Laser reflexotherapy in the combined treatment of neurogenic bladder dysfunction in children with caudal regression syndrome]. Vestn Ross Akad Med Nauk. 1994;(3):50-1. Russian. PMID: 7516224.
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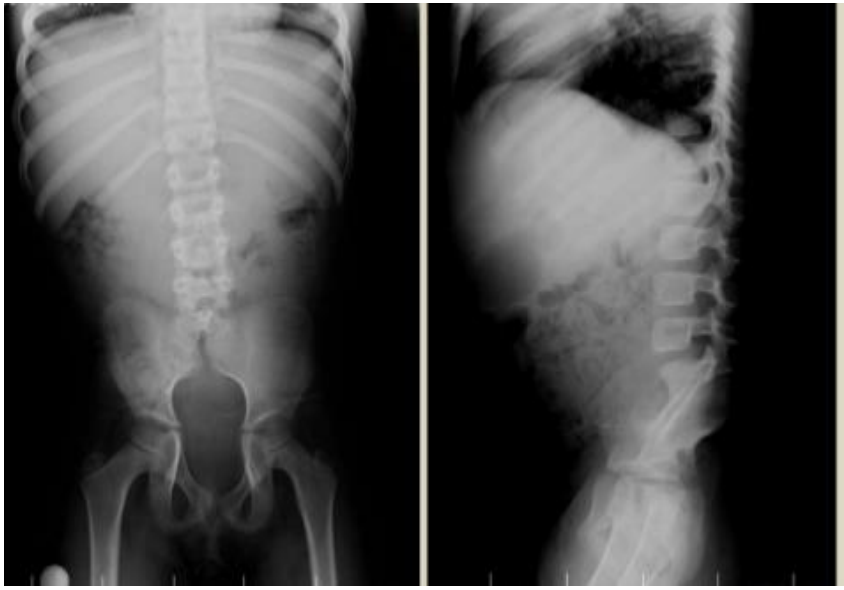
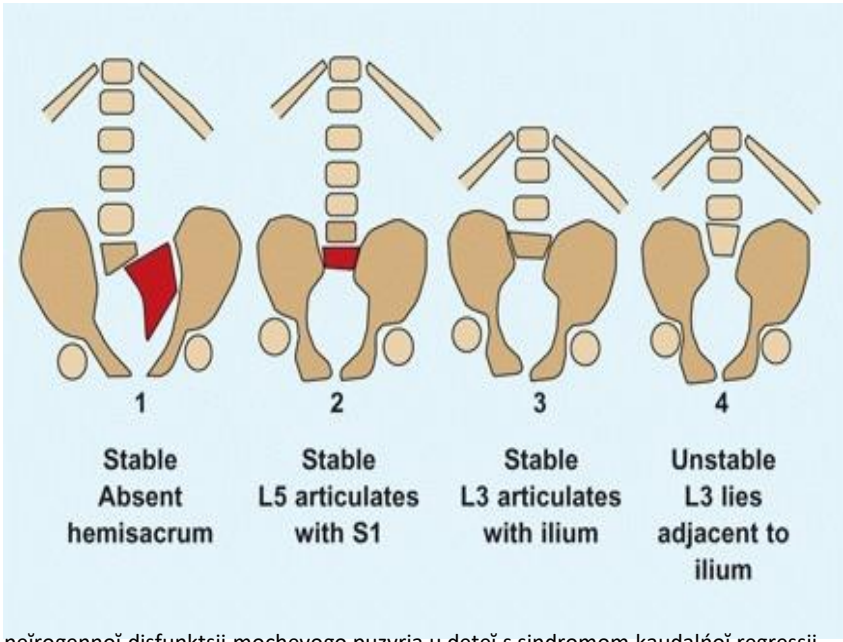
- Also termed as sacral agenesis.
- Due to abnormality of somite formation and disarrangements in the gene Hox.
- Sensorimotor paresis, narrow hips, hypoplastic gluteal muscles, mild foot deformities and gait abnormalities.
- CRS occurs in 1% pregnant women with diabetes.
- Radiological features show cigar shaped conus medullaris.
- Conus medullaris becomes blunt and ends above normal level.
- Absence of lumbosacral elements.
- Iliac bones overlap at midline.
- Ultrasound images show crossed legs images of foetus.





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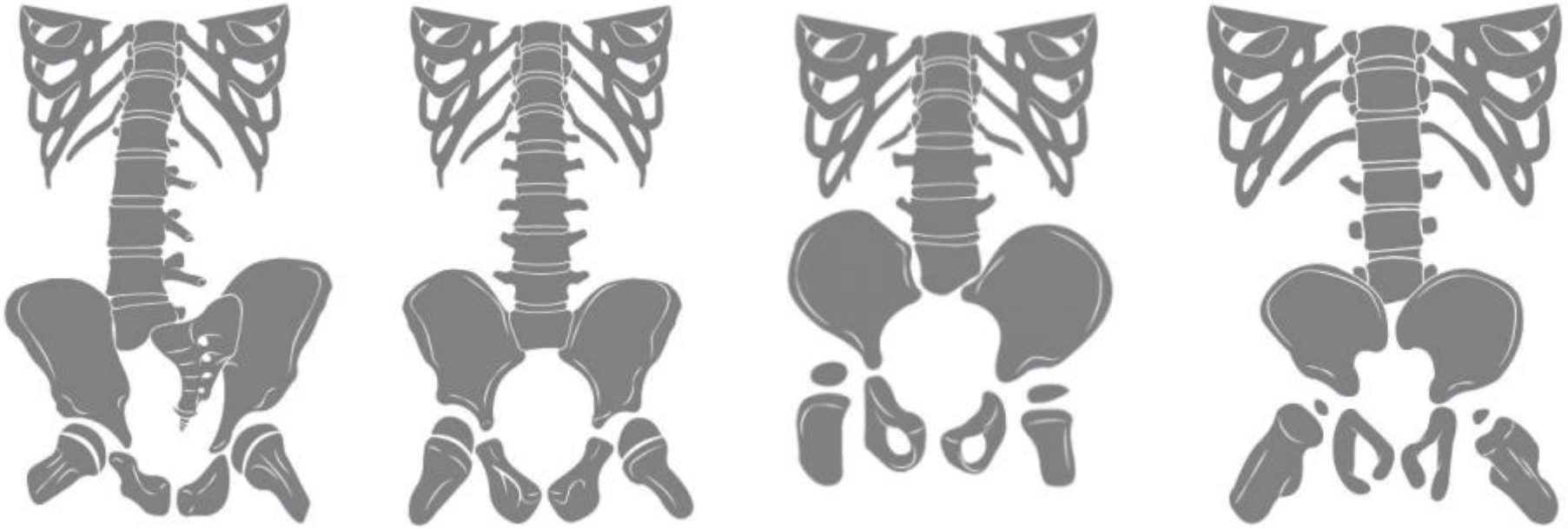

Type I: total or partial unilateral sacral agenesis
Type II: partial sacral agenesis with a partial but bilaterally symmetrical defect and a stable articulation between the ilia and a normal or hypoplastic first sacral vertebra (most common)
Type III: variable lumbar and total sacral agenesis with the ilia articulating with the sides of the lowest vertebra present.
Type IV: variable lumbar and a total sacral agenesis, the caudal end-plate of the lowest vertebra resting above either fused ilia or an iliac amphiarthrosis.
Type V: One set of leg tissue is absent. Aka Sirenomelia or Mermaid's syndrome.

