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Congenital Malformation of the Brain

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1. Introduction

More than 2000 different congenital malformations of the brain have been described in the literature, and their incidence is reported to be about 1 percent of all live births.¹ Since the congenital anomalies of the brain are commonly encountered in day to day practice, it is very important for every radiologist to be familiar with the basic imaging findings of common congenital anomalies to make a correct diagnosis necessary for optimum management of these conditions. Magnetic resonance imaging (MRI) is very useful in studying these malformations. The aim of this chapter is to provide an overview of all important and routinely encountered congenital malformations of the brain.

2. Normal brain development

Congenital anomalies of the brain are extremely complex and are best studied by correlating with embryological development. Basic events in normal brain development includes the following four stages:¹

Stage 1: Dorsal Induction: Formation and closure of the neural tube

- Occurs at 3-5 weeks
- Three phases: Neurulation, canalization, retrogressive differentiation
- Failure: Neural tube defects (Anencephaly, Cephalocele, Chiari malformations) and Spinal dysraphic disorders.

Stage 2: Ventral Induction: Formation of the brain segments and face

- Occurs at 5-10 weeks of gestation
- Three vesicles (prosencephalon, mesencephalon, and rhombencephalon) form the cerebral hemispheres/thalamus, midbrain, and cerebellum/brain stem respectively.
- Development of face
- Failure: Holoprosencephalies, Dandy Walker malformation, Cerebellar hypoplasia/dysplasia (Chiari-IV), Joubert syndrome, Rhombencephalosynapsis, Septooptic dysplasia, and facial anomalies.

Stage 3: Migration and Histogenesis

- Occurs at 2-5 months of gestation
- Neuronal migration from germinal matrix to the cortex
- Cortical organization
- Failure of Migration: Heterotopias, Agyria-pachygyria, Polymicrogyria, Lissencephaly, Schizencephaly, Corpus callosal agenesis, Lhermitte-Duclos disease.
- Failure of Histogenesis: Aqueductal stenosis, Arachnoid cyst, Megalencephaly, Microencephaly, Neurocutaneous syndrome (phakomatoses), Congenital vascular malformation, and Congenital neoplasms.

Stage 4: Myelination

- Begins at 6 months of gestation; matures by 3 years.
- Progresses from caudal to cephalad, from dorsal to ventral, and from central to peripheral
- Failure: Dysmyelinating diseases, Metabolic disorders.

3. Classification of congenital malformation of brain

A number of classification systems have been proposed, but with regards to our basic purpose, simplified classification of brain malformations has been taken into account, which is as follows:^{1,2}

a. Disorders of Organogenesis

- Neural tube closure
- Diverticulation and cleavage
- Sulcation/cellular migration
- Cerebellar hypoplasia/dysplasia

b. Disorders of Histogenesis

- Neurocutaneous syndromes (Phakomatoses)

c. Disorders of Cytogenesis

- Congenital vascular malformations
- Congenital neoplasms of brain

d. Disorders of Myelination

- Leukodystrophies

4. Imaging features of congenital malformations of the brain

4.1 Disorders of organogenesis

4.1.1 The chiari malformations

Chiari I Malformation^{1,2} (Figure 1)

- Termed as tonsillar ectopia
- Herniation of elongated peg like cerebellar tonsils into the upper cervical canal through the foramen magna.
- Abnormal tonsillar descent below the opisthion-basion line
 - At least 6mm in first decade
 - 5mm in the 2nd /3rd decade

- 4mm between 4th-8th decade, and
- 3mm by 9th decade
- Causes of abnormal tonsillar descent
 - Congenital asymptomatic tonsillar ectopia
 - Intracranial hypotension due to chronic CSF leak (sagging brain)
 - Long-standing compensated hydrocephalus
 - Craniovertebral anomalies (Basilar invagination, Platybasia, Atlantooccipital assimilation)
- Associated anomalies
 - Syringohydromyelia(30-60%)

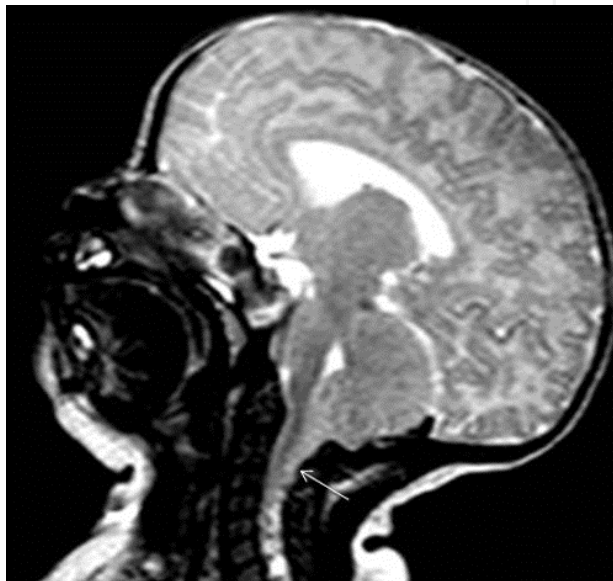


Fig. 1. Chiari I Malformation. Sagittal T1W MR scan of brain shows peg like low lying tonsil in upper posterior cervical canal (thin white arrow). The cerebellar tonsils lie more than 10mm below the foramen magnum. Note mild enlargement of supratentorial ventricular system.

Chiari II Malformation^{1,2} (Figure 2)

- Complex anomaly involving skull, dura, brain, spine and the cord
- Skull and dural involvement
 - Luckenschadel (lacunar skull), concave clivus and petrous ridges
 - Small and shallow posterior fossa with low lying transverse sinuses and torcular herophilli
 - Hypoplastic tentorium cerebelli with gaping (heart shaped) incisura
 - Hypoplastic, fenestrated falx cerebri with interdigitating gyri
 - Gaping foramen magnum
- Brain involvement
 - Cascading protrusion of vermian nodule, fourth ventricle choroid plexus and the medulla into the spinal canal with formation of cervicomedullary spur and kink
 - Upward herniation of cerebellar hemispheres and vermis through gaping incisura (towering cerebellum) producing tectal deformity (beaked tectum)
 - Cerebellar hemispheres creep around to engulf the brain stem
 - Large massa intermedia
 - Hydrocephalus (90%) with serrated appearance of lateral ventricles

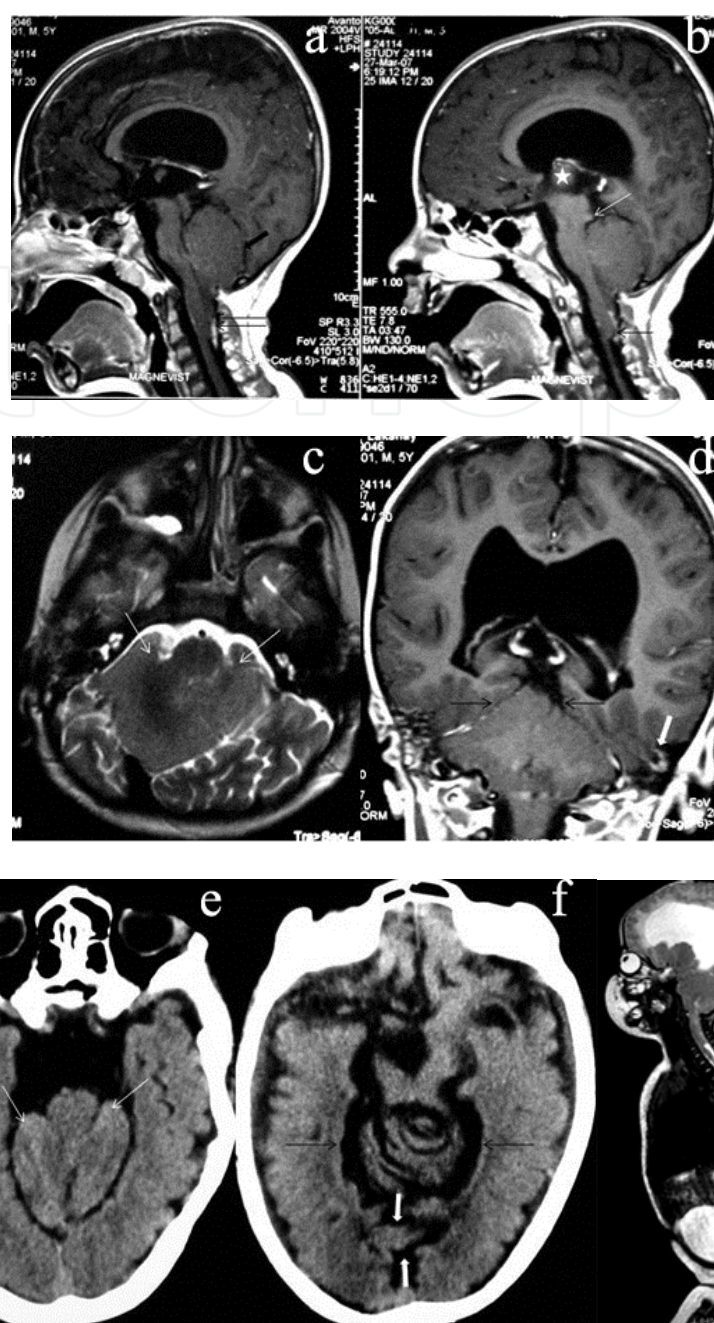


Fig. 2. Chiari II Malformation. Sagittal T1W MR images(a,b) of brain show a small posterior fossa, low lying tectorium cerebelli (thick black arrow), cascade of inferiorly displaced vermis and choroid plexus into upper posterior cervical canal (double thin black arrow), medullary spur (thin white arrow), large massa intermedia(asterix). The cerebellar hemispheres extend antero-medially(thin white arrow) to engulf the brain stem on axial T2W(c) image. Coronal T1W image(d) reveals low lying torcular herophili(thick white arrow), gaping incisura and towering cerebellum(thin black arrow). Also note bilateral hydrocephalus. Plain axial CT scan(e,f) another patient shows cerebellar hemispheres engulfing the brain stem, wide gaping incisura plugged with upward herniating dysmorphic cerebellum(thin black arrow), gross gyral interdigitation and stenogyria (thick white arrow). Screening of whole spine in another patient with ACM-II demonstrates associated lumbar myelomeningocele (asterix) on sagittal T2W image (g).

- Spine and cord involvement
 - Myelomeningocele (nearly 100%)
 - Syringohydromyelia (70-90%)
 - Diastematomyelia
- Associated anomalies
 - Corpus callosal dysgenesis, heterotopias, polymicrogyria

Chiari III Malformation^{1,2,3} (Figure 3)

- Features of Chiari II malformation with a low occipital or high cervical encephalocele
- The encephalocele may contain meninges, cerebellum, occipital lobe or brain stem. Cisterns and dural sinuses may also be present



Fig. 3. Chiari III Malformation. Sagittal T1w MR scan shows features of ACM II, evidenced as a large massa intermedia (asterix), tectal beaking (thin black arrow), inferior herniation of the cerebellar tissue through the foramen magnum into the upper posterior cervical canal (thin white arrow), and a low occipital meningoencephalocele (thick white arrow).

Chiari IV Malformation^{1,2}

- Severe cerebellar hypoplasia or dysplasia, small brain stem and large posterior fossa CSF space
- No hydrocephalus and other CNS anomalies

Cephaloceles^{1,2,4} (Figure 4-10)

- Characterized by protrusion of intracranial contents through a congenital defect in the dura and skull
- Usually located at or near the midline.
- Pathological classification of cephaloceles (based on the contents of the herniated sac)
 - Meningocele (leptomeninges and CSF)
 - Meningoencephalocele (leptomeninges, CSF and brain)
 - Meningoencephalocystocele (leptomeninges, CSF, brain and ventricles)
 - Atretic cephalocele (small nodule of fibrous fatty tissue)
 - Gliocoele (CSF lined by glial tissue)

- Anatomic classification of cephalocele (based on location)
 - Occipital - most common in America, Europe
 - Parietal
 - Temporal
 - Frontal or Frontoethmoidal (Sincipital) - most common in Asia
 - Transsphenoidal - uncommon
 - Nasal
- The herniated brain dysgenetic and non-functional
- Absence or erosion of the crista galli with enlargement of foramen cecum is a constant feature of a nasal cephalocele.

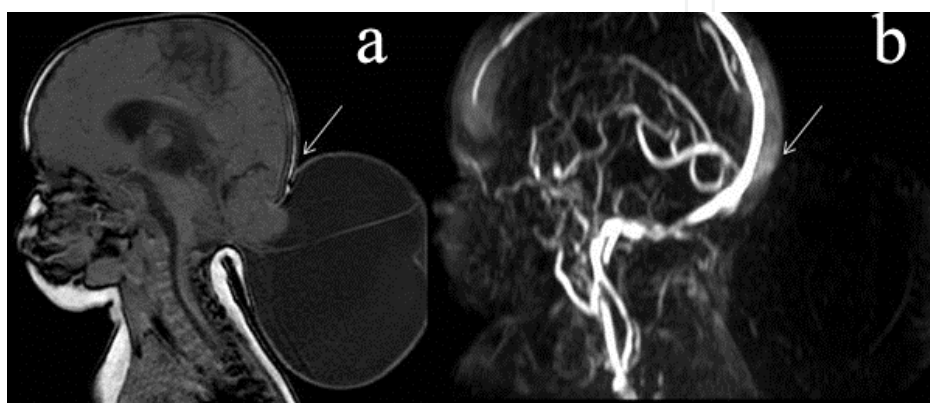


Fig. 4. Occipital cephalocele. Sagittal T1W (a) image shows herniation of severely dysplastic cerebellar tissue and the occipital lobe into a large CSF containing sac through an osseous defect in the occipital bone (thin white arrow). Thin strand of dysplastic brain tissue or septa can be seen traversing the CSF within the sac. Also note small posterior fossa, and deformed brain stem. 2D Time-of-flight venogram (b) demonstrates no herniation of dural venous sinuses in the cephalocele sac. This is important information for the surgeons.

