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A Case Of Spina Bifida Occulta With Tethered Cord Syndrome

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Clinical History

A 10-month-old baby presented with complaints of a lesion on the lower back since birth. The lesion was initially pinkish in color, present in the midline, 1×1.1 cm, irregular in shape, flat, and gradually changed color (presently brown). Puckering was present over the lesion.

No h/o increase in size or depth of the lesion/seizures/bowel or bladder dysfunction/increase in the size of the head.

It was initially diagnosed as a mole at a local medical facility and medications for local applications were prescribed. Recently, a discrepancy in lower limb length was noticed (left less than right) and Spina bifida occulta with a tethered cord was suspected and was admitted now for radiological investigations and further management.

PAST HISTORY: H/o fever + 20 days before admission. No h/o prior hospitalization, No h/o seizure activity or involuntary movements

ANTENATAL HISTORY: Mother did not take Folic Acid supplements during the first trimester of pregnancy. No h/o GDM/Gestational Hypertension/Thyroid disorder in the mother.

NATAL HISTORY: Baby was born by Full Term LSCS due to the non-progression of labour. Birth weight – 3.1 kg. The baby cried immediately after birth. Feeds were initiated within 2 hours of delivery and there was no difficulty noted. No NICU admissions.

POSTNATAL HISTORY: Baby passed urine and meconium within 24 hrs. Uneventful postnatal period.

DEVELOPMENTAL HISTORY: Baby has attained all milestones according to age. No developmental delay was noted.

IMMUNISATION HISTORY: Immunisation is completed as per schedule. BCG, DPT-1, OPV-0, HiB, HepB has been given.

NUTRITION HISTORY: The baby was exclusively breastfed for 6 months and weaning started with Ragi along with breastfeeds (4-5 times/day)

The baby's diet has mild deficiency of calories and proteins.

FAMILY HISTORY: Secondborn of two children of a non-consanguinous marriage. No similar history in the family. No h/o congenital anomalies.

The older child is an 8-year-old boy, alive and healthy.

Examination

A 10-month-old child is conscious, active, and alert.

VITAL SIGNS:

PR – 124 bpm

RR – 24 CPM

Temp.- 97.4°F

SpO2- 96-97% at room air

CFT- <3 sec

ANTHROPOMETRY

	PRESENT	EXPECTED	PERCENTILE (WHO)
WEIGHT	7.7 kg	9 kg	3rd-15th centile
HEAD CIRCUMFERENCE	45 cm	44 cm	50th-85th centile
LENGTH	72 cm	75 cm	15th-50th centile

HEAD TO TOE EXAMINATION

Normal.