Renshaw's Classifications for SA/CRS

Renshaw T.S. (1994) Sacral Agenesis. The Pediatric Spine – Principles and Practice. 1:2214, Raven Press, New York

- Recent treatment includes Growth Hormone along side rehabilitation.
- GH 0.3mg/day, 5 days/week, for 3 months.
- Low level laser therapy for inducing continence for about 3 months.

1. Nikolaev SN, Pospelov NV, Prityko AG, Khalpakhchian LKh. Lazerorefleksoterapiia v kompleksnom lechenii neĭrogennoĭ disfunktsii mochevogo puzyria u deteĭ s sindromom kaudalńoĭ regressii [Laser reflexotherapy in the combined treatment of neurogenic bladder dysfunction in children with caudal regression syndrome]. Vestn Ross Akad Med Nauk. 1994;(3):50-1. Russian. PMID: 7516224.
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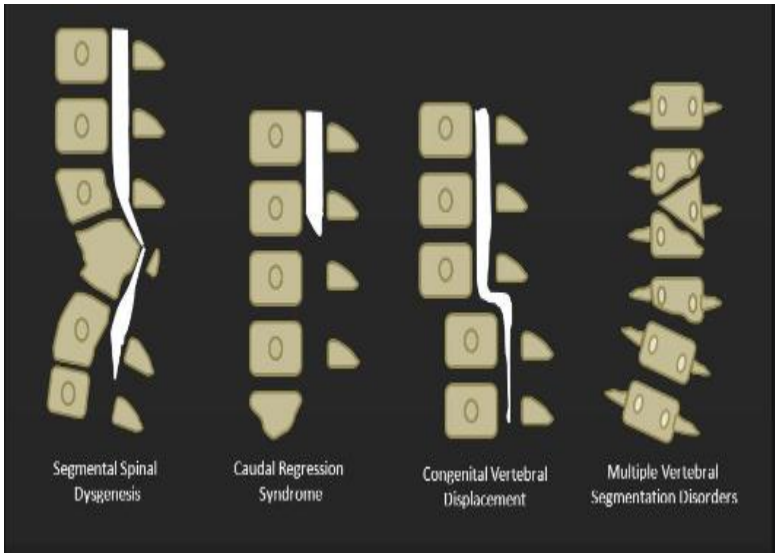
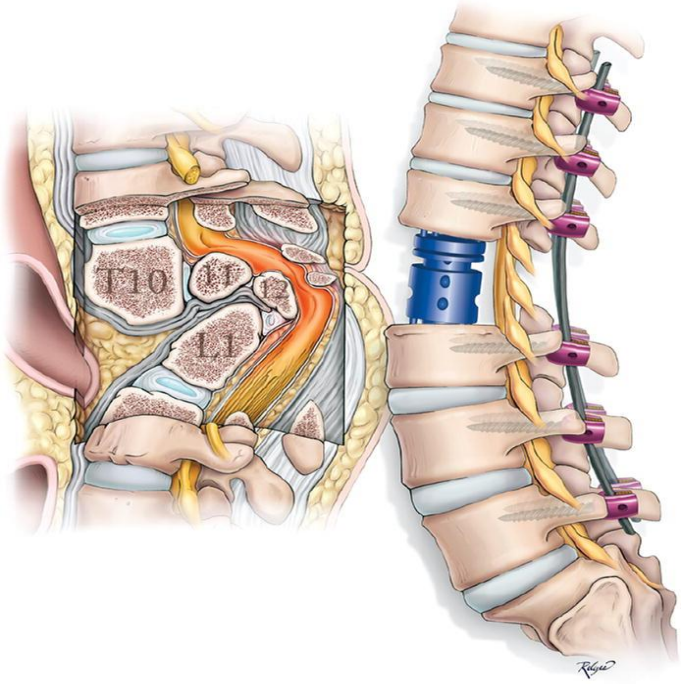
SEGMENTAL SPINAL DYSGENESIS

- It is an umbrella term used for various anomalies like segmental agenesis of thoracic or lumbar spine, segmental abnormality of spinal nerve or nerve roots, congenital paraparesis or paraplegia and congenital lower limb deformities.
- Complete disconnection at the level of dysgenesis.
- Normal upper spinal cord, abnormal affected cord devoid of spinal nerve roots, with bulky thickened distal cords.
- Most of the children present with kyphoscoliosis and motor impairments.
- Reduced tendon reflexes and neurogenic bladder.
- Anorectal anomalies with bilateral dysplastic hips present in nearly 90% cases.
- Type I SSD consists of mild congenital kypho-scoliosis, bulky distal cord without significant spinal canal compromise.





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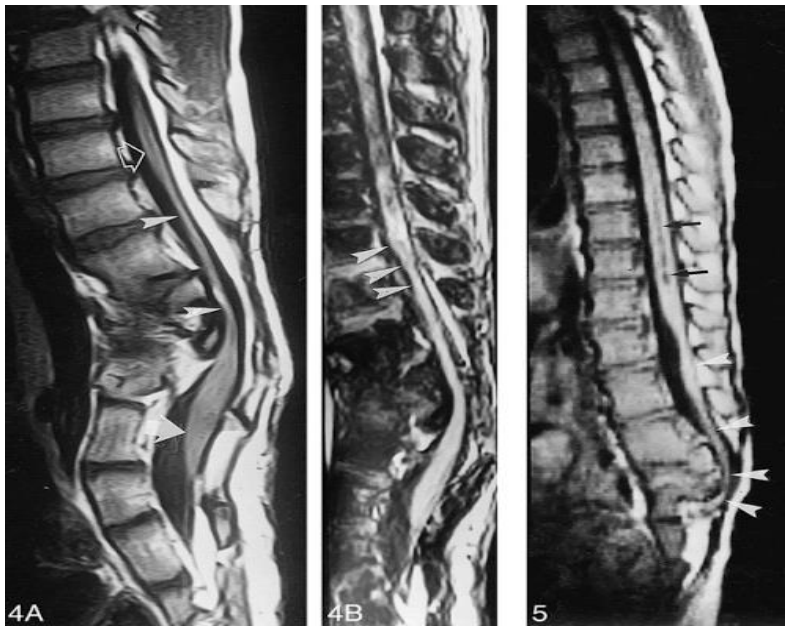
- Type I basically occurs due to loss of embryogenesis of spinal canal.
- Studies show close correlation to Vitamin A deficiency in embryos.
- In type II SSD there is segmental malformation of multiple vertebrate, spinal cord and underlying nerve roots.
- Causing severe kyphoscoliosis, gibbus deformity and severe spinal canal narrowing.
- Mutation in the Notch signalling pathway .
- Congenital vertebral defects are the primarily cause of type II SSD.
- Vertebral anomalies like hemivertebrae, block vertebrae, etc. causing spinal canal compression.
- Paraplegia due to congenital spinal stenosis.



