Developmental Brain Anomalies

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Developmental Anomalies

- Dorsal Induction (Neural tube defects)
- Ventral Induction
- Neuronal Migration & Organization
- Phakomatoses
- Myelination (Normal & Delayed)
- Destructive Brain & Skull Disorders
Disorders of Dorsal Induction

Neural Tube Defects

- Anencephaly
- Encephalocele
- Meningocele
- Chiari malformations
History: One day old boy born with an occipital mass
Dx: Chiari III - occipital meningoencephalocele - small posterior fossa - low tonsils, culpocephaly - scalloped clivus, beaked tectum - stenogyria.
Chiari I with syrinx
Chiari I Malformation

- Low cerebellar tonsils
- Small cisterna magna
- Craniocervical anomalies
- Hydromyelia
History: 5 month old boy with a myelomeningocele
Dx: Chiari 2 malformation
History: Newborn male with a mass on the lower back
History: 7 y/o boy with hydrocephalus presented for shunt followup
Dx: Chiari 2 - with agenesis of splenium, polymicrogyria, stenogyria, atretic straight sinus & enlarged deep veins.
Chiari II Malformation

- Caudal displacement of brainstem, cerebellum & 4th ventricle
- Small posterior fossa, concave petrous bone
- Tectal beaking, large mass intermedia
- Batwing frontal horns & hydrocephalus
- Fenestrated falx with interdigitation of medial cortical gyri
- Partial agenesis of the corpus callosum
- Myelomeningocele & hydromyelia
Disorders of Ventral Induction

- Holoprosencephaly
- Septo-optic dysplasia
- Corpus callosal anomalies
- Dandy-Walker syndrome
- Vermian hypoplasia or aplasia
History:  16 y/o female with a 20 week pregnancy

Dx:
History: Newborn girl with microcephaly

Dx:
Holoprosencephaly

Alobar type

- Microcephaly, hypotelorism, cyclopia
- Monoventricle
- Large dorsal cyst
- Fused thalami
- Absent interhemispheric fissure, falx, septum pellucidum & corpus callosum
- Absent superior, inferior & straight sinuses & internal cerebral veins
History: Newborn girl with an abnormal head ultrasound
Dx: Semilobar holoprosencephaly & midline facial cleft
Holoprosencephaly
Semilobar type

- Microcephaly, hypotelorism & cyclopia
- Partial development of posterior brain
  (Interhemispheric fissure, falx, dural sinuses, occipital/temporal lobes and horns)
- Partial cleavage of thalami
- Rudimentary corpus callosum
Holoprosencephaly

Lobar type

- Partial fusion of frontal lobes
- Shallow interhemispheric fissure (anteriorly)
- Square frontal horns & absent septum pellucidum
- Rest of brain normally formed
Septo-optic Dysplasia

De Morsier's syndrome

- Failure of optic vesicle & commissural plate
- Absent septum pellucidum
- Hypoplastic optic nerves & chiasm
- Pituitary/hypothalamic hypoplasia
Septo-optic Dysplasia
History: Newborn boy who had an abnormal intrauterine ultrasound
Dx: Complete agenesis of the corpus callosum
History: 1 y.o. child with developmental delay

Dx: Complete agenesis corpus callosum & septum pellucidum, Midline interhemispheric cyst, Falcine sinus vs. azygous ACA, Optic atrophy, Cortical dysplasia right insular cortex
Partial Agenesis of the Corpus Callosum
History: 24 y/o female with seizures
Dx: Interhemispheric lipoma & partial agenesis of corpus callosum
History: 18 y/o male
Dx: Lipoma of Corpus callosum
Dysgenesis of the Corpus Callosum
History: Newborn boy with abnormal head ultrasound
Dx: Dandy-Walker malformation
Dandy-Walker Syndrome

- Defective development of 4th ventricular roof
- Posterior fossa cyst; hydrocephalus: often
- Large posterior fossa; high torcula; absent falx in the posterior fossa
- Partial Dandy Walker lacks high torcula. Represents a form of cerebellar hypoplasia
- Mildest aspect of this spectrum would be mega cisterna magna
- DDX: arachnoid cyst, mega cisterna magna.
History: 45 y.o. woman with mental status changes
Mental status change {Page 2}

Dx:
Disorders of Neuronal Migration & Organization

- Lissencephaly, pachygyria
- Polymicrogyria, cortical dysplasia
- Schizencephaly
- Gray matter heterotopia
- Microcephaly
- Megalencephaly
Pachygyria / Lissencephaly
History: 55 y/o woman with refractory epilepsy for 30 years
Dx: Polymicrogyria & septo-optic dysplasia
History: 18 y/o male with chronic seizures
Dx: Pachygyria
History: 22 y/o woman with a history of seizures & right sided weakness
Seizures & weakness