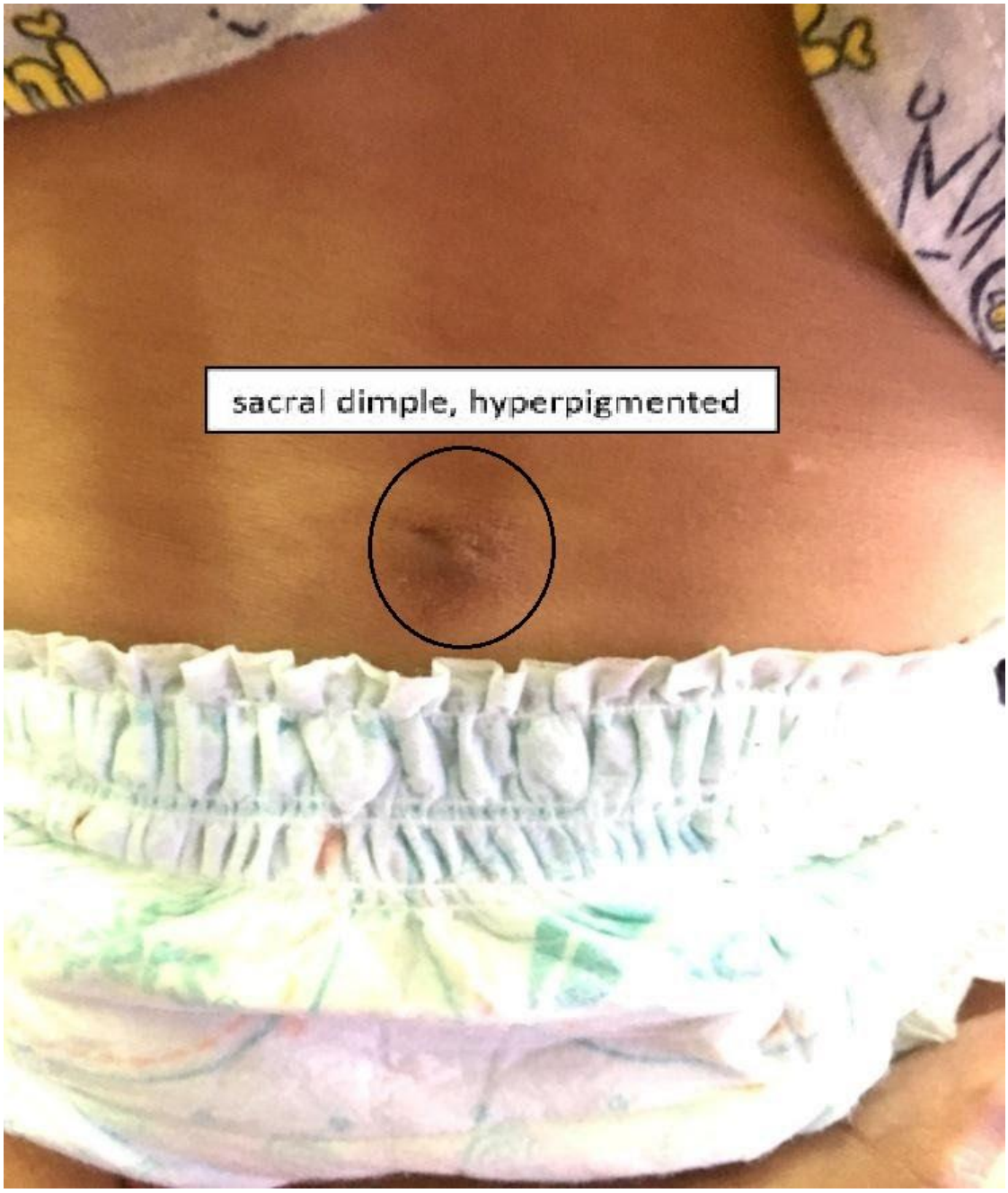
No icterus, no cyanosis, no edema, no lymphadenopathy

SYSTEMIC EXAMINATION

On CNS examination, the child was alert, conscious, and active.

Neurological observation was significant for SACRAL DIMPLE, BROWNISH COLOUR, 1×1.2 cm, IRREGULAR IN SHAPE, WITH DERMAL SINUS AND TUFT OF HAIR.

Limb discrepancy + (left lower limb shorter than the right lower limb)



On palpation, a skeletal defect was noted along the region of the sacral spine.

Cranial Nerves – B/L pupils reacting equally to light. No facial asymmetry.

Motor system- Tone normal in all 4 limbs. Power 5/5 in all 4 limbs.

The bulk of right thigh (22 cm) > Left thigh (21 cm)

Length of left lower limb (31 cm) < right lower limb (32 cm)

Sensory system- could not be assessed

No signs of meningeal irritation

Musculoskeletal system normal

CVS- S1, S2 heard. No murmurs.

RS- Normal Vesicular breath sounds heard. No added sounds.

PA- Soft, non-tender.

Investigations

Hb- 8.8 g/dL (normal 11-14 g/dL)