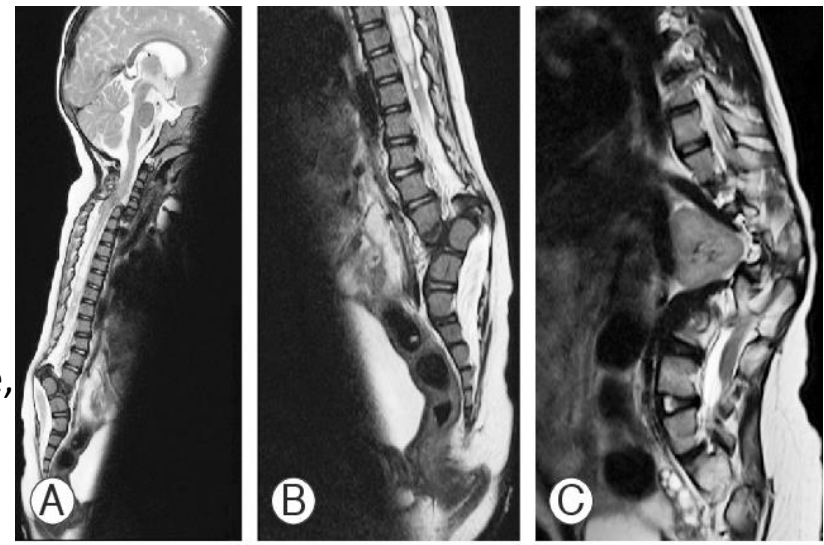


Fig. 5. (A, B) T2 sagittal section of the spine shows bifocal involve-

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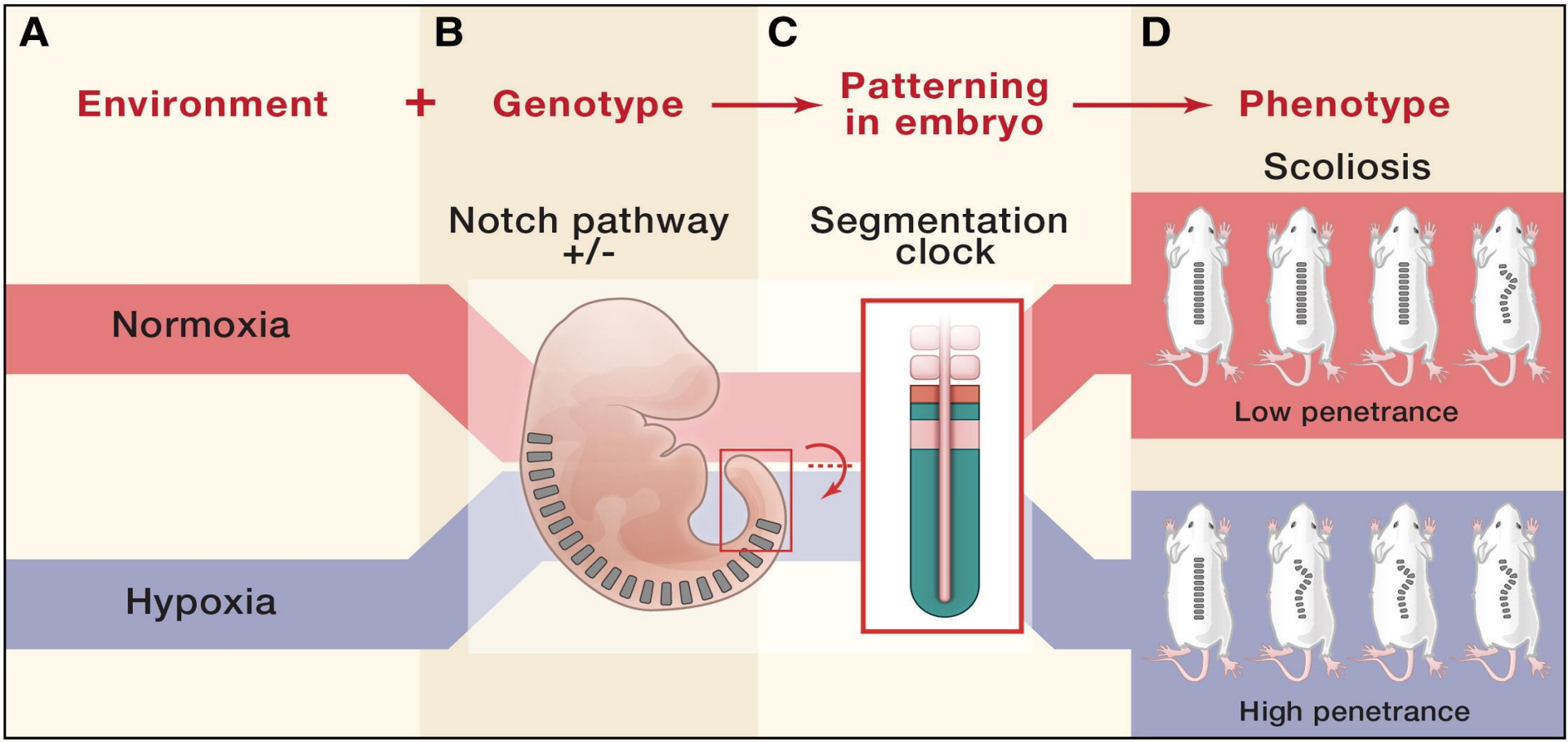
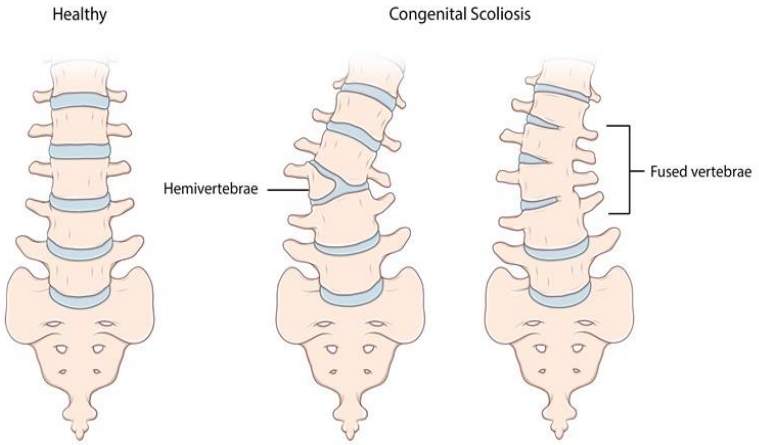
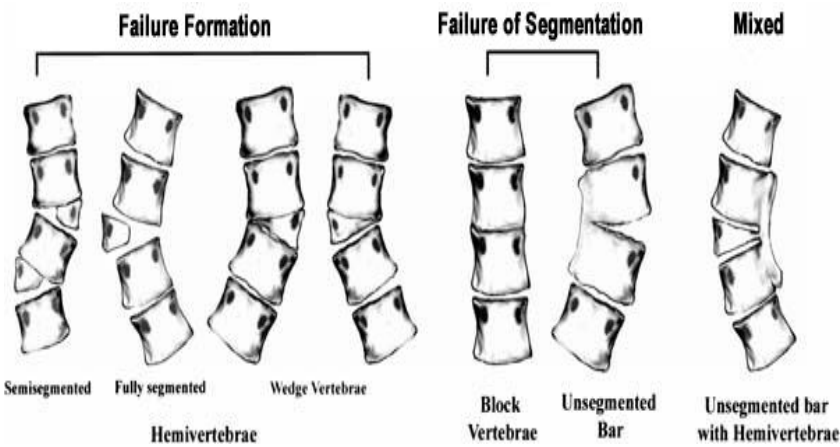
CONGENITAL SCOLIOSIS



- Vertebral anomalies within 5-6th week of gestation cause abnormal growth of spine and deformity.
- No genetic aetiology is said to be associated.
- Defined as lateral curvature beyond 10 degrees, aka, Cobb's angle.
- Primarily caused due to vertebral anomalies like hemivertebra (complete failure of formation of vertebra), wedge vertebra (partial failure of formation), unilateral unsegmented bar (unilateral failure of segmentation) and block vertebra (bilateral failure of segmentation).
- It is most commonly seen at the thoraco-lumbar junction, thoracic region and upper thoracic region in descending pattern.
- The prevalence of disease is 1-4% childbirths worldwide.