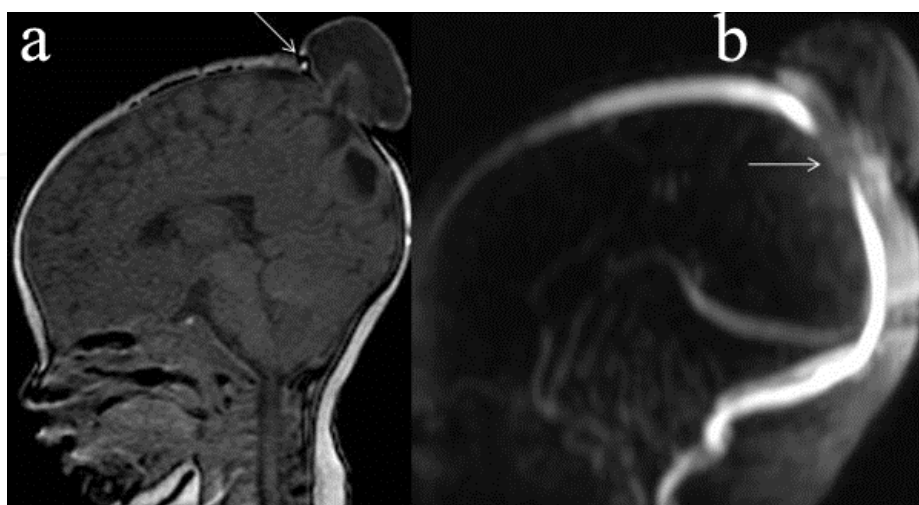


Fig. 5. Parietal cephalocele. Sagittal T1W (a) image of brain shows a small parietal cephalocele containing CSF and dysplastic brain tissue (thin white arrow). 2D TOF venogram (b) shows non-visualization of small segment of superior sagittal sinus in the region of osseous defect consistent with sinus thrombosis (thin white arrow).

- Differential diagnosis of a nasal cephalocele includes congenital nasal masses (e.g. dermoid) where crista galli is present but split.
- Antenatal ultrasound and MRI are useful in evaluation of content of the sac. MR venography is useful to assess major dural sinuses within the herniated sac, which may be responsible for major bleeding during surgery. CT is useful in demonstrating bony defect.
- Associated anomalies
 - Chiari II and III malformation (seen with occipital cephalocele)
 - Corpus callosum agenesis, Dandy-Walker malformation (seen with parietal cephalocele)

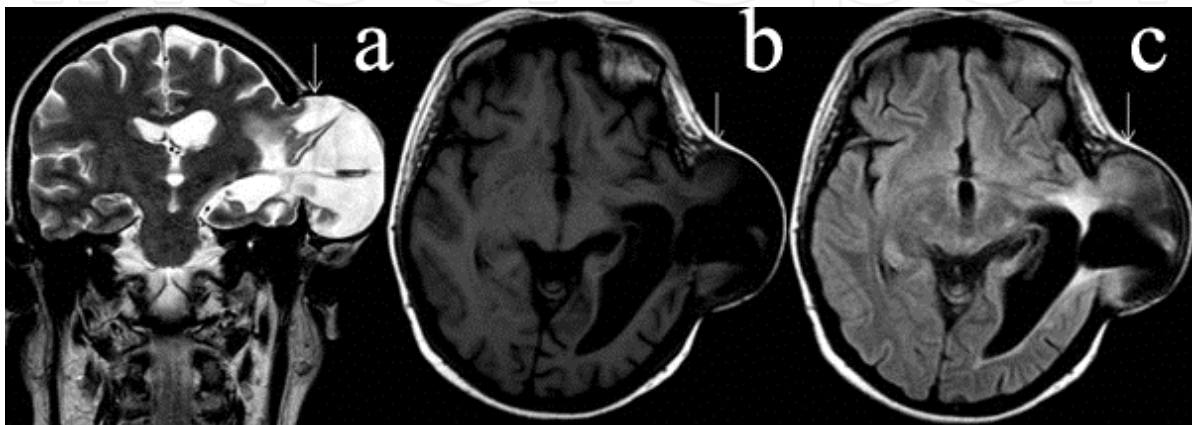


Fig. 6. Temporal cephalocele. Coronal T2W (a), axial T1W (b) and FLAIR (c) image of brain shows a left temporal meningoencephalocele (thin white arrow) containing dysplastic and disorganized left temporal lobe and CSF. Ipsilateral lateral ventricle is dilated and the temporal horn appears stretched towards the bony defect.

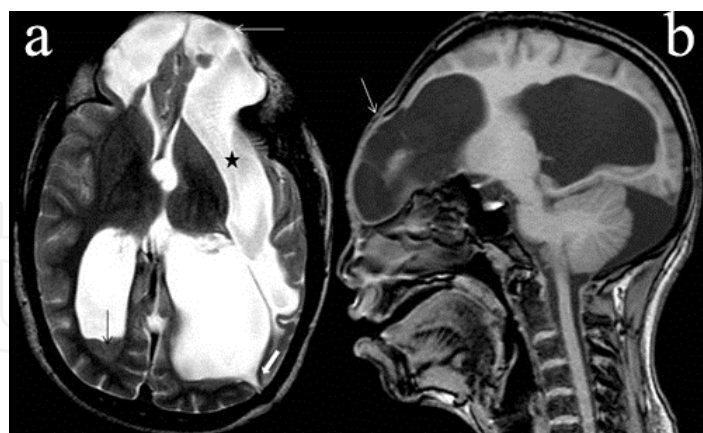


Fig. 7. Frontal cephalocele. Axial T2W(a) and sagittal T1W(b) image of brain demonstrates a large frontal midline meningoencephalocele (thin white arrow) containing dysplastic frontal lobes, stretched frontal horns and the CSF. Left cerebral hemisphere dysraphism is present, seen as a widely open CSF cleft (asterisk) extending across the left external capsule region, and communicating posteriorly with the dilated occipital horn and anteriorly with the cephalocele. Ipsilateral basal ganglion show abnormal morphology. Subependymal heterotopia of right occipital horn (thin black arrow) and a small, narrow left-sided posterior sphenoidal cleft (thick white arrow) is also noted.

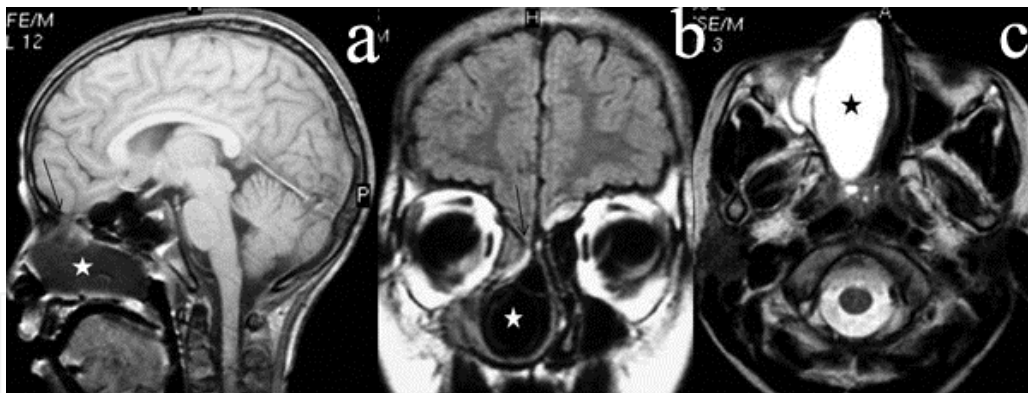


Fig. 8. Nasal cephalocele. Sagittal T1W (a), Coronal FLAIR (b), and axial T2W(c) images reveal a large right sided nasal meningocele containing the CSF (asterix). There is a defect in the skull base in the region of the cribriform plate and crista galli (thin arrow).

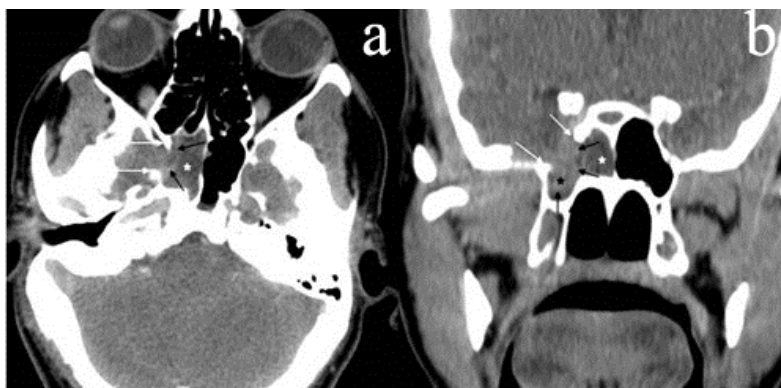


Fig. 9. Sphenoid encephalocele. Axial (a) and coronal (b) NCCT head shows a defect in right lateral wall of sphenoid sinus (thin white arrow) with herniation of small portion of inferomedial part of ipsilateral temporal lobe (black asterisk and thin black arrow). Associated sinusitis is seen involving the ipsilateral sphenoid sinus (white asterisk).

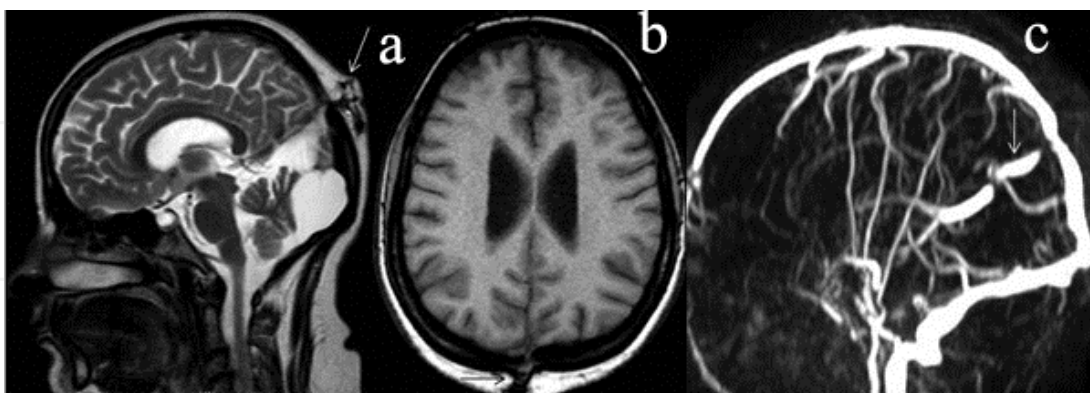


Fig. 10. Atretic occipital cephalocele. Sagittal T2W(a), axial T1W(b) image shows a small subcutaneous mass (thin white arrow) in high occipital region just external to a small defect in the calvarium. Note that the brain is not entering the cephalocele; instead, a thin strand of fibrous tissue is seen extending across the osseous defect, from the surface of the brain to the subcutaneous mass. Small posterior fossa arachnoid cyst is also seen. 2D TOF venogram (c) shows presence of median procencephalic vein within embryonic falcine sinus (thin white arrow) and absence of sagittal sinus.

Corpus Callosum Agenesis^{1,2} (Figure 11,12,13)

- Corpus callosum, a midline commissure connects two cerebral hemispheres
- Develops in cephalocaudal direction, beginning with genu then followed by the body and splenium. The rostrum is last to develop
- Two types: complete or partial
- Complete callosal agenesis
 - Absence of entire corpus callosum, cingulate gyrus and sulcus
 - High riding 3rd ventricle with spoke like orientation of gyri around it
 - Widely separated, parallel and non-converging lateral ventricles.
 - Probst bundles (longitudinal white matter tracts) indent superomedial lateral ventricles
 - Colpocephaly (dilated occipital horns) common, frontal horns small and pointed
 - High incidence of dorsal interhemispheric cyst

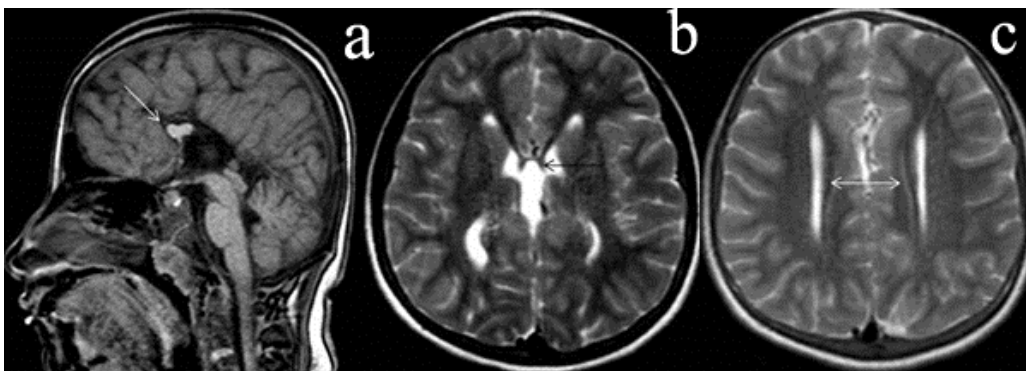


Fig. 11. Corpus callosal agenesis (partial). Sagittal T1W(a) and axial T2W (b,c) image shows presence of only the genu of the corpus callosum (thin white arrow), high riding third ventricle(thin black arrow),widely separated and parallel lateral ventricles(double thin white arrow).

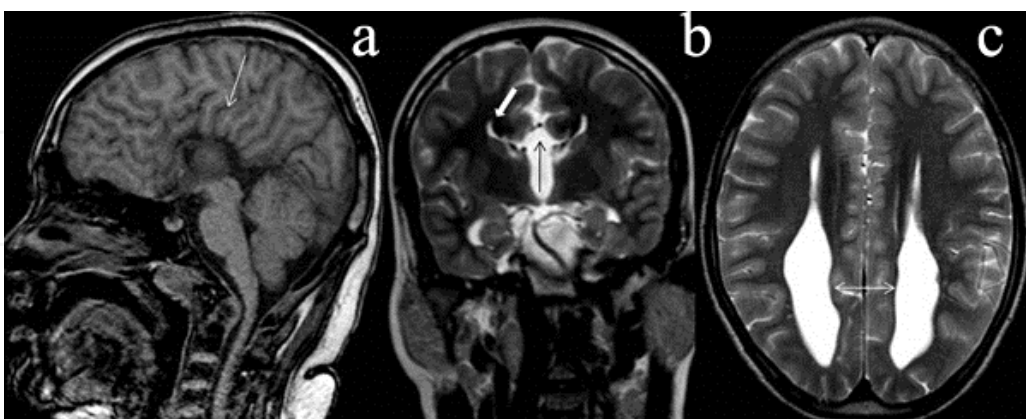


Fig. 12. Corpus callosal agenesis(complete). Sagittal T1W(a) and coronal T2W (b) image shows complete absence of the corpus callosum and cingulate sulcus (thin white arrow), high riding third ventricle communicating with the interhemispheric fissure(thin black arrow), and crescent shaped frontal horns indented medially by white matter tracts of Probst's bundles(thick white arrow). Widely separated and parallel lateral ventricles with colpocephaly are also seen (double thin white arrow) on axial T2W image(c).

