MR Imaging features of ‘Malformations of cortical development’ in the Pediatric age group

Saint Louis Children’s hospital,
Mallinckrodt Inst of Radiology
Tejaswini Deshmukh, Prakash Masand
Robert McKinstry
Disclosure

• No financial interests are associated with this educational material
Introduction

• Cortical malformations are frequently encountered in practice and it is important to familiarize oneself with the typical imaging features of some common entities.

• The following exhibit is an endeavor to present this complex topic in the form of a pictorial review.
Key: Abbreviations

- AX – Axial
- COR – Coronal
- SAG – Sagittal
- T1 – T1 weighted images
- T2 – T2 weighted images
- FLAIR – Fluid attenuation inversion recovery
- DTI – Diffusion tensor imaging.
Schizencephaly

- Gray-matter lined clefts extending from pial surface of cerebral cortex to ependymal lining of lateral ventricle
- Lined by polymicrogyric cortex
- Gyri and sulci radiate into the cleft
- Closed lip (small defect) or open lip (large defect)
Closed-lip Schizencephaly

AX T2

AX T1

COR T1
Open-lip Schizencephaly

Large cleft communicating with right lateral ventricle
Open-lip Schizencephaly

- Large cleft communicating with right lateral ventricle (large arrows)
- Periventricular calcifications : Intrauterine TORCH infection (arrowheads)
Septo-optic dysplasia

- Absence of septum pellucidum
- Hypoplasia of optic nerves
- Box-like frontal horns
- Associations: Schizencephaly, gray-matter heterotopia, Ventriculomegaly, Ectopic posterior pituitary, hypoplastic corpus callosum
Septo-optic dysplasia

Absent Septum Pellucidum

Hypoplastic Optic Nerves

AX T2

AX FLAIR

COR T1

Flow chart
Holoprosencephaly

- Failure of differentiation & cleavage of prosencephalon
- Incomplete separation of the cerebral hemispheres
- Absence/hypoplasia of falx cerebri with interdigitations of medial cerebral gyri
- Range from minor forms (lobar) to severe forms (alobar)
Holoprosencephaly

• Cavum septum pellucidum is always absent (in all 3 forms)

• Degree of callosal dysgenesis is important for prognosis

• Associated facial anomalies seen

• Some consider septo-optic dysplasia to be the most mild form (a.k.a lobar holoprosencephaly)
• **Alobar:**
  - Complete absence of interhemispheric falx
  - Fused thalami
  - Malformed cortex
  - Single horse-shoe shaped ventricle continuous with a dorsal cyst
Alobar Holoprosencephaly

- No separation of the cerebral hemispheres
- Single horseshoe shaped ventricle
- Absent interhemispheric fissure, falx cerebri, corpus callosum, and septum pellucidum
- Thalami are fused
- Dorsal cyst is seen
Alobar Holoprosencephaly

AX T1

AX FLAIR

AX T2
• **Semilobar**
  - Interhemispheric fissure partially present
  - Frontal and sometimes parietal lobes continue across the midline
  - Thalami are partially fused
  - Monoventricle
Semilobar Holoprosencephaly

Cranial sonogram
Semilobar Holoprosencephaly

- Incomplete separation of the cerebral hemispheres
- Monoventricle
- Partially absent interhemispheric fissure
- Thalami are partially fused
- Associated cleft lip
• Lobar
  - Only minor changes seen
  - Anterior falx, septum pellucidum absent
  - Hypoplastic frontal lobes & horns
Lobar Holoprosencephaly

Absent septum pellucidum and hypoplastic frontal horns
Microcephaly

- Head circumference < 3SD below normal
- Reduced white matter
- Microcephaly with Simplified gyral pattern
  - Too few sulci
  - Normal cortical thickness
- Microlissencephaly
  - Smooth cortical surface
  - Thickened cortex
Microcephaly

Decreased brain size with thinning of the mantle and increased subarachnoid space
Smooth cortical surface characteristic of microlissencephaly

AX T2  SAG T2  COR T2
Microcephaly

Small head circumference with a simplified gyral pattern, and too few sulci.
Hemimegalencephaly

- Hamartomatous overgrowth of part/all of a hemisphere
- **Cortex** – dysplastic with broad gyri, shallow sulci, indistinct gray-white matter junction
- **White matter** – heterogenous signal due to heterotopia and dysplastic neurons & glia
- **Lateral ventricle** – enlarged, straight frontal horn pointing superiorly and anteriorly
Hemimegalencephaly