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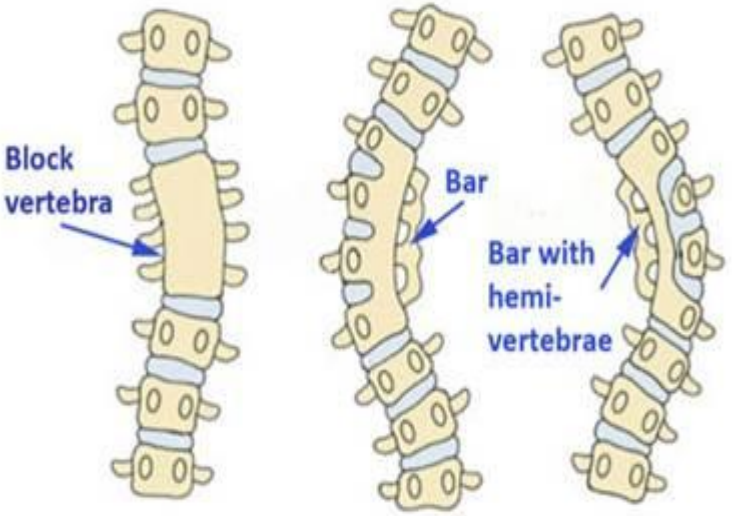
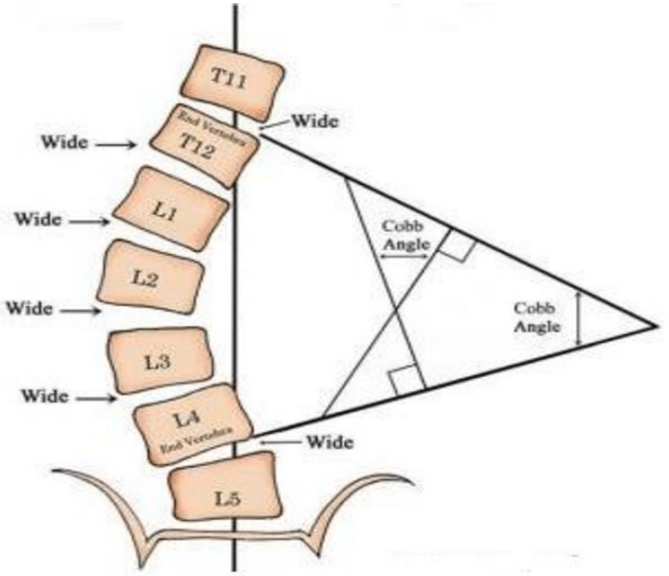
2. Blevins K, Battenberg A, Beck A. Management of Scoliosis. Adv Pediatr. 2018 Aug;65(1):249-266.

- Congenital scoliosis children usually get detected before 3 years of age.
- 26% patients deal with cardiac anomalies, 21% deal with genitourinal anomalies.
- Major treatment strategies include casting, physical therapy interventions and in extreme cases surgical interventions.
- Often associated with Chiari Malformation, tethered cord syndrome, lipoma, diastematomyelia, etc.
- Surgery is indicated in rapid progressive congenital scoliosis only.
- Bracing is not recommended in congenital scoliosis.





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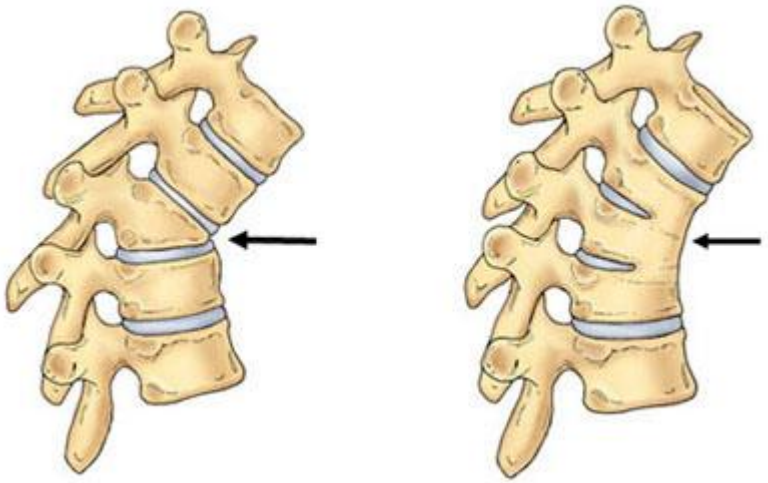
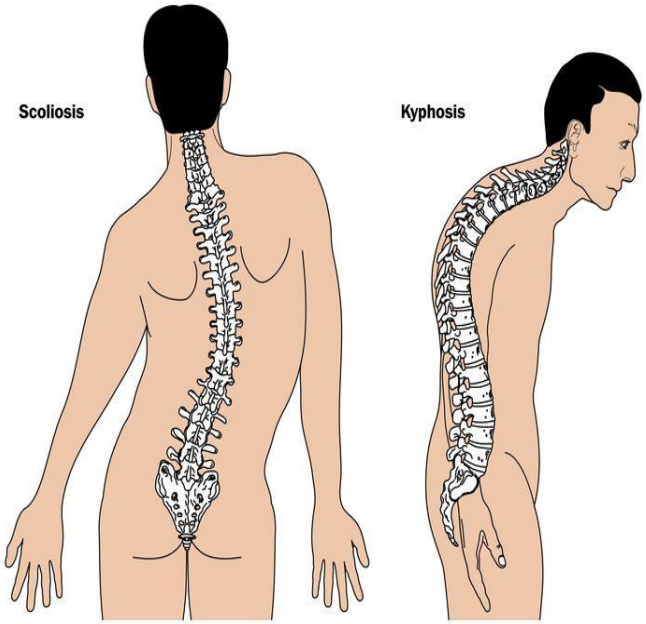
CONGENITAL KYPHOSIS

- In normal individual, cervical spine is lordotic, thoracic spine is 20-50 degree kyphotic, lumbar is again lordotic and sacral kyphotic. Kyphosis of greater than 50 degree is considered abnormal.
- Abnormal curving of spine causing slouched posture is termed as kyphotic posture.
- Vertebrae may be malformed or fused together.
- It is quite uncommon with frequent defects.
- In this the spinal deformity increases with growth.
- There are basic three types of congenital kyphosis.
- Type I is congenital failure of vertebrae formation.