- Partial callosal agenesis
 - Splenium and rostrum absent or hypoplastic
 - Genu and body present to various degrees
- Associated anomalies
 - Migration disorders (heterotopias, lissencephaly, schizencephaly)
 - Chiari II malformation
 - Dandy-Walker malformation
 - Holoprosencephaly
 - Corpus callosal lipoma

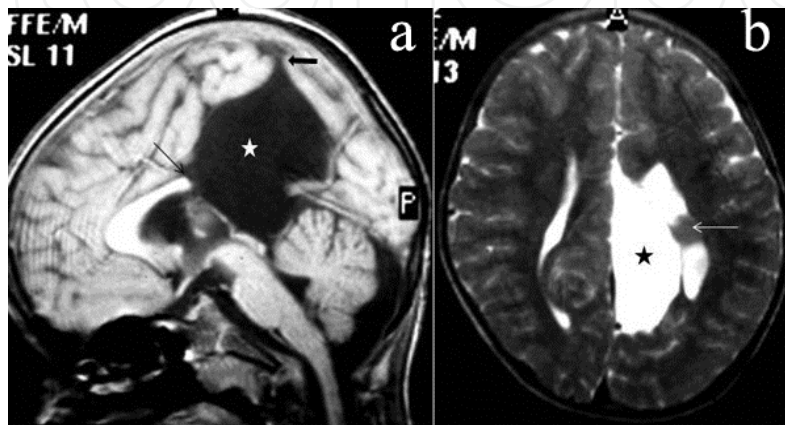


Fig. 13. Corpus callosal agenesis(partial) with dorsal interhemispheric cyst. Sagittal T1W(a) and axial T2W(b) image shows presence of the genu and anterior part of body of the corpus callosum while, the posterior body, splenium and rostrum is absent (thin white arrow). The lateral ventricles are widely separated and a moderately large dorsal interhemispheric cyst (asterix) is present which is seen communicating with the overlying subarachnoid space via a narrow schizencephalic cleft (thin black arrow). Solitary nodular heterotopia can be seen within the body of left lateral ventricle which is mildly dilated (thin white arrow).

4.2 Disorders of diverticulation and cleavage

Holoprosencephaly^{1,2,5} (Figure 14,15,16)

- Characterized by complete or partial failure of cleavage and differentiation of developing cerebrum (prosencephalon) into hemispheres and lobes.
- Cerebellum and brain stem relatively normal
- Classified into three types on the basis of degree of severity
 - Alobar (most severe)
 - Semilobar (moderately severe)
 - Lobar (mildest form)
- Precise boundaries among these three groups does not exist, intermediate cases may be identified
- Alobar Holoprosencephaly
 - Near complete lack of hemispheric cleavage
 - Crescent-shaped monoventricle
 - Absence of septum pellucidum, corpus callosum, falx cerebri, and interhemispheric fissure
 - Fused thalami and basal ganglia

- Associated anomalies: Dorsal interhemispheric cyst, severe craniofacial anomaly
- Semilobar Holoprosencephaly
 - Partial brain diverticulation
 - H-shaped holovertricle with rudimentary occipital and temporal horns. Rudimentary third ventricle may be present.
 - Septum pellucidum absent, callosal splenium may be formed, falx cerebri and interhemispheric fissure partially developed posteriorly.
 - Thalami and basal ganglia partially separated
 - Associated anomalies: Dorsal interhemispheric cyst, variable craniofacial anomalies

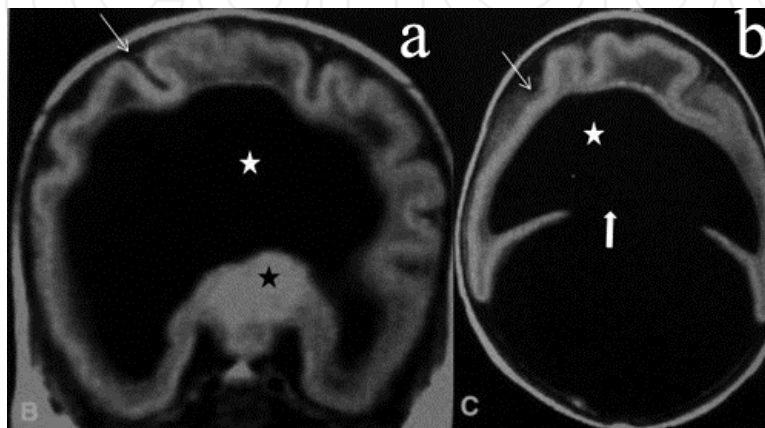


Fig. 14. Alobar holoprosencephaly. T1W coronal(a) and axial(b) MRI brain shows a large central monovertricle(white asterisk), thin cortical mantle(thin white arrow) and fused deep gray nuclei seen as a small central mass of gray matter(black asterisk). The crescentric monovertricle communicates with a large dorsal cyst (thick white arrow). (From Barcovich AJ. Pediatric Neuroimaging ((4th edn). Philadelphia: Lippincott Williams & Wilkins).

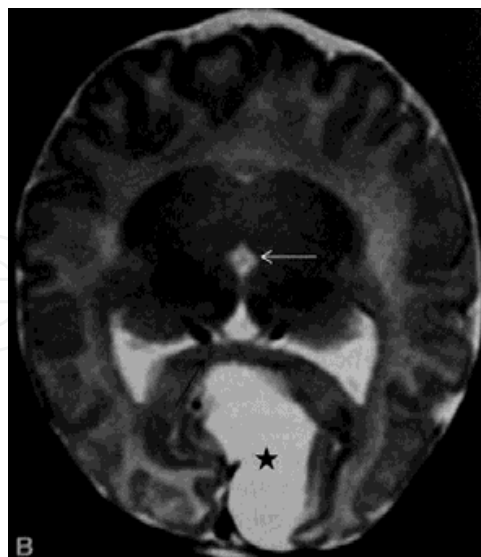


Fig. 15. Semilobar holoprosencephaly. Axial T2W MRI brain shows H-shaped monovertricle with rudimentary third ventricle (arrow). Basal ganglia and thalami are partially fused. Note the presence of callosal splenium(thin black arrow) and a small dorsal interhemispheric cyst(asterisk). (From Barcovich AJ. Pediatric Neuroimaging ((4th edn). Philadelphia: Lippincott Williams & Wilkins).



Fig. 16. Lobar holoprosencephaly. Axial T2W MRI brain shows separated basal ganglia and thalami, well formed third ventricle (small closed arrows) and splenium (large closed arrows), rudimentary frontal horns (small open arrows) and partial midline fusion of the frontal lobe cortex (thin large white arrow). The azygous anterior cerebral is also present (curved black arrow). (From Barcovich AJ. *Pediatric Neuroimaging* ((4th edn). Philadelphia: Lippincott Williams & Wilkins).

- Lobar Holoprosencephaly
 - Nearly complete brain cleavage
 - Squared-off or boxlike configuration of frontal horns due to absence of septum pellucidum
 - Septum pellucidum absent, variable degree of anterior extension of corpus callosum, well formed falx cerebri and interhemispheric fissure with some anteroinferior fusion of hemispheres
 - Thalami and basal ganglia well separated
 - Associated anomalies: Azygous anterior cerebral artery, mild or absent craniofacial anomalies

Septo-optic Dysplasia (de Morsier syndrome) ^{1,2,6}

- Milder form of lobar holoprosencephaly
- Characterized by hypoplastic optic nerves and optic chiasma, and absent septum pellucidum
- Associated hypoplasia of hypothalamic-pituitary axis seen in 2/3rd cases
- MR picture of small optic nerves and chiasma, widened anterior recess of 3rd ventricle and suprasellar cistern, and squared frontal horns
- Associated anomalies: Scizencephaly (most common)

4.3 Disorders of sulcation/cellular migration

Lissencephaly (Agyria-Pachygyria) ^{1,2} (Figure 17,18)

- Refers to “smooth brain” with absent or poor sulcation.
- Can be complete (agyria) or incomplete (pachygyria). Intermediate features of agyria and pachygyria may coexist

- Three types: Type I, II, and III
- Type I (classical) lissencephaly
 - Typical figure eight configuration of brain with oblique and shallow sylvian fissures.
 - Thickened cortex with flat broad gyri and smooth gray-white matter interface.
 - Colpocephaly
 - Associated with Miller-Dieker syndrome.

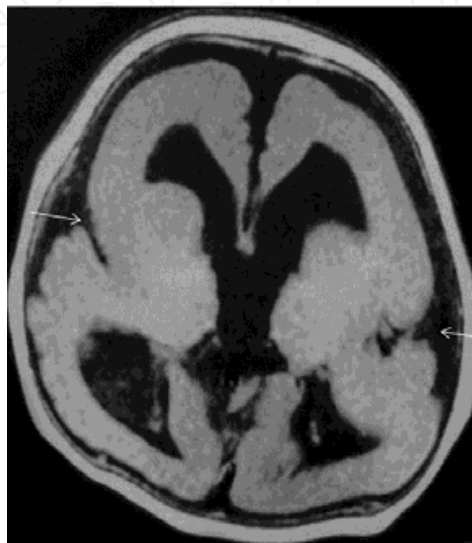


Fig. 17. Lissencephaly. Axial T1W image shows a complete smooth brain with thickened cortex and shallow sylvian fissures (arrows) giving the brain characteristic figure of eight appearance.

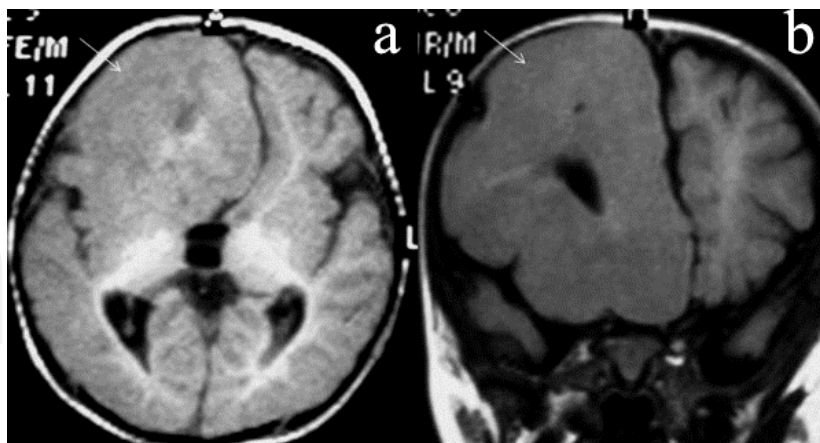


Fig. 18. Lissencephaly with hemimegalencephaly. Axial (a) and coronal (b) T1W image of brain shows right sided hemimegalencephaly and lissencephaly. Right frontal lobe is particularly enlarged, has a disorganized, thickened, nearly agyric cortex with complete loss of cortico-medullary differentiation (arrow). The anterior interhemispheric fissure is displaced to the opposite side by the hypertrophied frontal lobe; ipsilateral frontal horn is also enlarged.

- Type II (Cobblestone) lissencephaly

- Thickened cortex with polymicrogyric appearance. Concurrent hypomyelination of underlying white matter present.
- Associated with Fukuyama congenital muscular dystrophy, Walker-Warburg syndrome and muscle-eye-brain syndrome.
- Type III(cerebrocerebellar) lissencephaly
 - Microcephaly with moderately thickened cortex and hypoplastic cerebellum and brain stem

Nonlissencephalic Cortical Dysplasia^{1,2} (Figure 19,20,21)

- Two types: Polymicrogyria and pachygyria
- Polymicrogyria
 - characterized by diffusely thickened cortex with irregular, bumpy gyral pattern
 - MRI: thick cortex with flat surface, irregular gray-white matter junction

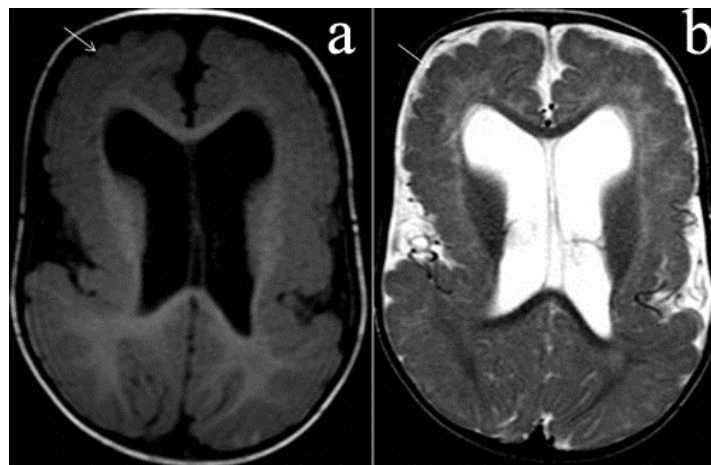


Fig. 19. Polymicrogyria. Axial T1W(a) and T2W(b) image shows shallow sylvian fissures giving figure of eight appearance to the brain. The cortex is diffusely thickened and has irregular bumpy gyral pattern (arrow) with relative paucity of underlying white matter. The cortico-medullary junction is also irregular.

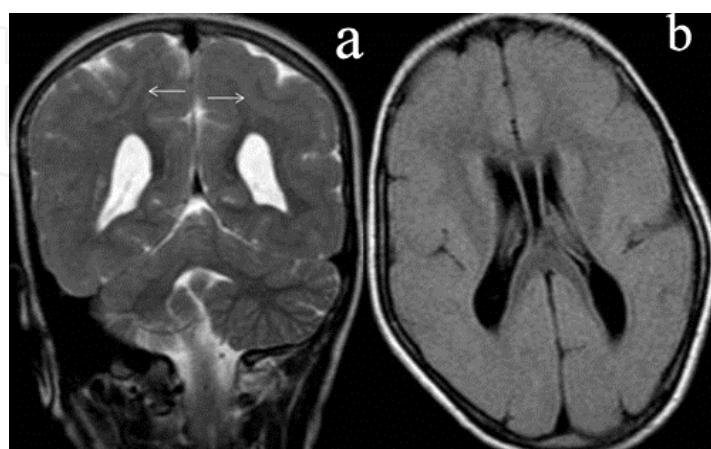


Fig. 20. Patchygyria with band heterotopia. Coronal T2W (a) and axial FLAIR (b) image shows very few, shallow sulci and thickened gyri. Also note band heterotopia, seen as bands of gray matter alternating with the white matter (arrow).