Dx:
History: 5 y/o girl with seizures & left hemiparesis
Dx: Right closed-lip schizencephaly
Left cortical dysplasia

Seizures {Page 2}
History: 46 y/o man with seizures for many years
Dx: Heterotopic gray matter
Gray Matter Heterotopia
History: Newborn with unilateral enlarged lateral ventricle on ultrasound
Dx: Hemimegalencephaly & GM heterotopia
The Phakomatoses

- Neurofibromatosis
- Sturge-Weber syndrome
- Tuberous sclerosis
- von Hippel-Lindau disease
- Ataxia-Telangiectasia
History: 7 y/o boy with headaches
Dx: Dysmyelination (vacuolar myelin)

Neurofibromatosis, Type 1

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History: 6 y/o boy with visual problems
Dx: NF 1 - vacuolar myelin & optic nerve glioma

Visual loss

{Page 3}
Peripheral Neurofibromatosis (NF I)
von Recklinghausen's disease

- Autosomal dominant - chromosome 17
- Cafe au lait spots, axillary freckles, Lisch nodules in iris
- Peripheral/plexiform neurofibromas
- Optic pathway gliomas
- Dysmyelination - Vacuolar myelin
- Sphenoid wing dysplasia
- Scoliosis, vertebral scalloping
History: 9 y.o. girl with multiple bilateral cranial nerve palsies
Cranial nerves

Dx: NF 2 with CN schwannomas
History: 40 y.o. man with pain & paresthesias in both arms & shoulders
Dx: NF 2 with intramedullary neurofibroma

Pain and Paresthesias

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Central Neurofibromatosis (NF 2)

- Autosomal dominant - chromosome 22
- Bilateral acoustic schwannomas
- Multiple meningiomas
- Paraspinal neurofibromas
- Spinal cord ependymomas
Sturge-Weber Syndrome
Sturge-Weber Syndrome
Encephalotrigeminal Angiomatosis

- Facial "port wine" vascular nevus
- Seizures, hemiparesis & hemianopsia
- Meningeal (pial) angiomatosis
- Cortical calcification, gyral enhancement
- Ipsilateral cerebral atrophy
- Enlarged deep venous system
  & choroid plexus
- Angiomas of choroid & visceral organs
History: 2 y/o girl with seizures
Tuberous Sclerosis
Bourneville's Disease

- Autosomal dominant inheritance
- Seizures, adenoma sebaceum, mental retardation
- Hyperpigmented nevi, shagreen patches, rhabdomyomas, angiomyolipomas, subungual fibromas
- Brain tubers/hamartomas: Subependymal, cortex & retina
- Giant cell astrocytoma
History: 48 y.o. man with headache and ataxia
Dx: Hemangioblastoma

History: Ataxia
Hemangioblastoma
von Hippel--Lindau Disease

CNS Angiomatosis

- Autosomal dominant inheritance
- Hemangioblastomas of cerebellum & spinal cord
- Imaging: cystic mass with vascular enhancing nodule
- Retinal angiomas
- Renal cell carcinoma, Pheochromocytoma
Normal Myelination

- Begins during 29th week of gestation
- At birth, myelin present in brain stem & cerebellum - other white matter has long T1 and long T2
- Myelination & decreasing water content shorten T1 and T2
- IR sequence best for first 6 months
- T2 image: Isointense phase at 9-12 months - adult pattern > 3 years
23 week premature baby

T2WIs

FLAIR
Normal Brain Development

T1WIs
History: Newborn with high pitched cry after difficult delivery.
Dx: Normal newborn
History: 4 week old boy (gestational age = 36 wks) with persistent apnea
History: 10 month old boy with developmental delay
Delayed development

FLAIR images
Dx: Markedly delayed myelination
Delayed Myelination
Destructive Brain & Skull Disorders
History: 44 y/o man with seizures
Porencephaly

- Focal brain destruction early in life
- Causes: vascular occlusion, hemorrhage, trauma, post-surgical
- Smooth-walled, fluid-filled cystic cavity
- Ventricular communication variable
- Fluid isointense to CSF
History: 23 week premature baby with suspected herpes encephalitis
23 week premie

Dx: Grade 2 germinal matrix hemorrhage
History: A 31 y/o male with chronic headaches

Dx: Aqueductal stenosis
History: Newborn with a large head
Dx: Hydrocephalus with Aqueductal stenosis
History: 17 month old boy with head size decreasing from 75 to 5 percentile in 1 year
Dx: Craniosynostosis of coronal & lambdoid sutures
History: 28 week fetus with abnormal ultrasound
Dx: Teratoma