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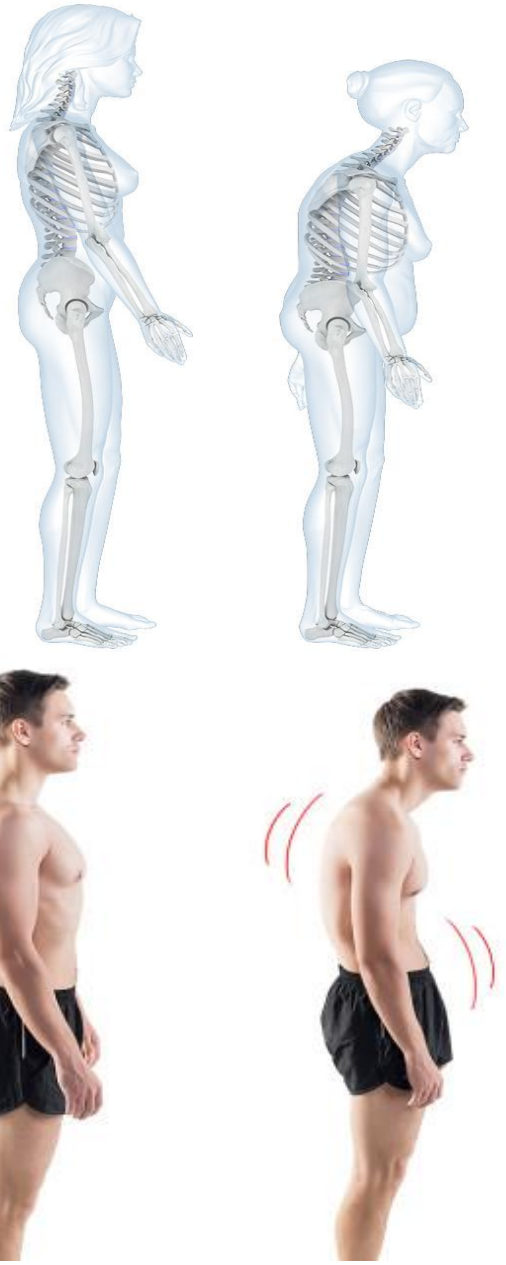
- Type II is failure of vertebral body segmentation.
- Type III is combination of failure of formation and segmentation of vertebral body.
- Most commonly occurs at the thoracolumbar and lower thoracic region.
- Bracing has no role in congenital kyphosis.
- Surgical interventions like posterior fusion, anterior fusion, or anterior osteotomy with posterior fusion could be opted for better results.
- Progression of kyphosis may lead to paralysis.
- Larger deformities may lead to obstruction to movement and pain.



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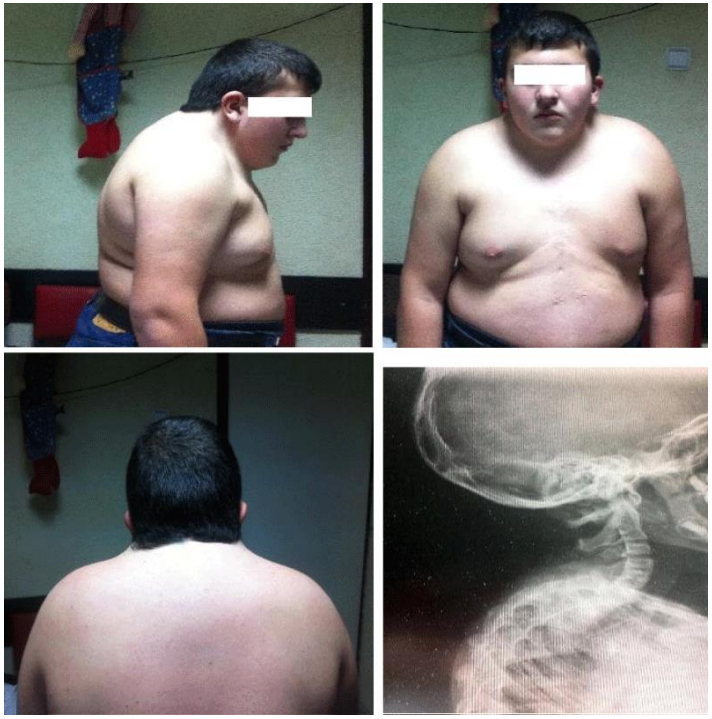
KLIPPEL- FEIL SYNDROME

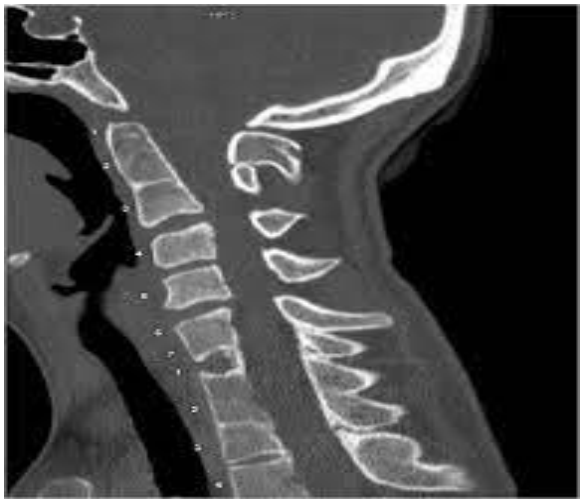
- Congenital defect in the cervical vertebrae which includes fusion of two or more vertebrae.
- There are basic three types:

  Type I: fusion of whole cervical spine.

  Type II: fusion of 1 or 2 cervical spine.

  Type III: Type I or II with thoracic and lumbar spine anomalies.
- Also termed as congenital brevicollis syndrome.
- It is an autosomal dominant disorder.
- Distinctive features includes short statured neck, limited range of motion which is very low than normal ranges and low hairline at the neck.
- Neural compression occurs at the spinal foramina which creates pain (radiating type) at the fused vertebra level.
- As age and disease progresses it leads to spinal stenosis.