- Pachygyria
 - Focal areas of thickened and flattened cortex with blurred gray-white matter junction
- Both types of cortical dysplasia show relative paucity and marked T2 prolongation of underlying white matter

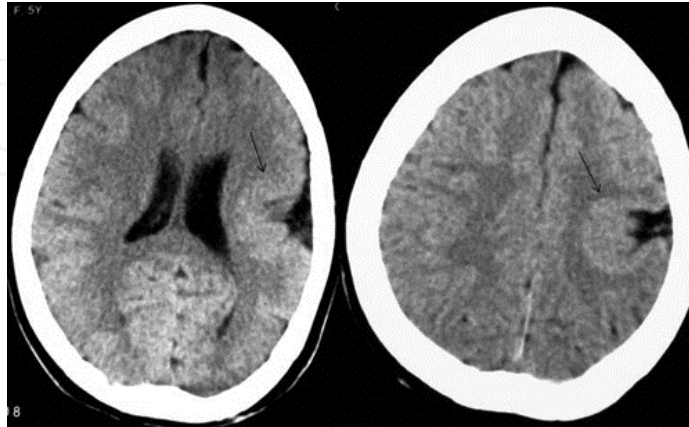


Fig. 21. Focal cortical dysplasia. Axial NCCT brain shows focal cortical dysplasia seen as localized thickening and infolding of the cortex (arrow) in left fronto-parietal junction region with paucity of underlying white matter and prominence of overlying sulcus.

Heterotopias^{1,2,7} (Figure 22,23)

- Characterized by the presence of normal neurons at abnormal sites
- Result of arrested neuronal migration from periventricular germinal zone to the cortex along the radial glial fibers
- Two types: Nodular type(common), band (laminar) type (uncommon)
- Nodular type:
 - Multiple masses of gray matter which are of variable size
 - Common location: subependymal or subcortical
 - Focal or diffuse
 - Subependymal focal nodules indent the ventricular wall, whereas diffuse heterotopias border the walls of the lateral ventricle and are X-linked.
 - Heterotopias are isointense to normal gray matter in all pulse sequences and do not enhance on administration of intravenous contrast. They are best appreciated on medium tau inversion recovery sequences.
 - The differential diagnosis is subependymal nodules (SENs) of tuberous sclerosis. On MRI, SENs are not precisely isointense to gray matter, however, occasionally show enhancement after contrast administration. They are often calcified.
 - Large dysplastic and disorganized masses of ectopic gray matter may mimic intracranial mass, and produce severe deformity of ipsilateral ventricle.
 - Subcortical heterotopias are less frequent.
- Band or laminar type
 - A layer of neurons interposed between the ventricle and cortex, seen as alternating layer of gray and white matter band
- The cortex overlying the heterotopia is nearly always abnormal with pachygyria or polymicrogyria.

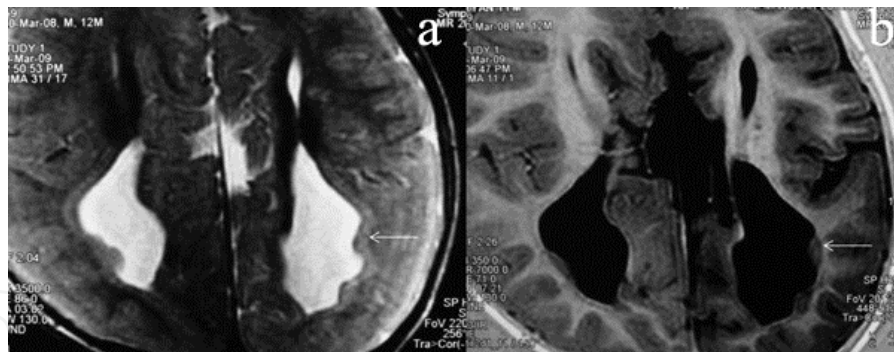


Fig. 22. Nodular heterotopia. Axial T2W(a) and T1W medium tau inversion recovery(b) image shows subependymal nodular heterotopias(arrow). Also note colpocephaly, widely separated and parallel lateral ventricles along with a small dorsal interhemispheric cyst in a patient of partial callosal agenesis.

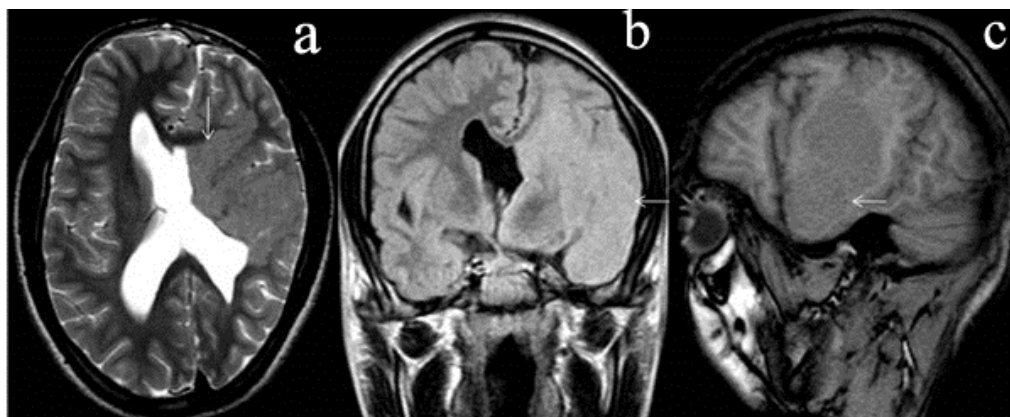


Fig. 23. Focal cortical dysplasia with mass heterotopia. Axial T2W (a) image demonstrates a heterotopic mass of gray matter indenting the left lateral ventricle(arrow). On FLAIR coronal(b) and T1W sagittal(c) image, the ipsilateral temporal lobe has a thickened, agyric cortex and shows complete loss of gray-white matter differentiation.

Schizencephaly (split brain) ^{1,2,8,9} (Figure 24,25)

- Characterized by presence of heterotopic gray matter lined cleft that extends from the ventricular (ependyma) to the periphery (pial surface) of the brain, traversing through the white matter.
- Can be unilateral or bilateral
- Two types: Closed lip (type I) or open lip (type II)
- Closed lip (type I) schizencephaly
 - Walls of the cleft oppose each other and there is no intervening CSF
 - Imaging shows an outpouching or nipping at the ependymal surface of the cleft
- Open lip (type II) schizencephaly
 - Walls of the cleft are widely separated and the cleft is occupied by CSF
 - Severe form of open lip schizencephaly has an appearance which is called “basket brain”.
 - Closest differential is porencephalic cyst in which CSF space is lined by gliotic white matter, in contrast to gray matter lined schizencephaly.

- Associated anomalies: heterotopias, septo-optic dysplasia, absence of septum pellucidum and callosal dysgenesis

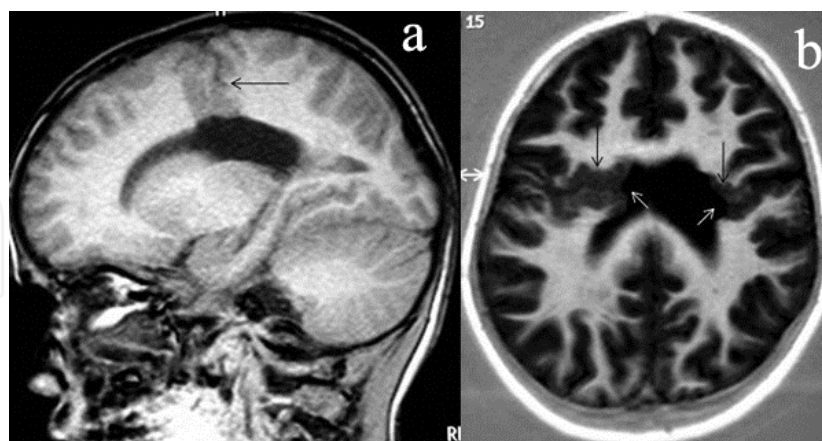


Fig. 24. Bilateral closed lip scizencephaly. Sagittal T1W(a) and axial T1W medium tau inversion recovery(b) image shows bilateral gray matter lined tract with closely opposed walls(thin black arrow), extending from the lateral ventricle to the cerebral convexity. Note nipping in the lateral wall of bilateral lateral ventricles (thin white arrow).



Fig. 25. Unilateral open lip scizencephaly. Axial NCCT brain shows a wide open, gray matter lined CSF cleft extending from left lateral ventricle to the cerebral cortex (arrow).

Hemimegalencephaly^{1,2,9,10} (Figure 18)

- Hamartomatous overgrowth of a part or all of one cerebral hemisphere
- MR shows enlargement of a part or whole of one cerebral hemisphere, ipsilateral ventricle is frequently dilated and the frontal horn is stretched. The cortex is affected by diffuse migration anomaly (polymicrogyria, pachygyria) and the underlying white matter is gliotic and dysmyelinated
- Rarely, associated enlargement and dysplasia of ipsilateral cerebellar hemisphere and brain stem may be present, a condition referred as total hemimegalencephaly
- Heterotopias may be present
- Associated anomalies: Epidermal nevus syndrome, Klippel-Trenaunay-Weber syndrome, Neurofibromatosis type 1

4.4 Disorders of cerebellar hypoplasia/dysplasia

Dandy-Walker Complex^{1,2,11} (Figure 26,27)

- Includes Dandy-Walker Malformation and Dandy-Walker Variant
- Dandy-Walker Malformation Characterized by
 - Large posterior fossa with cystic dilatation of the fourth ventricle; that elevates the tentorium, torcular Herophili and the transverse sinuses above the lamdoid suture (lamdoid torcula inversion)

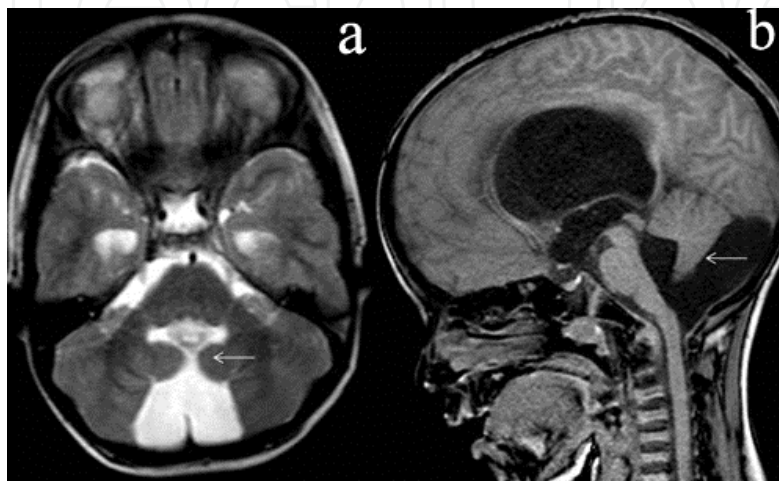


Fig. 26. Dandy-Walker Variant. Axial T2W (a) and sagittal T1W(b) image shows that inferior half of the vermis is hypoplastic (arrow) and the fourth ventricle communicates with enlarged cistern magna. Associated hydrocephalus is also present.

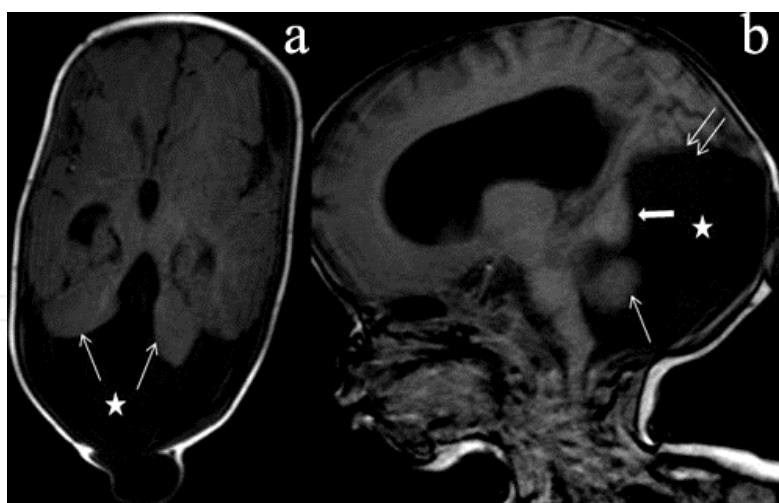


Fig. 27. Dandy-Walker Malformation. T1W axial(a) and sagittal (b) image of brain shows the classic findings of a Dandy-Walker malformation. The posterior fossa is enlarged and filled with a large cyst that is actually a huge fourth ventricle (asterix). The vermis is small, hypogenetic and is everted over the posterior fossa cyst (thick white arrow). The cerebellar hemispheres are hypoplastic (thin white arrow), and appear to be pushed laterally by the cyst on axial image and located beneath the everted vermis on sagittal image. The tentorium is elevated (double thin arrow). Note moderate hydrocephalus and small occipital encephalocele communicating with the posterior fossa cyst.

- Partial or complete vermian agenesis associated with hypoplastic cerebellar hemispheres. The hypoplastic superior vermis is everted above the cyst and the cerebellar hemispheres which are displaced anterolaterally against the petrous ridge appear “winged outward”.
 - Brain stem compressed against the clivus
 - The cistern magna, albeit present, is effaced
 - Hydrocephalus present in 80% of untreated cases
 - Associated anomalies: Corpus callosum agenesis (20-25%), heterotopias, schizencephaly, cephaloceles
- Closest differentials: Mega cisterna magna and posterior fossa arachnoid cyst where vermis is well developed.
 - Dandy-Walker Variant characterized by
 - Inferior vermian hypoplasia with communication of fourth ventricle and cistern magna through an enlarged vallecule giving characteristic “Key-hole deformity”.
 - The tentorial position and posterior fossa size are normal
 - Brain stem is usually normal and hydrocephalus is uncommon.

