E.1. Diagnosis and Management of Hydrocephalus and Spinal Dysraphism

a. Hydrocephalus

i. Definition of acute and chronic hydrocephalus:

Hydrocephalus is the imbalance between spinal fluid production and absorption leading to a build-up of cerebrospinal fluid (CSF).

ii. Normal physiology and pathophysiology of hydrocephalus:

Cerebrospinal fluid (CSF) is a liquid that normally surrounds the brain and spinal cord, serving to cushion the nervous tissue as well as wash away metabolic byproducts. It probably serves other functions, which are not as well understood. CSF is made within the brain by a specialized tissue known as choroid plexus. This specialized modified cuboidal epithelium secretes CSF at a constant rate of .37 cc/minute, or roughly 500 ccs per day. The CSF production rate is energy dependent and constant. The composition of this fluid is similar to an ultrafiltrate of plasma. Choroid plexus in found within the 4 ventricles of the brain, the two lateral ventricles, the midline third ventricle and the fourth ventricle of the posterior fossa. The CSF normally circulates through these ventricles by way of communication pathways. The two lateral ventricles join the third ventricle by way of the Foramena of Monro. The third ventricle joins the fourth ventricle by way of the Aqueduct of Sylvius, and CSF exits the fourth ventricle by way of the midline foramen of Magendie and the lateral Foramina of Luschka. CSF then circulates up over the convexity of the cerebral hemispheres where it is absorbed by another specialized tissue, the arachnoid granulations. The arachnoid granulations are a single cell layer of cuboidal epithelium which allow CSF to cross into the venous system in an energy passive process by forcing fluid across the membrane, utilizing the pressure differential between the intracranial pressure (ICP) and the pressure within the venous sinuses. Normally, the arachnoid granulations can absorb several times more than that which is produced; however, if the arachnoid granulations become scarred because of trauma or infection, or if they become obstructed by blood products from a hemorrhage, then CSF can no longer be absorbed well and begins to build up in the subarachnoid spaces, putting pressure on the brain. If CSF flow is blocked at this level, this is termed "communicating hydrocephalus". If the CSF pathways are blocked elsewhere, such as at the level of the aqueduct of Silvius, then this is termed obstructive hydrocephalus. As the intracranial pressure begins to rise in response to a build up of CSF, the patient becomes symptomatic.

iii. Symptoms of hydrocephalus

aa. Acute:

If the blockage of CSF flow happens rapidly, such as following a sudden hemorrhage, the brain has little time to compensate and the intracranial pressure rises rapidly and to a sufficient degree to cause rapid deterioration into coma. Patients will typically experience headache, nausea and vomiting, followed in a period of a few hours by confusion, agitation and then somnolence. If acute hydrocephalus is not treated emergently, the patient’s intracranial pressure will reach the point at which cerebral perfusion is compromised and the patient will deteriorate from coma to death.

bb. Chronic:

If there is partial obstruction or partial blockage to CSF absorption such that CSF pressure builds gradually, then the brain accommodates to this change at first, and the onset of symptoms is more insidious. This is typically seen in patients with slow growing tumors. They typically develop headaches and nausea at night or upon awakening in the morning, with improvement in their symptoms as they get up and walk around. In infants with open sutures, chronic hydrocephalus manifests by increased head growth, which crosses percentiles on the growth curve.

iv. Signs of hydrocephalus

aa. Acute:

In the young child with an open fontanelle, an infant will develop a bulging fontanelle and separation of the cranial sutures. Rapid head growth is noted in premature and newborn infants. These young infants may also have intermittent episodes of bradycardia and apnea. They may develop crossing of the eyes in response to stretching of the abducens nerves or may develop a conjugate downgaze, termed the "setting sun" sign. In the young infant, irritability and somnolence is a late occurrence.