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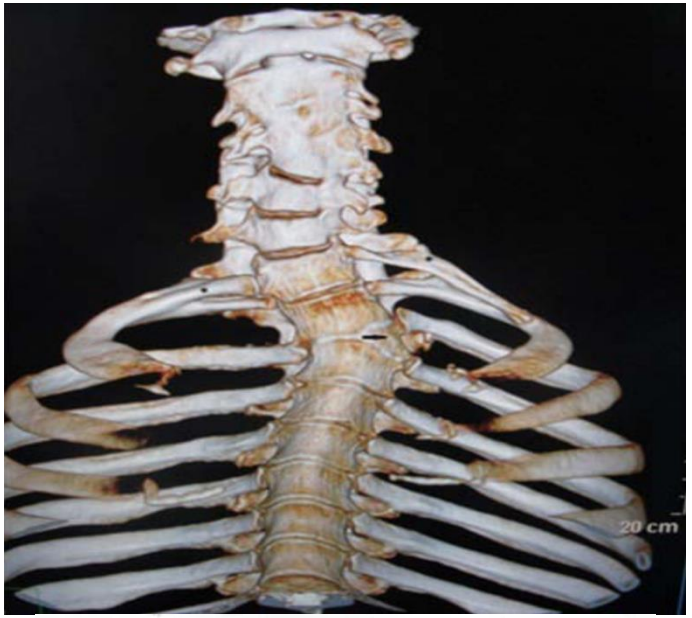
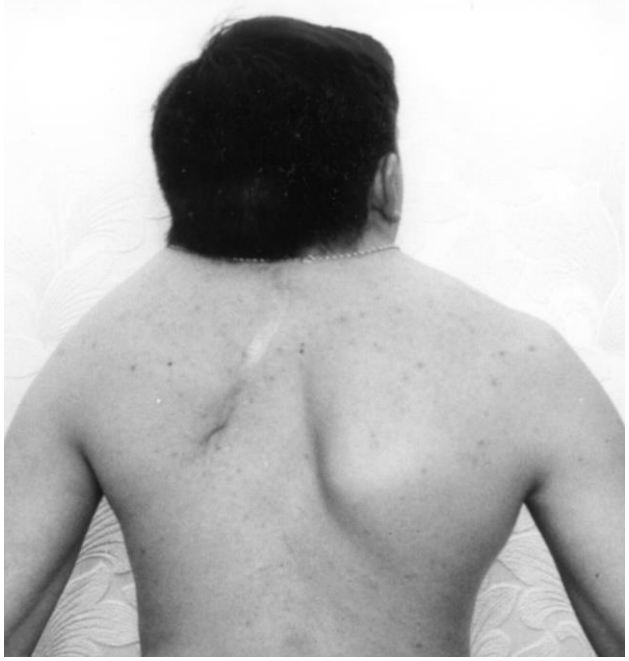
- Vertebral artery is a common anomaly in patients.
- Caused due to genetic mutation of GDF6 and GDF3 genes.
- These genes are responsible for bone formation and bone development.
- Often related to spina bifida and scoliosis.
- Often delay in treatment leads to development of radiculopathy and myelopathy.
- Physiotherapy along with NSAIDS and Opiods are the key treatment procedures.
- Surgical interventions are not frequent with the disease.
- Major symptoms include paralysis, ataxia, hearing problems, vertigo, and in severe cases spasticity.





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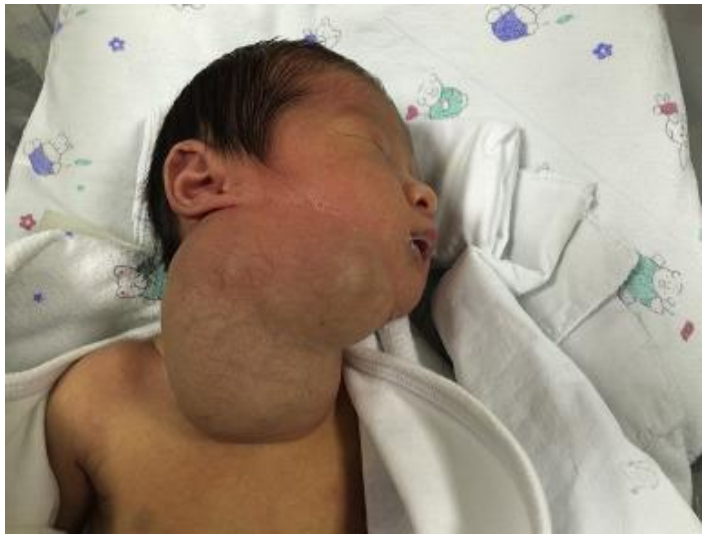
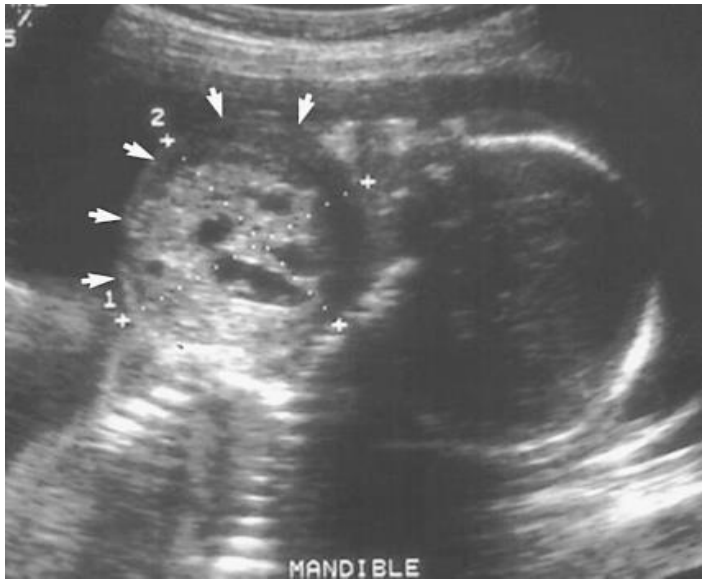
TERATOMA

- Teratomas are group of cells having teeth, hair, nails and cells like a foetus.
- Due to abnormal differentiation of fetal germ cells.
- Sacroccygeal teratomas form 40% of teratomas and other common sites are cervical spinal canal, mediastenum, brain, etc.
- Women more commonly suffer from benign and mature teratomas whereas males commonly suffer from immature and malignant teratomas.
- Various types of teratomas like spinal dermoid cysts are present most commonly in the lumbo sacral region and cause neural tube closure.
- Diagnosis is made through ultrasound and MRI scans prenatal period.





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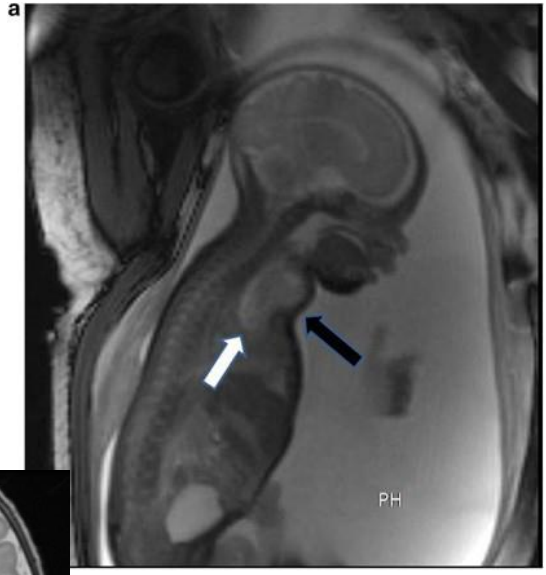
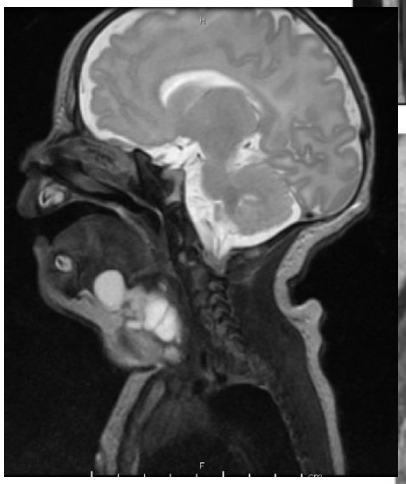
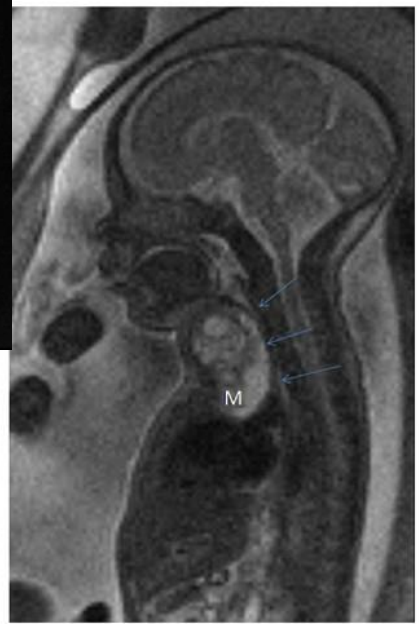
- Often correlated with steal syndrome in which the fetal blood supply is diverted towards teratoma.
- Prevalence is 1 in 35,000-40,000 cases.
- 30-50% cases are diagnosed prenatally.
- Fetal sacroccygeal teratoma are the most common congenital germ cell tumor.
- Cervical teratomas lead to hyper extension and flexion of neck of child towards one side.
- Cervical teratomas cause neonatal airway obstruction.
- Surgical and chemotherapeutic interventions are the mainstay treatments available.