TLC – 12,470 cells/cumm

RBC – 3.75 million/cumm

Platelets – 5.19 lakh/cumm

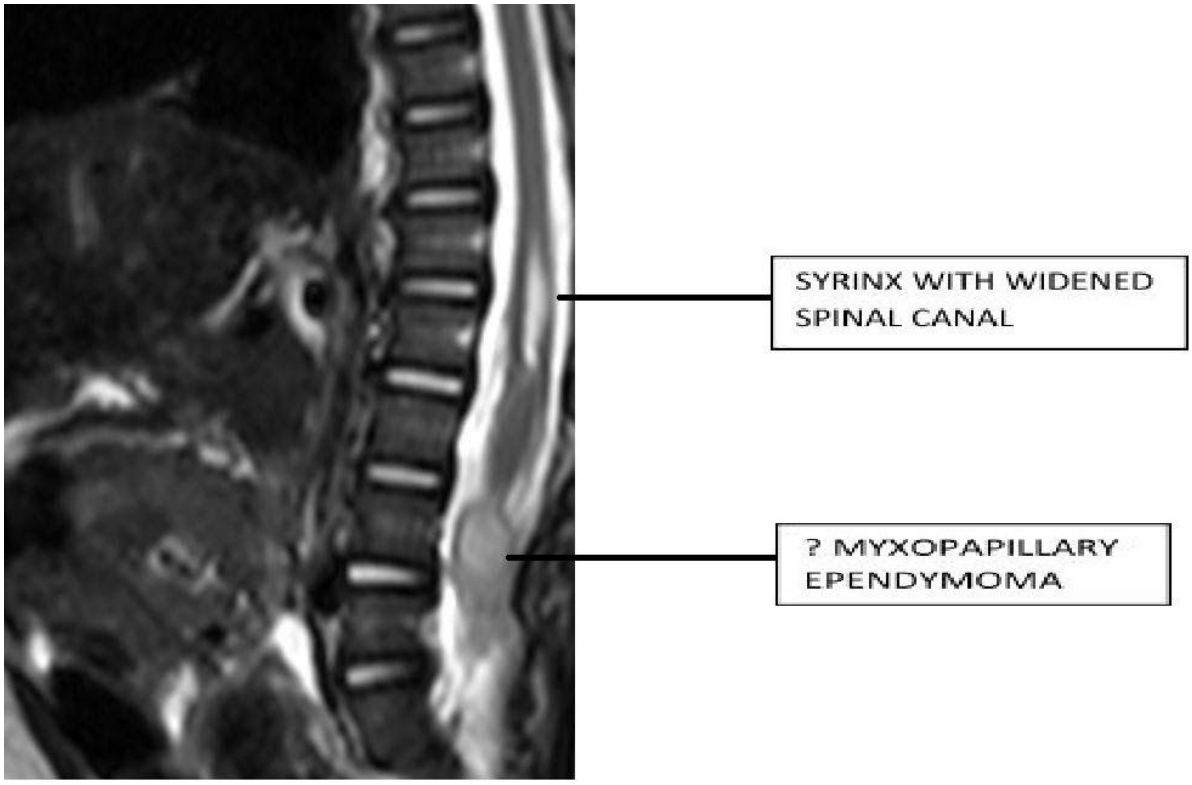
LFT, RFT, Electrolytes – Within Normal Limits.