Mega Cistern Magna^{1,2} (Figure 28)

- Characterized by variable size dilatation of the cistern magna which, however, freely communicates with both the fourth ventricle and the adjacent subarachnoid spaces (can extend up to the straight sinus superiorly and C1-C2 level inferiorly). In severe cases enlargement of posterior fossa and scalloping of the occipital squamae may be present.
- The fourth ventricle, vermis and cerebellar hemispheres are normal. No hydrocephalus.
- Vermian hypoplasia may mimic mega cistern magna, hence, it is important to look at the size of the vermis for vermian hypoplasia which can be symptomatic.

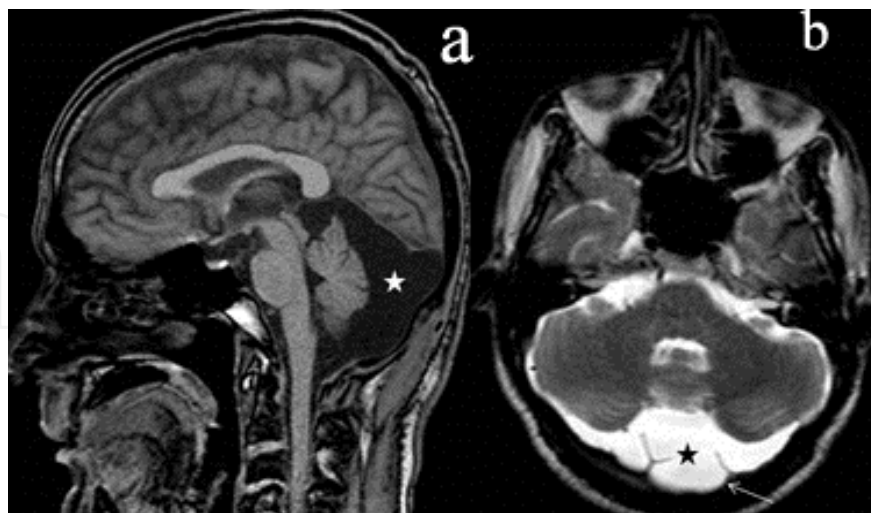


Fig. 28. Mega Cisterna Magna. Sagittal T1W(a) and axial T2W(b) image demonstrates an intact vermis with enlarged posterior fossa CSF space(asterix) that extends superiorly above the vermis and communicates with adjacent CSF space. Prominent scalloping of the occipital squamae is also seen (arrow). No hydrocephalus present.

Posterior Fossa Arachnoid Cyst^{1,2,12} (Figure 29)

- The posterior fossa arachnoid cyst is a CSF collection within the layers of arachnoid membrane which does not communicate fully with the fourth ventricle or adjacent subarachnoid spaces.
- The fourth ventricle, vermis and cerebellar hemispheres are normal but may be displaced by the cyst.
- On imaging, cyst appears as a non-enhancing round to oval lesion attenuation and signal intensity that closely parallels CSF.
- Differential diagnosis includes Dandy-Walker Malformation, Mega cisterna Magna.
- Note: In general the arachnoid cysts are more frequently supratentorial (75%), particularly at the temporal poles. Other relative common location includes the posterior fossa, the suprasellar and quadrigeminal cisterns.

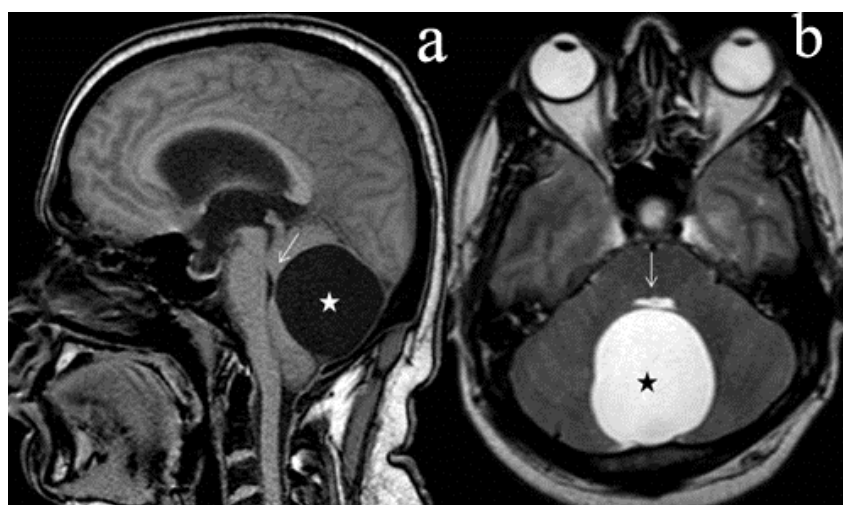


Fig. 29. Posterior fossa arachnoid cyst. Sagittal T1W(a) and axial T2W(b) image shows a classical posterior fossa arachnoid cyst(asterix). Note normally formed but displaced fourth ventricle (arrow) and vermis.

Chiari IV Malformation^{1,2}

- Severe cerebellar hypoplasia or dysplasia, small brain stem and large posterior fossa CSF space
- No hydrocephalus and other CNS anomalies

Joubert's Syndrome (Congenital Vermian Hypoplasia)^{1,2,13,14} (Figure 30)

- Characterized by inherited vermian dysgenesis, enlarged superior cerebellar peduncles and a high riding fourth ventricle.
- On MR imaging, the vermis is completely or partially absent. Superior fourth ventricle is bat-wing or umbrella shaped (on axial image) and has a convex roof (on sagittal image). The superior cerebellar peduncles are elongated, thin, running parallel to each other. Isthmus (area of pontomesencephalic junction) is narrow. Midbrain has the typical "molar tooth" appearance.
- Hydrocephalus is absent
- All patients of Joubert's syndrome should be screened for occipital encephalocele (30%), callosal dysgenesis, cortical dysplasia, hypothalamic hamartoma, and ocular, hepatic & renal diseases.

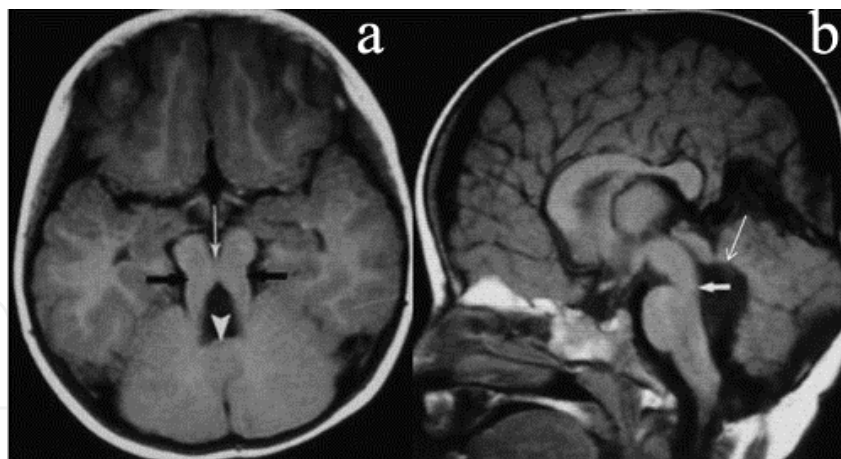


Fig. 30. Joubert's syndrome (Congenital vermian hypoplasia). Axial T1W image(a) at level of mid brain shows characteristic "molar tooth" appearance of the midbrain secondary to the narrow isthmus (thin white arrow) and elongated superior cerebellar peduncles (thick black arrow). Note severe vermian hypoplasia (arrow head). On sagittal T1W(b) image the fourth ventricle has a peculiar upward convex appearance (thin white arrow). Note narrow isthmus (thick white arrow).

Rhombencephalosynapsis^{1,2,15} (Figure 31)

- Rare abnormality characterized by vermian agenesis or hypogenesis combined with midline fusion of cerebellar hemispheres, peduncles and dentate nuclei.
- On MR imaging, the folia and sulci are continuous throughout the midline on axial section and monolobated cerebellum lies in mid-sagittal plane on sagittal image. The Posterior fossa is small.
- Associated anomalies: ventriculomegaly (common), callosal dysgenesis, absent septum pellucidum, cephalocele and schizencephaly.

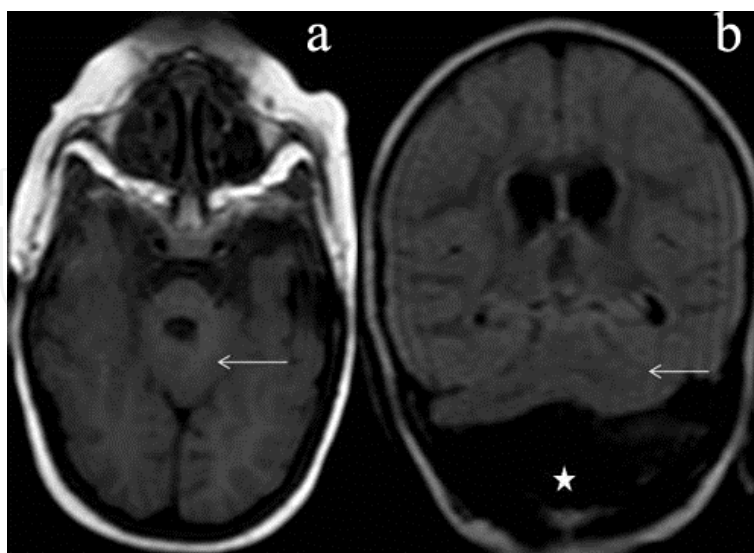


Fig. 31. Rhombencephalosynapsis. T1W axial(a) and FLAIR coronal(b) MR image demonstrates lack of normal vermis and midline fusion of the cerebellar hemispheres (thin white arrow). Also note markedly enlarged cisterna magna (asterix) indenting the posterior aspect of fused cerebellar hemispheres.

Tectocerebellar Dysraphism¹

- Rare abnormality characterized by vermian hypo-aplasia, occipital cephalocele, and marked deformation of quadrigeminal plate and the brain stem. Fusion of colliculi forms a tectal beak which points towards the site of the cephalocele. The cerebellar hemispheres usually engulf the brain stem.
- Associated anomalies: hydrocephalus and other supratentorial abnormalities.

Lhermitte-Duclos Disease^{1,2} (Figure 32)

- Also known as dysplastic gangliocytoma of the cerebellum
- Uncommon cerebellar dysplasia characterized by gross thickening of the cerebellar folias with or without mass effect.
- MR imaging demonstrates a pseudomass having laminated or folial pattern of increased signal on T2-weighted scans. The lesion may or may not enhance after administration of contrast. Mass effect and displacement of fourth ventricle may occur. Calcification or hydrocephalus may be present.

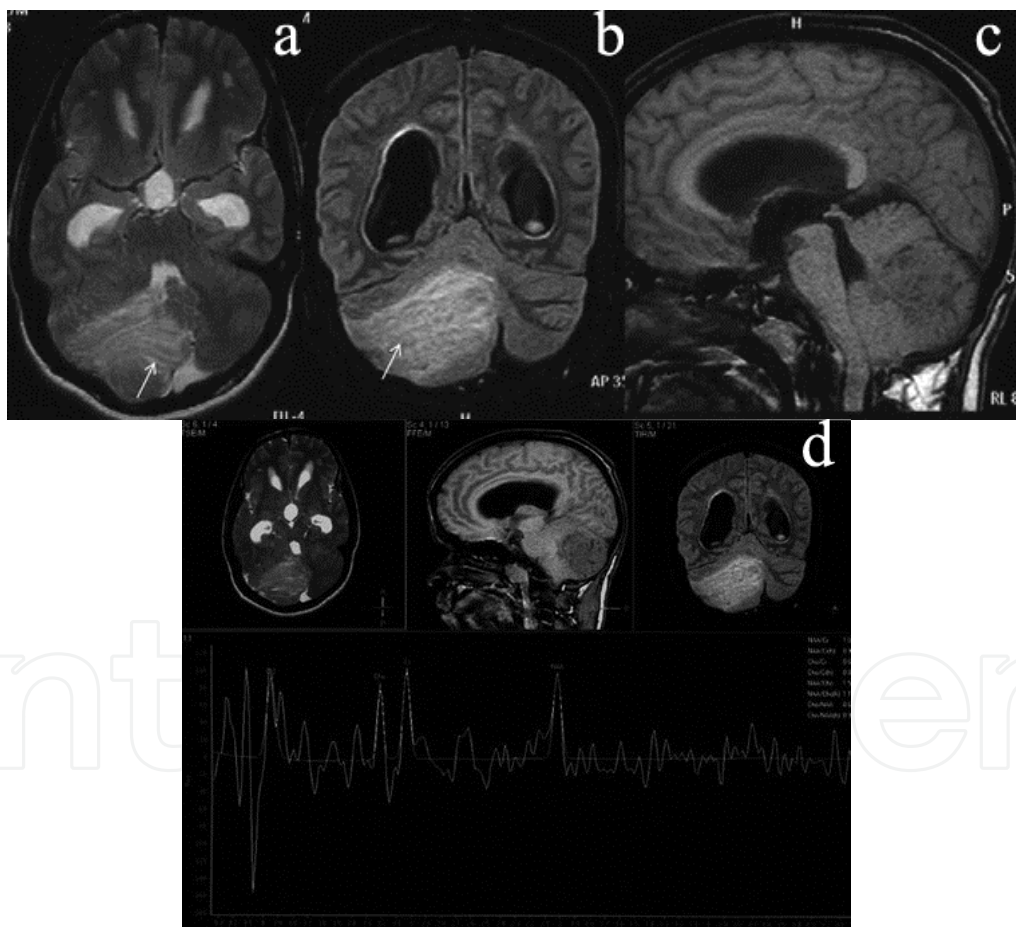


Fig. 32. Lhermitte-Duclos syndrome. Axial T2W (a), coronal FLAIR(b), and sagittal T1W(c) MR image demonstrates a large laminated-appearing T2W/FLAIR hyperintense and T1-weighted hypointense mass involving the right cerebellar hemisphere. Note gross thickening of the cerebellar folias (arrow). No perilesional edema present. However, mass effect on the fourth ventricle with moderate hydrocephalus can be seen. Proton MR spectroscopy (d) reveals normal metabolites peak.

- Associated anomalies: Cowden syndrome (common), megalencephaly, heterotopias, cortical dysplasia, multiple visceral hamartomas and neoplasms.