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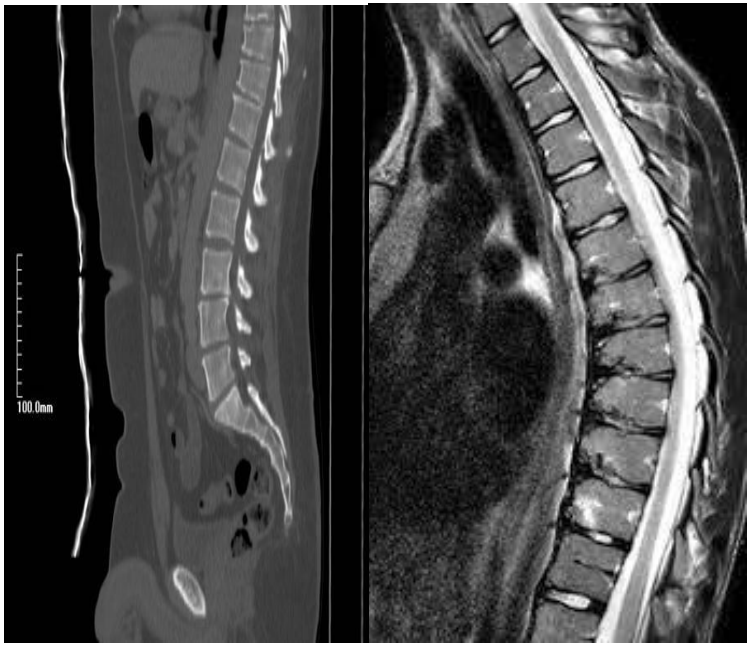
SCHEUREMANN'S DISEASE

- Also known as juvenile kyphosis.
- Due to deformed thoracic or thoracolumbar region.
- Increase in the kyphosis leads to various anomalies.
- Stooped posture is seen due to wedge shaped vertebra.
- Commonly diagnosed between 12-14 years of age.
- Wedge shape and deformed posture causes the anterior longitudinal ligament to get thickened at the site.
- Site of deformed vertebra forms the schmorl's nodes.
- Varied sign and symptoms occur due to compression of the spinal cord at the level.
- Basic signs include muscle cramps, chest pains, formation of hump while bending.
- In rare circumstances, decreased lung capacities and breathing difficulties persist.



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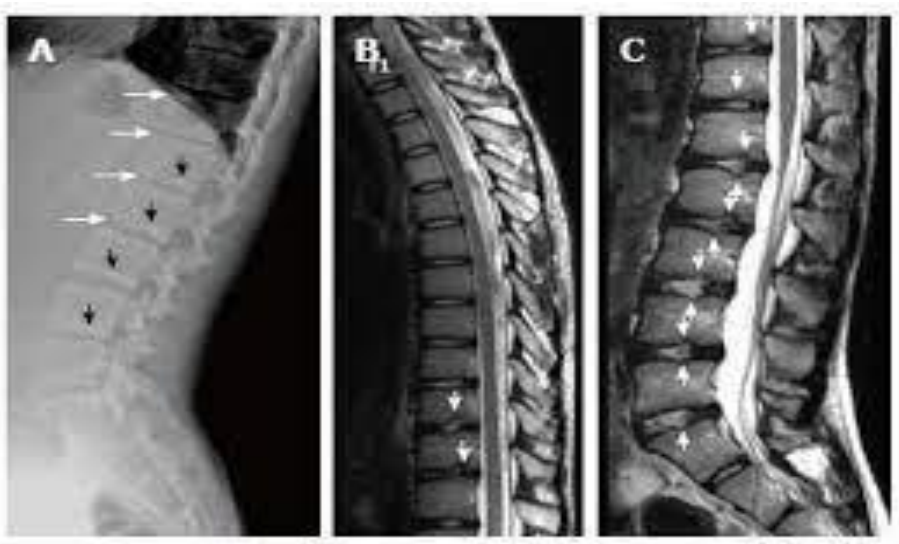
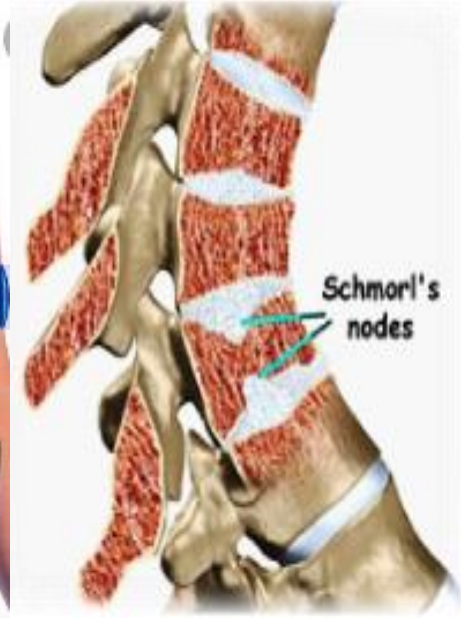
- Major complications include chronic low back pain, progressive deformity, neurological deficit, cardio-respiratory deficits.
- Compression occurs at the initial starting level of kyphosis.
- Diagnosis can be made on the basis of X-rays and posture analysis.
- Management depends on the degree of kyphosis.
- Surgically posterior spinal fusion can be done to reduce the curvature.
- For pain management, moist heat packs, cryotherapy, short wave diathermy, etc. can be used.
- Physiotherapy along with bracing is recommended.