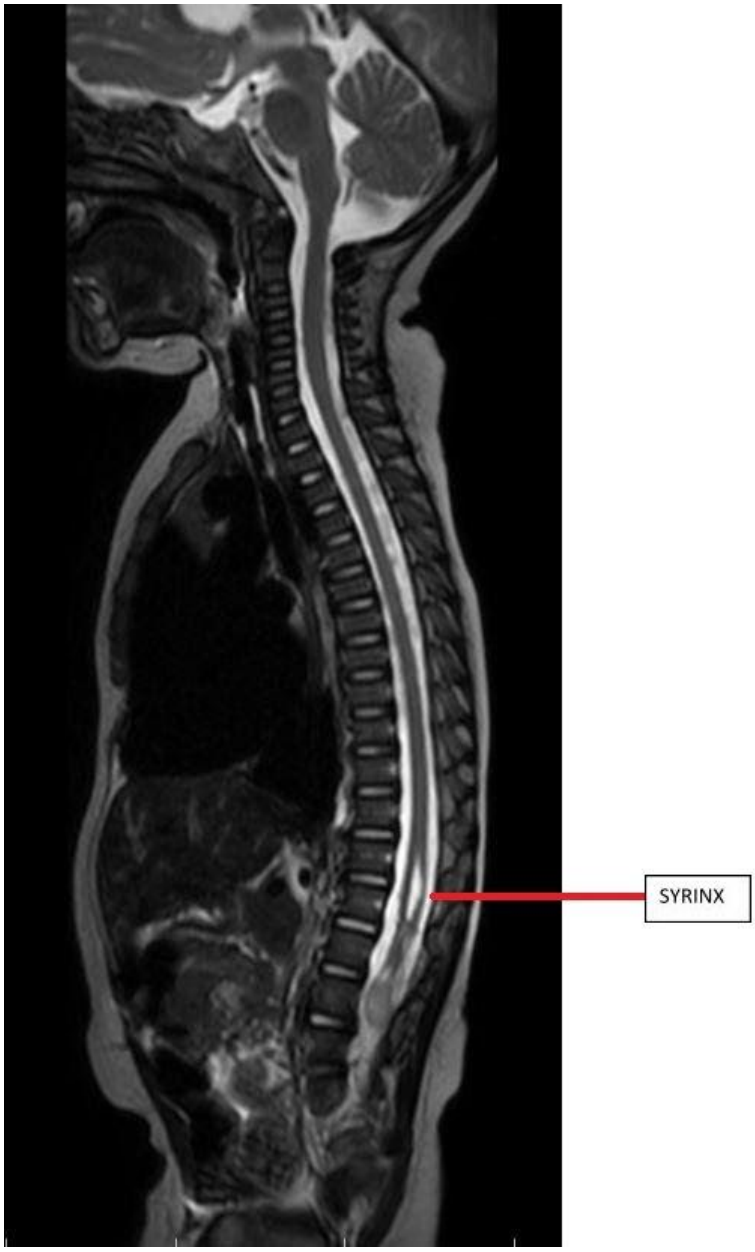
MRI LUMBAR SPINE (under general anesthesia) –

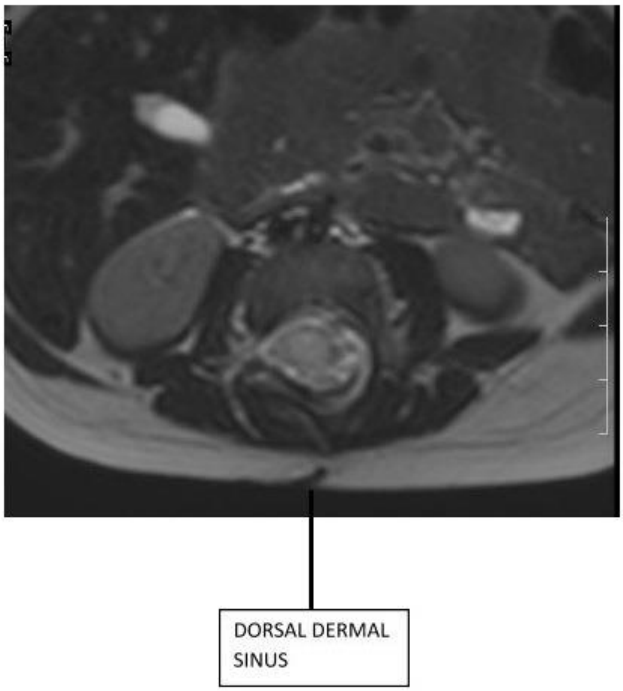
Well defined intradural lesion measuring 24x9x12 mm arising from filum terminale and seen extending from L4-S1 vertebrae. It is causing a mass effect on the Cauda Equina fibers displacing them anteriorly. Similar lesion is seen at the level of S2 vertebra measuring 3×3 mm. ? MYXOPAPILLARY EPENDYMOMA

Intramedullary hyperintensity noted in the center of the spinal cord extending from L1-L2 level measuring 20mm in length with a maximum thickness of 3 mm – SYRINX

A linear hypointense tract measuring 1.5 mm in width is seen extending from the cutaneous plane at L4 level, descending antero-inferiorly, and terminating at the spinous process of the L5 vertebra. – DORSAL DERMAL SINUS


SYRINX WITH WIDENED SPINAL CANAL
? MYXOPAPILLARY EPENDYMOMA


SYRINX



Diagnosis

1. Spina Bifida Occulta with Tethered Cord Syndrome

2. ? Myxopapillary Ependymoma with Syrinx at L1-L2 with Dorsal Dermal Sinus at L4-L5

3. ? Lipoma

Treatment

Plan for neurosurgical intervention through a midline vertical approach with intraoperative nerve monitoring, which would include –

1. Excision of the dermal sinus tract
2. L4-S2 laminectomy and excision of the space occupying lesion (? Lipoma/Myxopapillary ependymoma)
3. Detethering of the cord.