In older children and adults, the development of papilledema occurs over hours to days and may not be seen in the acute state. Severe headache, vomiting and alteration of mental status are followed by somnolence as cortical perfusion becomes further compromised.

bb. Chronic:

Chronic hydrocephalus beyond infancy often manifests similarly to the syndrome of Normal Pressure Hydrocephalus in which the patient presents with an ataxic gait, urinary incontinence, and either dementia or a decline in short-term memory. Patients may describe intermittent headaches which are typically worse upon arising first thing in the morning or which awaken them from sleep at night. They may have chronic papilledema or optic atrophy with constriction of their visual fields.

v. Radiographic diagnosis of hydrocephalus

aa. Skull x-ray:

Skull radiographs can demonstrate findings of raised intracranial pressure, are inexpensive, and are readily available in most physicians' offices. Separation of the cranial sutures, demineralization of the sella turcica, or a J-shaped sella may indicate chronically raised intracranial pressure.

bb. Ultrasound:

In the newborn infant with an open fontanelle, sonography at the bedside can demonstrate the ventricular size and large subdural collections. Small subdurals can be missed. Insonation through the mastoid can image the posterior fossa and rule out 4th ventricular masses.

cc. Computerized tomography:

This is the imaging modality of choice for screening for hydrocephalus. It is relatively inexpensive and is of sufficient detail to rule out most tumors which might obstruct the ventricular system.

dd. Magnetic resonance imaging:

MRI gives the highest resolution image of the brain. It has the advantage of multiplanar imaging, which can be useful in determining subtleties such as agenesis of the Foramen of Monro or aqueductal stenosis. With resolution down to 0.5mm, a MRI is unlikely to miss even the smallest of tumors.

vi. Differential diagnosis of hydrocephalus

aa. Acute:

The differential diagnosis of acute hydrocephalus is age dependent. In the premature infant, it will most commonly be secondary to a spontaneous intraventricular hemorrhage. In the newborn, it may be secondary to a congenital abnormality of the CSF pathways, may be secondary to neonatal meningitis, or may be caused by a congenital brain tumor.

bb. Chronic:

Chronic hydrocephalus is caused by a slowly growing brain tumor until proven otherwise. It may be secondary to a congenital abnormality such as aqueductal stenosis, or possibly to one of the chronic meningitides, but CT or MRI must rule out a mass lesion first.

vii. Treatment of hydrocephalus:

The treatment of hydrocephalus is dependent upon its cause. Acute hydrocephalus secondary to hemorrhage or infection is often transient and can be managed by temporary CSF drainage, either by serial lumbar punctures (LPs) or by placement of a temporary ventricular drain until the underlying pathology has been dealt with. Chronic hydrocephalus has classically been treated by a shunt, which is a plastic tube and valve system which offers a manmade plumbing system to replace the natural one which is no longer working. This most commonly involves placement of a tube into the ventricle which exits the skull through a drilled hole (burr hole) and is connected to a one way pressure regulating valve and then a distal tube which drains excess fluid into another body cavity where it can be absorbed. Most often, this is the peritoneum.

b. Spina Bifida

i. Definition of Spina Bifida:

Spina bifida is a developmental abnormality in which there is incomplete fusion of the dorsal elements composing the roof of the spinal canal. If this birth defect is skin covered, it is termed "occulta". If the defect is open and the neural elements are exposed, it is termed "cystica" or "aperta".

ii. Normal development and abnormal development related to spina bifida:

In the course of normal development, the human embryo at day 18 of gestation is composed of 3 primordial layers of tissue, the ectoderm, the mesoderm, and the endoderm. Shortly thereafter, the ectoderm begins to develop two raised areas, one on either side of the primitive streak. These folds of tissue comprise the neural crest tissue, which curves together and fuses across the midline. This fusion expands in both the rostral and caudal directions to form the neural tube. The anterior neuropore closes at around day 24 of life and the posterior neuropore around day 28. Failure of closure of the anterior neuropore will cause anencephaly,

whereas failure of closure of the posterior neuropore is associated with spina bifida cystica.

iii. Signs of spina bifida

aa. Open:

Spina bifida cystica of aperta is being increasingly diagnosed by prenatal ultrasound. When not diagnosed prenatally, it generally becomes readily apparent at birth, with the fetus being born with a large head and a myelomeningocele. This condition is usually associated with additional abnormalities such as pes cavus deformity of the feet and neurogenic bowel and bladder.

bb. Closed:

Spina bifida occulta can be a variant of normal, with 5% of the population demonstrating incomplete fusion of the neural arches on spine x-ray. Most of the time, this is not associated with neural abnormalities. At times, the incomplete arch is accompanied by other midline lumbar ectodermal abnormalities. These include an abnormal pit in the skin, representing a rudimentary sinus tract, an abnormal lipomatous collection, a tuft of hair, or an area of cutis aplasia (abnormal skin similar to a birth mark). When found on screening physical exam, this should alert the physician to the possibility of an underlying dysraphism. The filum terminale is the terminal extension of the pia of the spinal cord. It forms a small linear structure, which normally connects the end of the conus medullaris to the dura at the end of the thecal sac. In the fetus, the spinal cord extends to the end of the sacral spinal canal, but over time, there exists a differential rate of growth between the vertebrae and the neural elements such that the end of the spinal cord migrates rostrally within the spinal canal. At birth, the end of the conus in normally around L3, and by six months of age, it is normally between T12 and L2. In cases of abnormal development, a thickened filum terminale, a spinal lipoma, or the bony spicule associated with diastematomyelia may serve to tether the spinal cord and prevent the normal rostral migration. This tethering will lead to progressive dysfunction of the distal spinal cord, that which controls bowel and bladder function, sexual function, and distal lower extremity function.

iv. Symptoms of spina bifida

aa. Open:

Spina bifida aperta is generally too obvious to be symptomatic. The infant has deformity of the lower extremities, an enlarged head circumference, and a neural tube defect, which is obvious. In some instance, these infants may develop symptoms of hindbrain compression secondary to a Chiari malformation. This malformation is commonly found in patients with spina bifida aperta and results in their brainstem structures being in their cervical spinal canal. Chiari symptoms at this age may consist of drooling, feeding difficulties, a hoarse or high pitched cry, vocal cord paralysis or other signs of lower cranial nerve dysfunction.

bb. Closed:

The symptomatic spina bifida occulta typically becomes symptomatic beyond infancy, typically following a growth spurt. Classically, the young child who has become toilet trained begins to experience urinary incontinence and urgency. This is often accompanied by back pain exacerbated by exercise, similar to the

syndrome of lumbar pseudoclaudication seen in elderly adults. Numbness of the legs, dysesthesias in the lower extremities and motor symptoms can also be seen. Most of these children will have a history of chronic constipation.

v. Radiographic diagnosis of spina bifida

aa. Plain radiographs:

Plain radiographs of the spine are generally diagnostic. Occasionally, more subtle abnormalities will be found such as the midline upper lumbar calcification of diastematomyelia or widening of the pedicles from a chronic intraspinal mass such as a spinal arachnoid cyst.

bb. Ultrasound:

In the newborn period, the dorsal arches of the spine are cartilaginous and have yet to ossify. In the first few weeks of life, ultrasound over the distal spine can adequately image the spinal canal and cord to determine the level of the conus medullaris, detect intraspinal lipomas and fluid collections such as syringomyelia. At this age, a thickened filum terminale can also be detected.

cc. CT and CT myelography:

The CT scan is an excellent mode of imaging abnormal bone anatomy and is important in defining abnormal segmentation defects such as butterfly vertebrae or diastematomyelia. In the MRI era, myelography is only rarely performed in the child.

dd. MRI:

This is the imaging modality of choice for defining abnormalities of the neural elements associated with spina bifida occulta or tethering of the spinal cord. In the newborn period MRI may be difficult due to respiratory and cardiac motion artifact. If the infant is clinically stable, most pediatric neurosurgeons prefer to wait until the infant is 3-6 months old to perform this study.

vi. Treatment:

In the infant born with a myelomeningocele, repair is usually performed within the first 48 hours of life. The early repair of a leaking myelomeningocele is believed to prevent the development of meningitis. Ninety percent of these infants will also have hydrocephalus and will require placement of a ventriculo-peritoneal (VP) shunt. The repaired myelomeningocele always scars into the walls of the spinal canal at the site of repair; therefore, all of these infants have, by definition, a tethered spinal cord. Over time, at least a fourth of children with spina bifida will become symptomatic from this tethering and will require further surgeries to untether the spinal cord.

In the asymptomatic infant noted to have spina bifida occulta detected by a midline lumbar cutaneous signature mark, treatment continues to be controversial. Most pediatric neurosurgeons will untether the asymptomatic infant due to reports of progressive neural dysfunction noted in many infants, which are followed over time without treatment. Given that all neonates are incontinent, it is difficult to assess bowel and bladder function at this age, and once lost, surgical intervention cannot reliably restore function, but can only halt the deterioration; therefore, adequate evidence exists to support prophylactic untethering.