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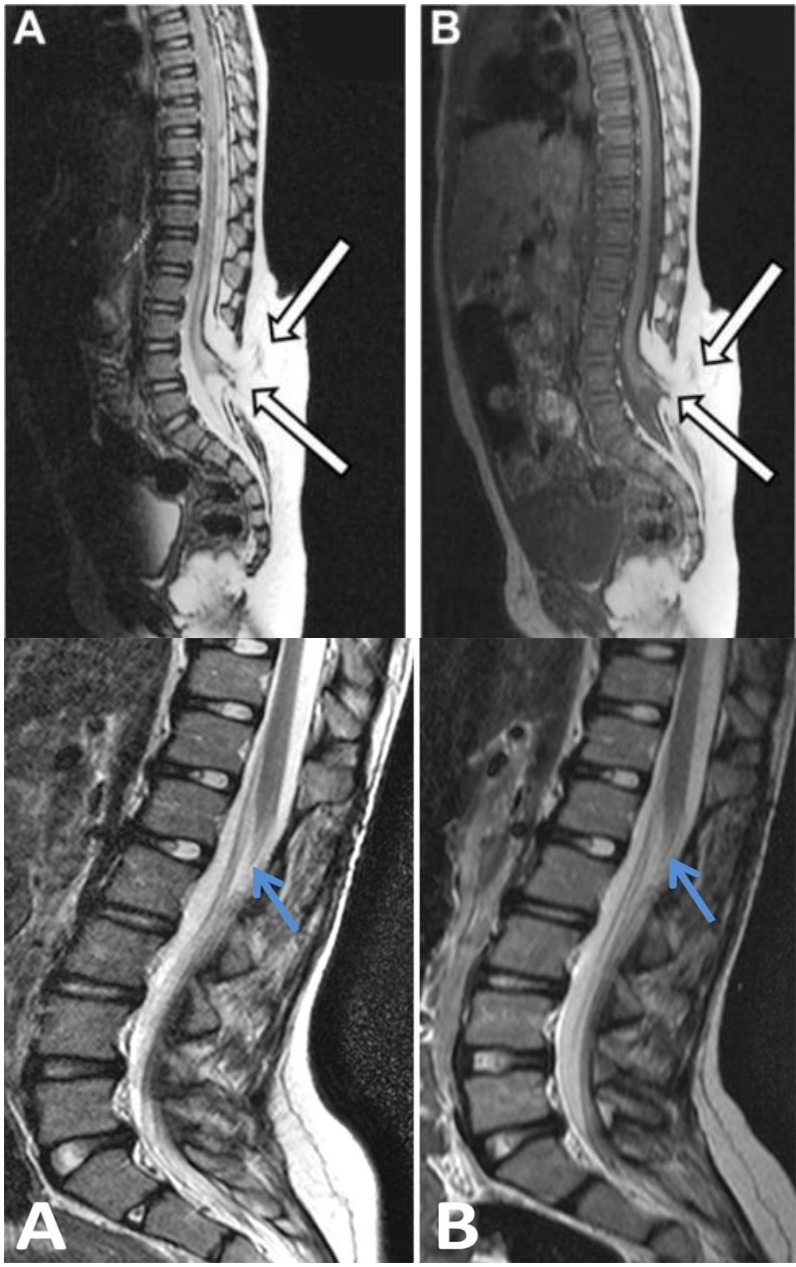
TETHERED CORD SYNDROME

- Group of sign and symptoms of motor and sensory abnormalities as a result of tension in the spinal cord and spinal canal.
- Abnormally low position of conus medullaris.
- As the traction and stress over the spinal cord increases, blood flow and oxidative metabolism gets hampered.
- Causes may be varied like spina bifida, split cord formation, lumbosacral lipomas, neuroenteric cysts, myelomeningocoele, etc.
- In neonates, urinary dribbing may be the first sign apart from radiological features.
- Scoliosis and lower extremity deformities are common.
- Children usually present with progressive motor dysfunction, sensory anomalies, scoiliosis, foot deformities, decreased control over bladder and bowel, etc.



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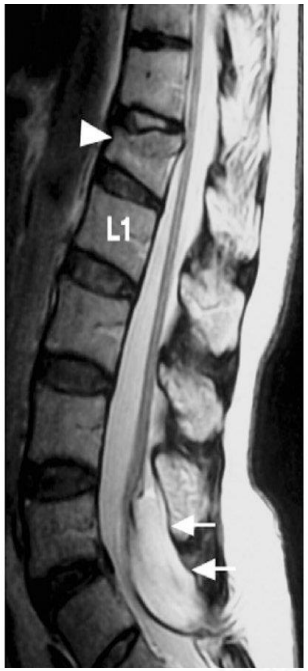
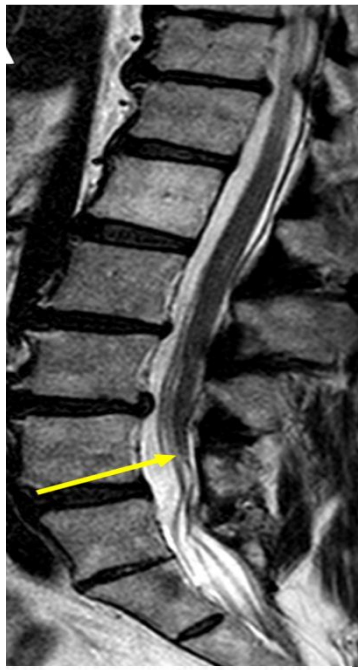
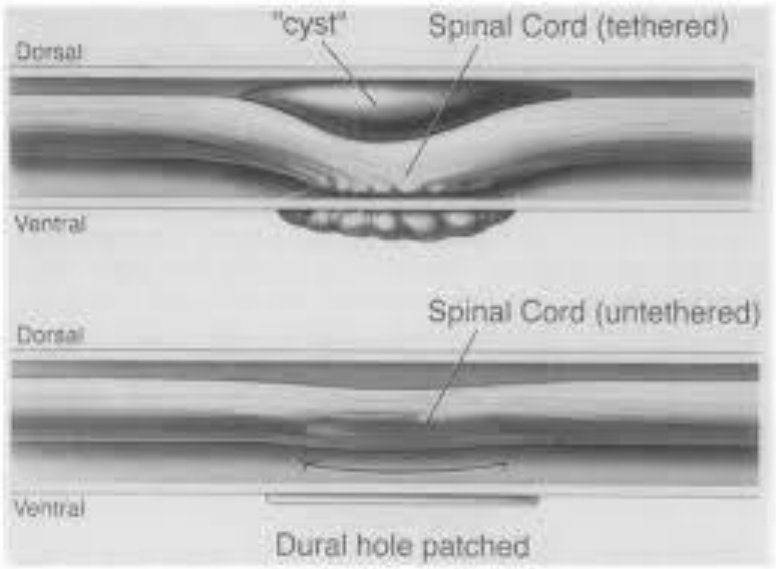
2. Hertzler DA 2nd, DePowell JJ, Stevenson CB, Mangano FT. Tethered cord syndrome: a review of the literature from embryology to adult presentation. Neurosurg Focus. 2010 Jul;29(1):E1.

- Pain is somewhat restricted to the low back pain.
- Limb length discrepancies and gait abnormalities are frequent in children.
- Spasticity and clonus appear.
- Most commonly detrusor hyper reflexia is present.
- Symptoms are present only in patients with worsening conditions, there may be condition where tethered cord is present without symptoms.
- Surgical intervention includes untethering of spinal canal.
- Physiotherapeutic intervention is rendered symptomatically.
- Complications which may arise post surgical intervention include infection, bleeding, damage to spinal cord, loss of bladder and bowel control, etc.







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THANK YOU