• Enlarged right hemisphere
• Dysplastic right cortex
• White matter hyperintensity
• Blurred gray-white matter junction distinction
Hemimegalencephaly

- Enlarged left hemisphere
- Dysplastic left cortex with shallow gyri and few sulci
- White matter hyperintensity
- Blurred gray-white matter junction distinction
Hemimegalencephaly

Abnormal cortex, 3D volume rendered images
Hemimegalencephaly

- Subtle enlargement of right hemisphere
- Abnormal hyperintense signal within the right periventricular white matter
Tuberous Sclerosis

- Inherited tumor disorder with multiorgan hamartomas
- Subependymal nodules, may calcify
- Subependymal giant cell astrocytoma
- Cortical / subcortical tubers (flame shaped)
Tuberous Sclerosis

Subependymal nodules

AX T2  AX T1  AX PC T1
Tuberous Sclerosis

Calcified subependymal nodules

T2*

AX T2

AX FLAIR
Tuberous Sclerosis:
Cortical tubers:
- Increased signal on T2 and FLAIR (arrows)
- Calcification in cortical tubers (arrowheads)
Tuberous Sclerosis

- Cortical tubers
- Subependymal nodules
Tuberous Sclerosis

Subependymal giant cell astrocytoma

AX T1

AX FLAIR

AX, COR, SAG PC T1

Flow chart
Lissencephaly

- Smooth brain - paucity of sulcal & gyral development
- Thick cortex
- Hour-glass configuration (figure of “8”) of brain with shallow sylvian fissures
- **Complete form** - Agyria (absent gyri)
- **Incomplete form** - Pachygyria (few broad, flat gyri)
Lissencephaly

- Smooth figure of “8” configuration of the brain
- Abnormal 4-layer cortex
- Arrested neuronal migration from the germinal matrix to the cortical surface
- Developmental delay
- Seizures
Lissencephaly

- Genetics
- LIS1 (commonest): Hemizygous deletion or mutation
- XLIS or DCX:
  - lissencephaly in hemizygous males
  - subcortical band heterotopia in carrier females
Lissencephaly

- Large LIS 1 gene deletion on Chromosome 17p13.3 plus deletion of 14-3-3 epsilon
- Facial features
- Prominent forehead with bitemporal wasting
- Short nose with upflared nares
- Micrognathia
- Hypertelorism
- Low set ears
Lissencephaly

- Agyria, with hourglass configuration caused by shallow sylvian fissures
- Prominent occipital horns of lateral ventricles
Lissencephaly

Smooth brain - absence of sulcal & gyral development, with shallow sylvian fissures
Lissencephaly

Lissencephaly with associated left microphthalmia and retinal detachment = Miller Dieker syndrome
Lissencephaly

Head CT scan - Lissencephaly
Lissencephaly

Shallow Sylvian fissures with bilateral broad and flattened gyri
Polymicrogyria

• Neurons reach the cortex but distribute abnormally forming multiple small undulating gyri
• Irregular cortical surface & gray-white matter junction
• Infolding of cortex (cortical dimple)
Polymicrogyria

- Abnormal convolutional pattern of the brain characterized by an excessive number of abnormally small gyri
- Migrational defect (but not a developmental arrest) causing a cobblestoning defect on the surface of the brain
Symmetric Polymicrogyria
Focal Polymicrogyria

Left polymicrogyria, mimics a mass
Focal Polymicrogyria

Cortical dimple in the left parietal lobe, at site of focal abnormality
Polymicrogyria

Bilateral excessive abnormally small gyri
Bilateral Perisylvian Syndrome

- Genetic disorder with polymicrogyria lining the Sylvian fissures
- Characterized by seizures as well as specific array of clinical findings, including hypotonia
Bilateral Perisylvian Syndrome
Heterotopias

- Collections of normal neurons in abnormal locations anywhere from the subependymal region to the cerebral cortex
- Imaging characteristics match gray matter
- **Types**: Subependymal, Focal subcortical, Band heterotopia
Subependymal Heterotopia
Focal Subcortical Heterotopia with polymicrogyria
Band Heterotopia

• Also referred to as:
  - Double cortex heterotopia or diffuse heterotopia
• Relative lack of normal interdigitations of white matter
• Symmetric ribbons of gray matter in the centrum semiovale separated from the cortex and the ventricular walls by surrounding white matter
Band Heterotopia
Band Heterotopia

Band of gray matter, flanked by white matter on both sides
Summary

- We have now seen the MR imaging features and typical appearances of some of the common malformations of cortical development.
- The broad classification used is easy to remember and helpful in characterizing these entities.