4.5 Disorders of histogenesis

“Neurocutaneous syndromes” or “Phakomatoses” constitute a group of congenital malformations which are characterized by cutaneous lesions associated with CNS anomalies. Some of the common neurocutaneous syndromes are described below.

Neurofibromatosis^{1,16,17} (Figure 33-42)

Neurofibromatosis type 1 (NF 1)

- Also known as Von Recklinghausen disease or peripheral neurofibromatosis
- Accounts for > 90% of all NF cases
- Incidence = 1:2000 to 3000 live births
- Diagnostic criteria: two or more of the following findings are present
 - Six or more café-au-lait spots (≥5mm in pre-pubertal children and ≥15mm in post-pubertal period)
 - One plexiform neurofibroma or two or more neurofibromas of any type
 - Two or more pigmented iris hamartomas (Lisch nodules)
 - Optic nerve glioma
 - Axillary or inguinal freckling
 - Osseous lesions such as dysplasia of greater wing of sphenoid, pseudoarthrosis
 - First degree relative with NF-1
- CNS lesions present in 15-20% cases. These include
 - Optic nerve glioma (most common CNS lesion), may extend to involve the optic chiasma, optic tract, optic radiation and the lateral geniculate bodies.
 - Nonoptic gliomas may involve the brain stem, tectum, and periaqueductal region.
 - Plexiform neurofibroma is a hallmark of NF-1. It is an unencapsulated neurofibroma along the path of major cutaneous nerve of the scalp and neck, which commonly involves the first (ophthalmic) division of trigeminal nerve. It is often associated with dysplasia of sphenoid bone and bony orbit.
 - Non-neoplastic hamartomatous lesions (80%) of basal ganglia and white matter. Majority of lesions show no mass effect or contrast enhancement. These lesions may increase in size or number in early childhood, diminishes with age and rarely observed into adulthood.
 - Other intracranial lesions include astrocytic proliferation of the retina, intracranial aneurysms, vascular ectasia and a progressive cerebral arterial occlusion disease akin to moyo-moya pattern.
 - Spinal lesions may include cord astrocytoma / hamartoma, dural ectasia and lateral/anterior intrathoracic meningoceles.
 - Skeletal dysplasias may include hypoplasia of sphenoid bone and bony orbit, kyphoscoliosis, scalloping of posterior aspect of the vertebral bodies

Neurofibromatosis type 2 (NF 2)

- Also known as central neurofibromatosis
- Incidence = 1:50,000 live births
- Cutaneous manifestations rare

- CNS lesions present in 100% cases. These include
 - Bilateral acoustic schwannomas, hallmark of NF-2
 - Schwannomas of other cranial nerves. Trigeminal nerve is next most frequently involved nerve, albeit, any cranial nerve may be affected (with the exception of the olfactory and optic nerves).
 - Meningiomas, often multiple
 - Choroid plexus calcification
 - Spinal lesions include cord ependymomas, meningiomas, or multilevel bulky schwannomas of exiting roots

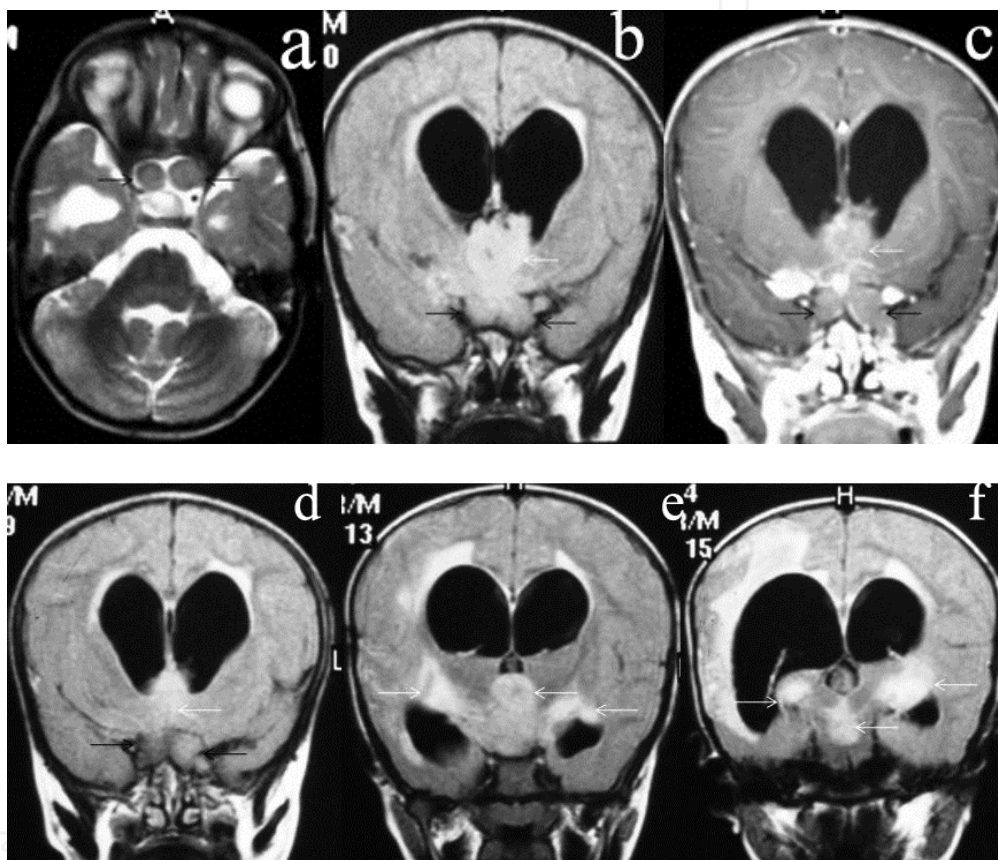


Fig. 33. Neurofibromatosis type 1: Opticochiasmatic-hypothalamic pilocytic astrocytomas. Axial T2-weighted (a) and coronal FLAIR (b) MR image shows enlargement of bilateral optic chiasma (thin black arrows) and ill-defined hyperintensity involving the hypothalamus (thin white arrow) and adjacent brain. Coronal T1-weighted post contrast image (c) demonstrates mild to moderate enhancement of the optic chiasma/hypothalamus but marked enhancement of the lesions involving the adjacent brain parenchyma. Moderate obstructive hydrocephalus is also present. FLAIR coronal images (d-f) of the same patient shows further extension of the optic pathway glioma to involve bilateral medial temporal lobes, basal ganglia region, mid brain and pons (thin white arrow). These lesions appear as ill-defined areas of high signal intensity on Flair images. The enlarged optic chiasma (thin black arrow) and obstructive hydrocephalus are also seen in these images.

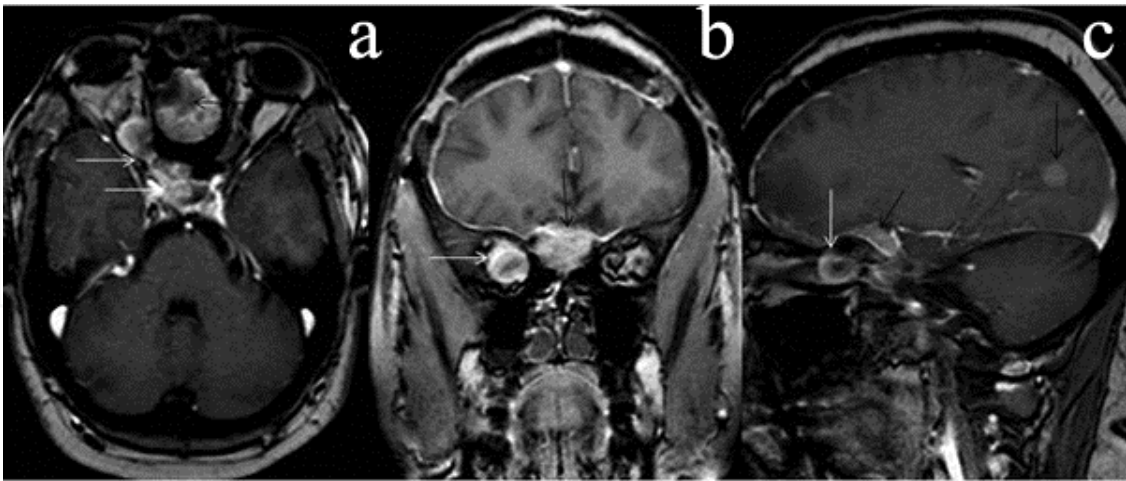


Fig. 34. Neurofibromatosis type 1: Optic nerve glioma with multiple meningiomas. Post contrast T1-weighted axial(a), coronal(b) and sagittal(c) MR images of brain reveal markedly enhancing right optic nerve glioma(thin white arrow) which extends posteriorly to involve the optic chiasma. Associated multiple enhancing meningiomas are also present (thin black arrow).

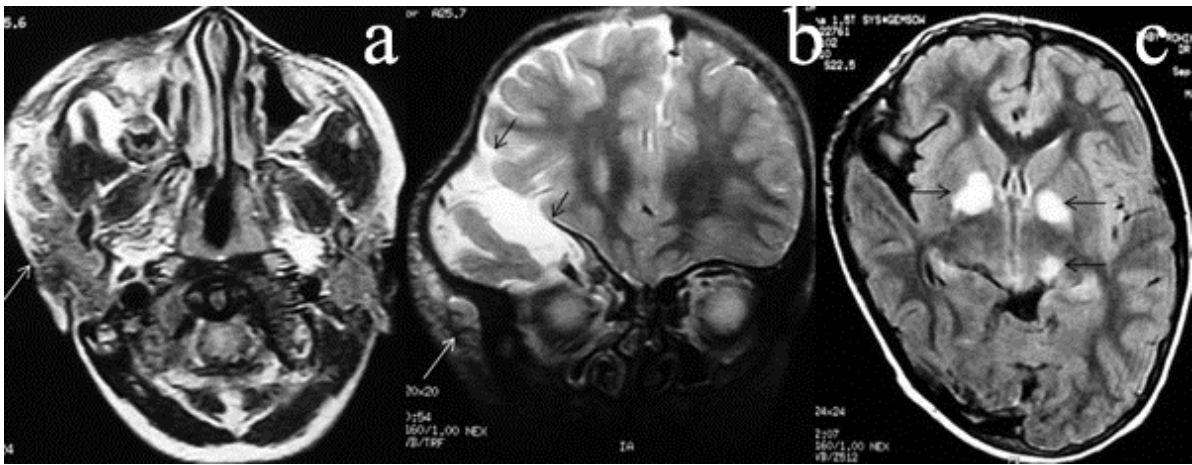


Fig. 35. Neurofibromatosis type 1: Plexiform neurofibroma with sphenoid wing dysplasia. T2W axial(a) and coronal(b) MRI brain shows right temporal and infratemporal plexiform neurofibroma(thin white arrows). Lateral part of the right sphenoid wing is absent and the dysplastic temporal lobe is seen protruding through the dehiscence sphenoid bone into the orbit(thin black arrow). FLAIR axial image(c) of the same patient shows multiple hyperintense nonneoplastic hamartomatous lesions involving bilateral basal ganglia and left midbrain region(arrows).

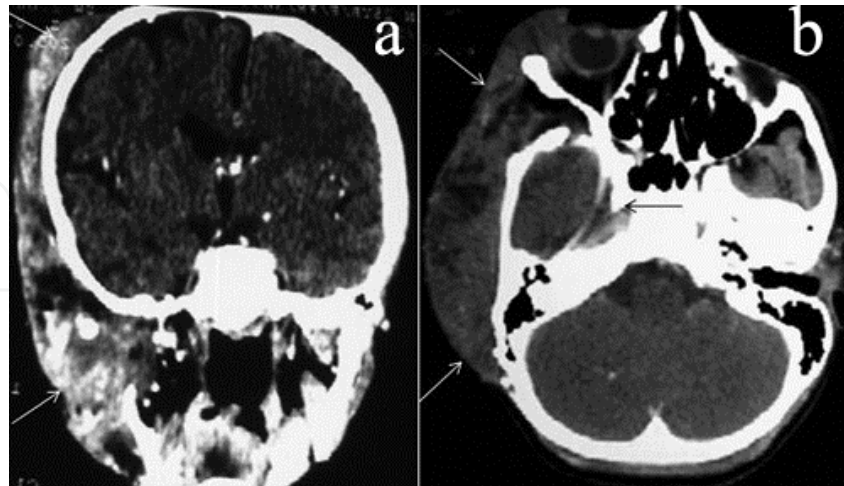


Fig. 36. Neurofibromatosis type 1: Plexiform neurofibroma with sphenoid wing dysplasia. Contrast enhanced CT brain, coronal (parenchymal window)(a) and axial (bone window)(b) view shows left sided plexiform neurofibroma of subcutaneous soft tissue(thin white arrow) and sphenoid wing dysplasia(thin black arrow).

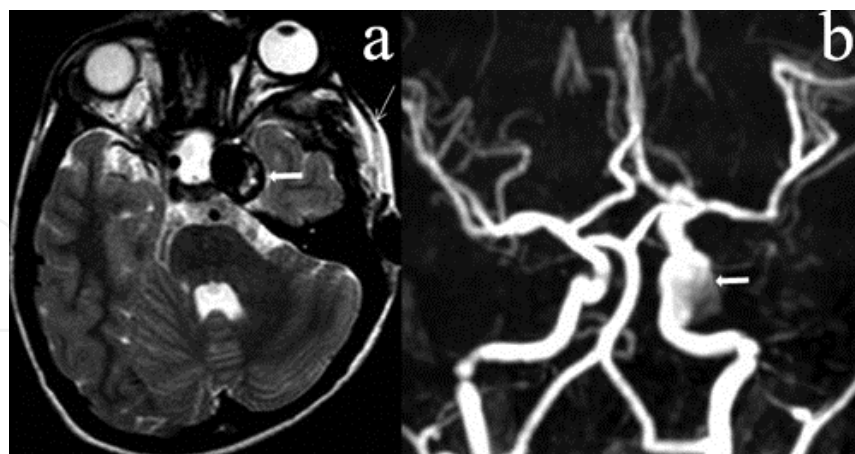


Fig. 37. Neurofibromatosis type 1: Vascular abnormalities. Axial T2W MR image of brain(a) shows severe left sphenoid wing dysplasia and subcutaneous soft tissue plexiform neurofibroma(thin white arrow). Associated left internal carotid artery aneurysm (thick white arrow) is seen on T2-weighted axial image(a) and TOF angiogram(b).