Discussion

The Neural tube is the structure in the developing embryo that eventually becomes the brain, spinal cord, and the tissues that enclose them. The neural tube forms early in pregnancy and closes by the 28th day after conception. [1]

Neural tube defects are one of the most common congenital anomalies due to the failure of closure of the neural tube during embryogenesis. These defects can be open (exposed neural tissue), e.g. craniorachischisis, anencephaly; or closed (neural tissue covering intact), e.g. Encephalocele, Spina Bifida Occulta. [1]

Specific folate deficiencies at the cellular level may be responsible for NTDs because of deranged bioavailability. There can be presence of autoantibodies binding the folate receptors and blocking cellular uptake of folates. [1,2].

Folates play a major role in nucleotide synthesis. Rapidly dividing cells of the neural tube in embryos require synthesis of a large number of nucleotides to allow DNA replication [1] and hence deficiency of folate during the first trimester of pregnancy has been implicated in the increased incidence of NTDs.

Spina bifida, depending on the level of the lesion, can cause interruption of the spinal cord at that site- resulting in paralysis of lower limbs, bowel/bladder dysfunction, anesthesia of overlying skin, and abnormalities of hips, knees, and feet. [3] It is also commonly associated with hydrocephalus and Arnold Chiari malformation which cause an increase in size of the head.

Spina Bifida Occulta is the failure of bony fusion of the vertebral column that is covered by skin, with or without an overlying tuft of hair, lipoma, or sinus. [4]

Tethered cord syndrome (TCS) is a common association with Spina Bifida, as seen in the above-discussed case as well. It is a neurological disorder caused by tissue attachments that limit the movement of the spinal cord within the spinal column. These attachments can cause an abnormal stretching of the spinal cord. The magnitude and degree of traction govern the reversibility of cord dysfunction.

Failure of regression of the caudal cell mass gives rise to a hypertrophic or fibrous filum terminale. Decreased viscoelasticity of a thickened or fatty filum places traction on the lower spinal cord, displacing the conus below the level of L2. TCS presents with cutaneous and neurological symptoms. [5]

Cutaneous features (predominantly seen in neonates and infants): Dimples at the level of lumbar/upper sacral spine, hypertrichosis, hemangioma, dermal sinus, subcutaneous lipomas.

Neurological features: Lower extremity weakness, muscle atrophy, sensory loss in the perineal region, abnormal gait (toe walking, due to tight heel cords), Limb discrepancies. [5]

Detethering is performed if clinical signs and symptoms of deterioration are present. 10-20% of children may require repeat surgeries for correction as symptoms of tethering may reappear during periods of growth. [6]

Syrinx or Syringomyelia is the presence of fluid-filled cyst within the spinal cord, which may grow over time. When the syrinx is present in the distal third of the cord, it is called "Terminal Syringomyelia" and is often associated with tethered cord and spina bifida. [7] Radial tension on the spinal cord due to intermittent low pressure of the cord parenchyma may draw in interstitial fluid and result in enlargement of the syrinx.

MRI is the gold standard for diagnosis and Syringomyelia is considered one of the most important radiological findings in cord tethering.

Immediate surgical intervention is not necessitated unless the patient is symptomatic or the syrinx is extensive. [8]

Ependymomas are rare tumors which take origin from the neuroectoderm and are classified as Grade I – myxopapillary ependymoma, subependymoma; Grade II ependymoma; Grade III – Anaplastic ependymoma [9]

Myxopapillary Ependymoma is slow-growing, usually benign, and occurs in the lower part of the spinal column. They are known to recur locally and are treated with aggressive surgery, chemotherapy, radiation therapy. [10]

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