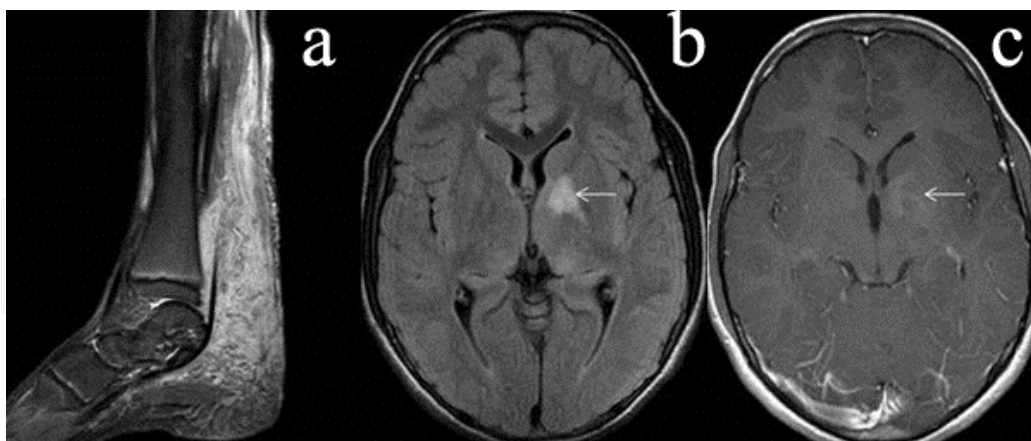


Fig. 38. Neurofibromatosis type 1: Peripheral Plexiform neurofibroma. Sagittal T1W post contrast image of lower limb (a) of another patient shows diffuse plexiform neurofibroma of left leg. Screening of brain revealed T2-hyperintense (b) focus in left basal ganglia region showing minimal contrast enhancement(c) but no mass effect, consistent with non-neoplastic hamartomatous lesion of brain. The patient had no neurological complains.

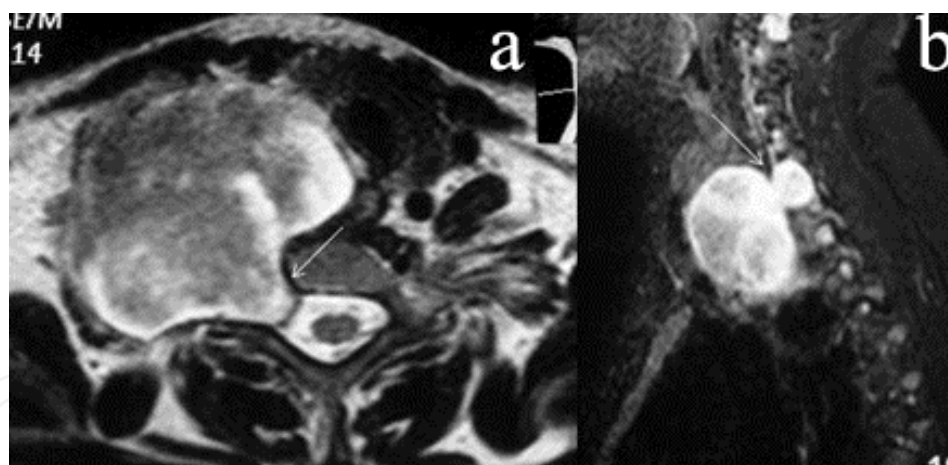


Fig. 39. Neurofibromatosis type 1: various spinal lesions. MRI of three different patients showing various spinal lesions found in NF-1. Axial T2W (a) and sagittal T1W post contrast (b) image of cervical spine demonstrates a large right paravertebral neurofibroma with intraspinal extension causing widening of ipsilateral neural foramen (thin white arrow). Sagittal T2W MRI of cervico-thoracic spine (c) shows multiple intradural extramedullary neurofibromas (thin white arrows). Solitary intramedullary hyperintense lesion (thin black arrow), which did not enhance after contrast administration (not shown) probably represents the benign white matter lesion similar to that observed in the brain. T2W-sagittal MRI spine (d) of another patient with NF-1 shows lumbar canal dural ectasia with posterior vertebral scalloping (arrows).



Fig. 40. Neurofibromatosis type 1: various spinal lesions. MRI of three different patients showing various spinal lesions found in NF-1. Axial T2W (a) and sagittal T1W post contrast (b) image of cervical spine demonstrates a large right paravertebral neurofibroma with intraspinal extension causing widening of ipsilateral neural foramen (thin white arrow). Sagittal T2W MRI of cervico-thoracic spine (c) shows multiple intradural extramedullary neurofibromas (thin white arrows). Solitary intramedullary hyperintense lesion (thin black arrow), which did not enhance after contrast administration (not shown) probably represents the benign white matter lesion similar to that observed in the brain. T2W-sagittal MRI spine (d) of another patient with NF-1 shows lumbar canal dural ectasia with posterior vertebral scalloping (arrows).

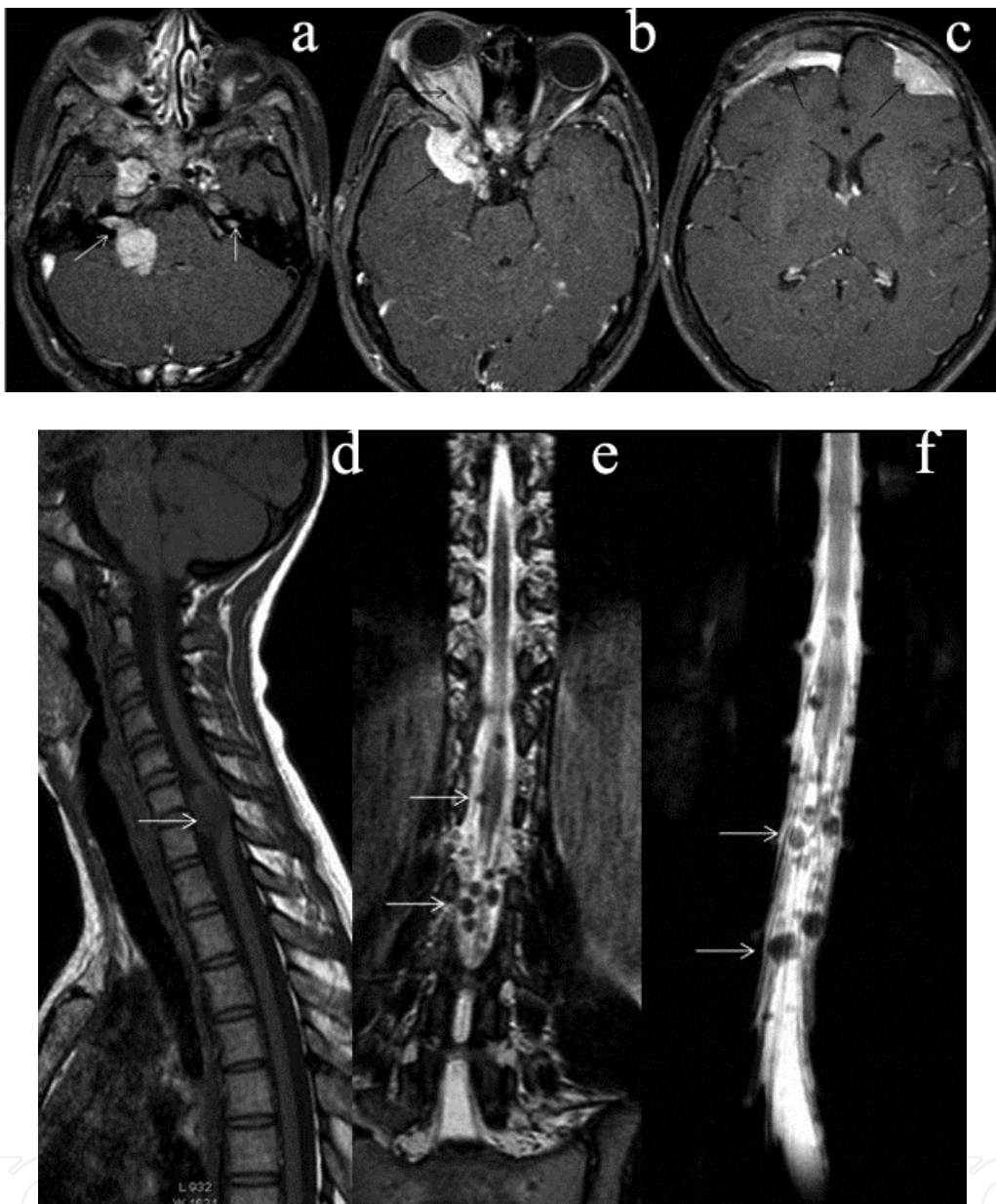


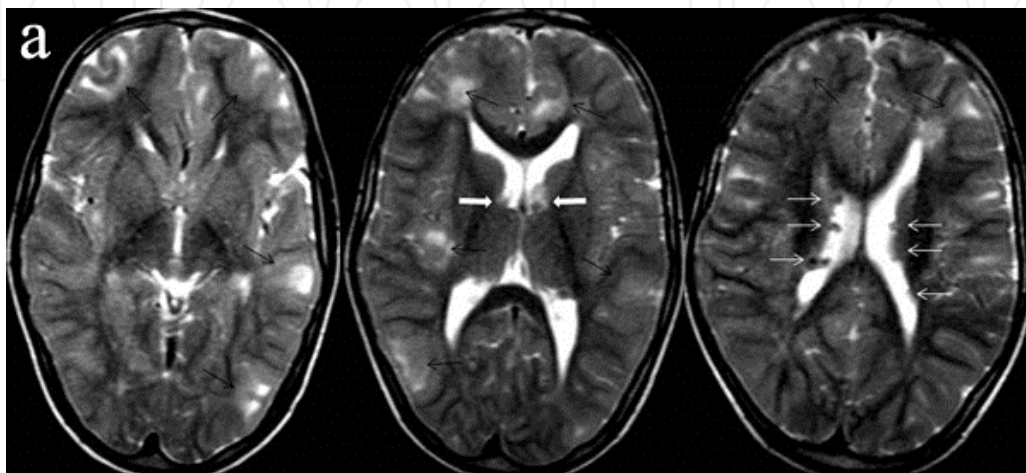
Fig. 41. Neurofibromatosis type 2: Axial T1W post contrast MR images(a-c) of brain demonstrate bilateral acoustic schwannomas(thin white arrows). Right schwannoma appears as a large homogenous enhancing right CP angle mass with intracanalicular extension and the left one is seen as a small intracanalicular enhancing mass. Multiple meningiomas(thin black arrows) are also present seen as enhancing extra-axial masses in right medial temporal and bilateral frontal regions. Right optic nerve meningeoma is also seen completely filling the intraconal space. Non-contrast sagittal T1W(d) and coronal T2W image(e) of whole spine of the same patient demonstrates low cervical region meningeoma(d) and multiple rounded lumbar region nerve root schwannomas(e), best appreciated on MR myelogram(f).



Fig. 42. Neurofibromatosis type 2: Nonneoplastic choroid plexus lesions. Axial NCCT brain of another patient with NF-2 shows extensive nonneoplastic bilateral choroid plexus calcification(arrows).

Tuberous sclerosis^{1,16,18} (Figure 43,44)

- Also known as Bourneville disease or multiple hamartomatous syndrome
- Incidence=1:10,000 to 50,000 live births
- Classical triad of popular facial lesions (adenoma sebaceum), seizure disorder and mental retardation
- CNS lesions include
 - Cortical tubers or hamartomas present in 95% cases of TS, are characteristic lesions at pathologic examination. On MR imaging, these lesions may expand and distort the affected gyri and show age-related signal changes. Enhancement following contrast administration occurs in less than 5% cases. Calcification of theses lesions increases with age.



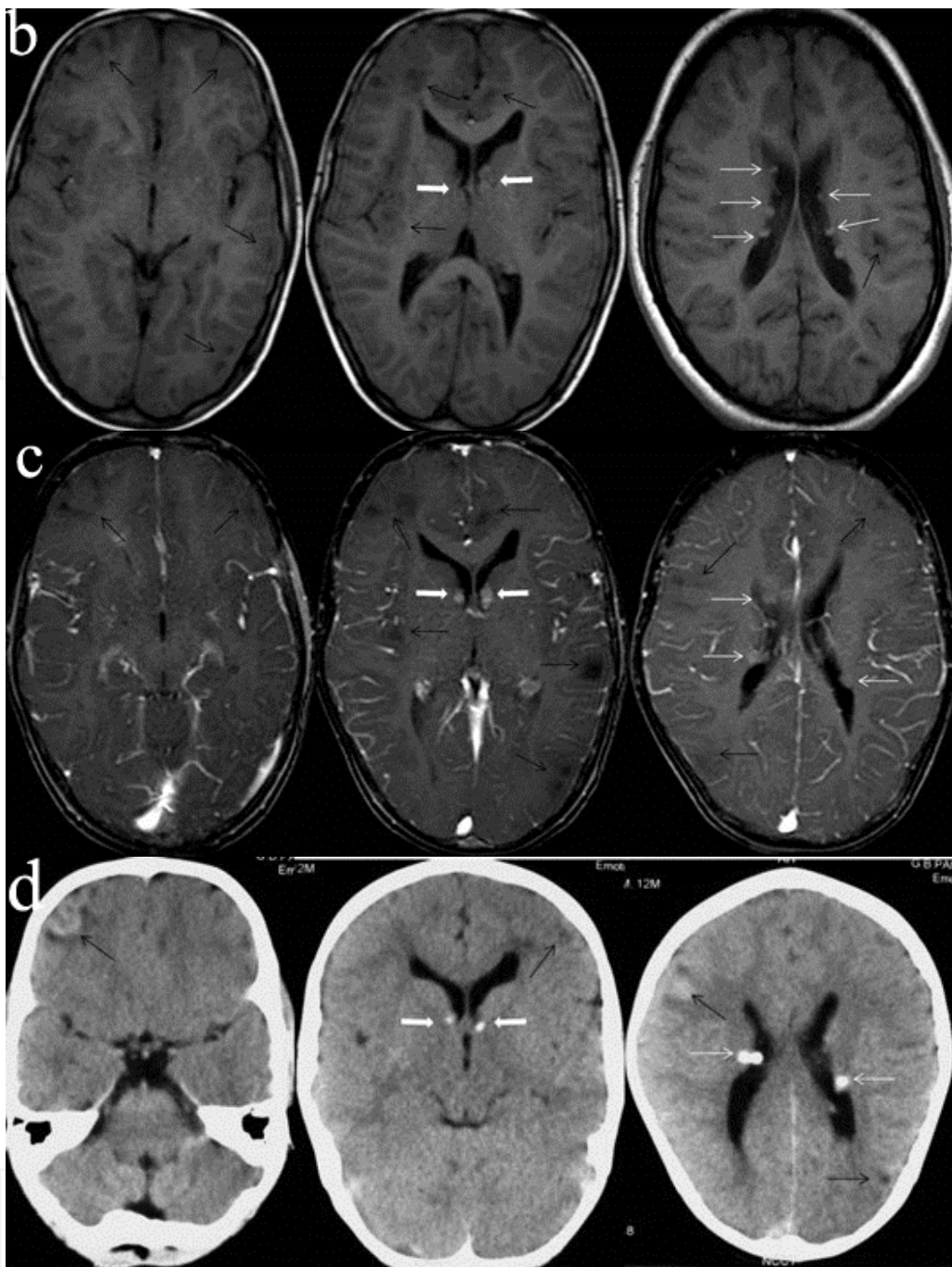


Fig. 43. Tuberosclerosis. Axial T2W MRI brain(a) demonstrates multiple hyperintense cortical tubers(thin black arrows), bilateral hyperintense giant cell tumors at the foramen of monro (thick white arrows) and multiple hypointense subependymal nodules(thin white arrows). On corresponding axial T1W images (b) the cortical tubers appear hypointense to gray matter; while the giant cell tumors and subependymal nodules appear isointense to the gray matter. Corresponding axial T1W post contrast images(c) demonstrate non-enhancement of the cortical tubers, mild enhancement of the subependymal nodules and marked enhancement of giant cell tumors. On corresponding axial NCCT(d) the cortical tubers are hypo to hyperdense in attenuation, while the giant cell tumors and subependymal nodules appear calcified.

- Subependymal nodules or hamartomas (SENs) present in 95% cases of TS, are the hallmark of tuberous sclerosis. They are most commonly located on the ventricular surface of caudate nucleus, just behind the foramen of Monro followed by atria and temporal horns of lateral ventricles, third and fourth ventricle. On MR imaging, these nodules are usually hypointense to white matter on T2-weighted sequences (because of calcification) and may show minimal contrast enhancement. Closest differential includes nodular heterotopias which parallel cortex in signal on all MR pulse sequences and do not enhance after administration of intravenous contrast.
 - Subependymal giant cell astrocytoma (SGCAs) present in 15% cases of TS, are located at or near the foramen Monro. On MR imaging, SGCAs show intense, uniform contrast enhancement. These lesions are frequently calcified. Obstructive hydrocephalus is common with SGCAs.
 - Dysplastic/disorganized benign white matter lesions. MR imaging shows four distinct patterns of these lesions: (i) straight or curvilinear bands that extends from the ventricle through the white matter to the cortex, (ii) wedge-shaped lesions, (iii) tumefactive or conglomerate foci, and (iv) cerebellar radial bands. Like cortical tubers, these lesions also show age-related signal changes, and contrast enhancement in about 15% cases.
 - Other CNS lesions include retinal hamartomas (50%), intracranial aneurysms, vascular ectasia and a progressive cerebral arterial occlusion disease akin to moyamoya pattern.
- Non-CNS lesions include hamartomatous growths in multiple organ system.

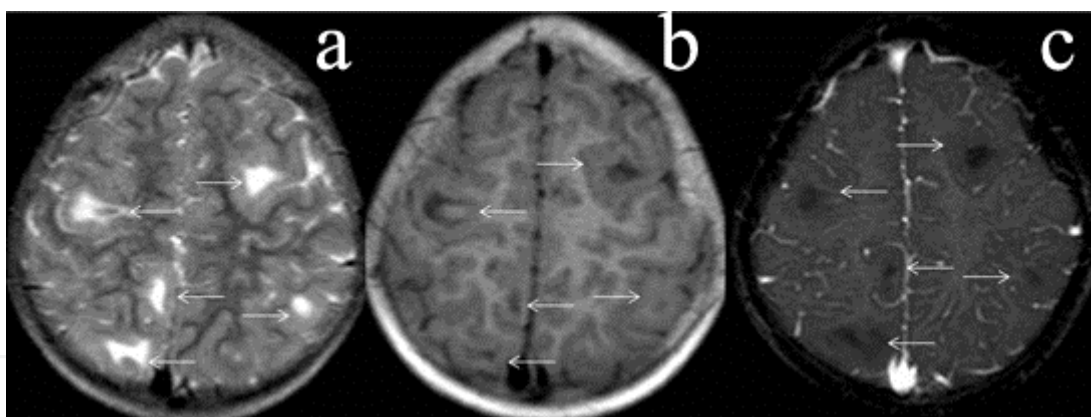


Fig. 44. Tuberous sclerosis. Axial T2W(a), T1W(b) and T1W post contrast image(c) of another patient demonstrates multiple cortical tubers appearing hyperintense on T2WI, hypointense on T1WI and showing no post contrast enhancement.

Sturge-Weber Syndrome^{1,16} (Figure 45,46)

- Also known as encephalotrigeminal angiomas
- Characterized by facial port wine vascular nevus flammeus in the trigeminal nerve distribution (1st division most commonly involved), leptomeningeal venous angiomas of ipsilateral brain, hemiparesis, homonymous hemianopia and seizure.
- Pathologically, there is leptomeningeal venous angiomas with congenital absence of cortical veins; therefore the blood is shunted towards the hypertrophied deep medullary veins and thence to the choroid plexus. This results in venous stasis and vascular congestion with hypoxia of the affected cortex. Slowly progressive atrophy

and dystrophic calcification of brain underlying the angioma occurs. The angioma itself does not calcify.

- Usually unilateral, rarely bilateral
- Occipital and posterior parietal lobe on the side of facial angioma is most commonly involved.
- Tram-track or gyriform pattern of cortical calcification underlying the leptomeningeal angioma is diagnostic of the syndrome. The calcification is unusual before two years of age. Calcifications are best seen on plain CT, T2W and GRE image.

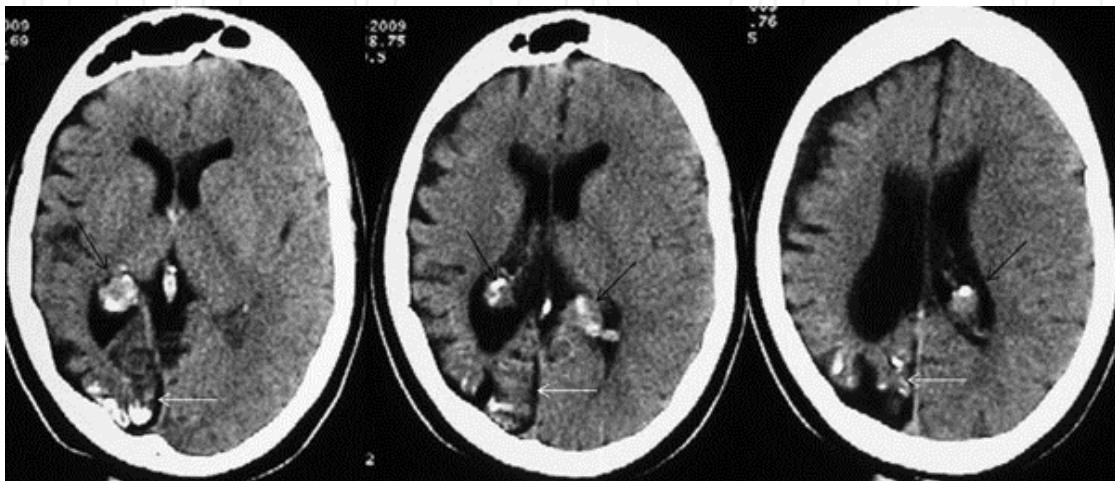


Fig. 45. Sturge-Weber syndrome. Unenhanced axial CT brain shows diffuse right cerebral hemispheric atrophy with dystrophic calcification (gyriform) of occipito-parietal cortex (thin white arrows). Associated enlargement of ipsilateral subarachnoid space, lateral ventricle and the frontal sinus is also present. Bilateral choroid plexus are enlarged (thin black arrows).

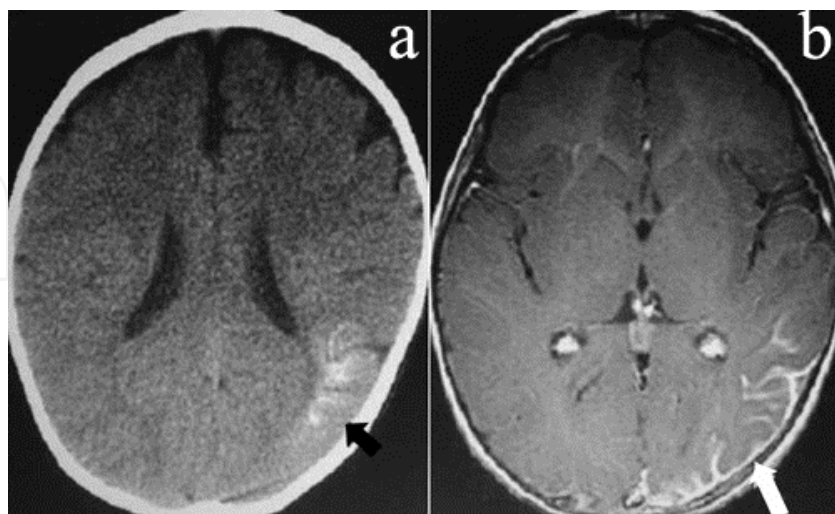


Fig. 46. Sturge-Weber syndrome. Unenhanced axial CT of brain (a) demonstrates gyriform calcification (thick black arrow) of left parieto-occipital cortex. After administration of contrast (b) there is marked enhancement of overlying leptomeningeal venous angiomas (thick white arrow).

- Severe cortical atrophy results in marked dilatation of ipsilateral ventricle, thickening of calvarium and prominence of ipsilateral sinuses.
- The pial angiomas, cortical infarcts and enlarged ipsilateral choroid plexus show intense post contrast enhancement. Abnormal medullary and subependymal veins are demonstrated on CEMR and MR angiogram.
- Ocular lesions in the form of sclera and choroidal angiomas may be seen on MRI in one-third cases.
- Associated anomalies: Klippel-Trenaunay syndrome

Von Hippel-Lindau syndrome^{1,16} (Figure 47)

- Is a multisystem disease characterized by presence of cysts, angiomas, and neoplasms of the CNS and abdominal viscera.
- CNS lesions include
 - Cerebellar (75%), retinal (50%) and spinal cord (25%) hemangioblastomas. Supratentorial hemangioblastomas are extremely rare.
 - On MR imaging, majority of hemangioblastomas have cystic appearance with intensely enhancing mural nodule. Between 20-40% tumors are solid. Contrast enhanced MRI has increased sensitivity for detection of small lesions. Flow voids in the afferent and efferent vessels supplying the tumor can often be detected. The angiographic appearance of the hemangioblastoma is highly characteristic, showing tangles of tightly packed vessels that become opacified in the early arterial phase.

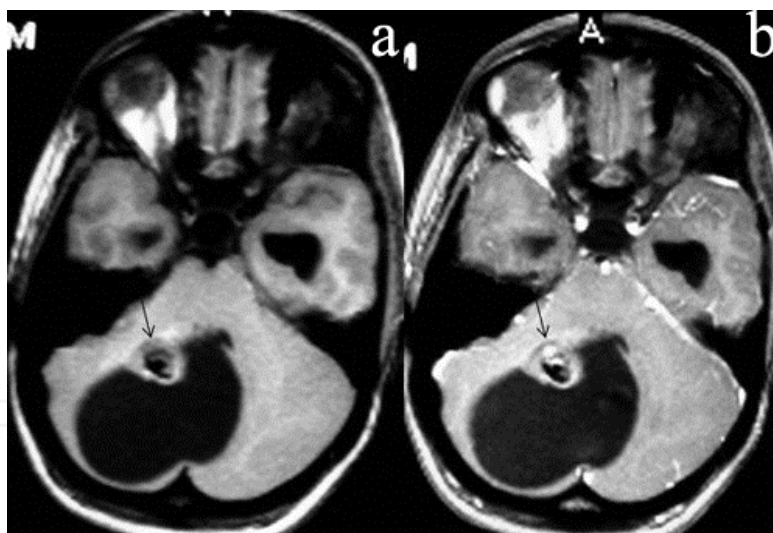


Fig. 47. Von Hippel-Lindau syndrome: Cerebellar hemangioblastoma. Axial T1W (a) MRI brain shows a right cerebellar hemispheric cyst with a mural nodule containing a curvilinear flow voids (arrow). Corresponding post contrast image (b) shows enhancing vessels within the mural nodule (arrow).

5. Summary

Congenital malformations of the brain are both complex and multiple. The neuroradiologic diagnosis of such anomalies requires a basic understanding of normal brain development and pathogenesis. The aetiologies associated with development anomalies may result from a

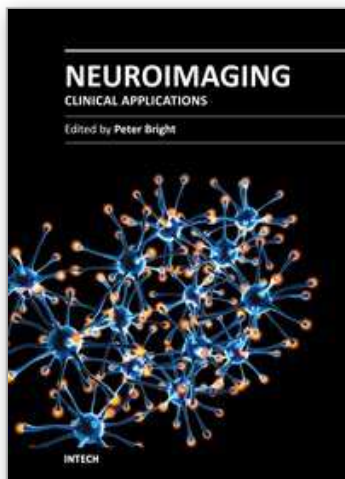
variety of insults from genetic to environmental. Abnormalities associated with the neural tube and the neural plate generally occur within the first 28 days of gestation. On the other hand, abnormalities associated with cellular proliferation and migration in the CNS generally occur after the 28th day of gestation. This chapter will cover malformations associated with both of these periods.

Congenital anomalies of the brain are commonly encountered in day to day practice. Nevertheless, diagnosing it correctly is of paramount importance. Imaging plays an important role in reaching the correct diagnosis necessary for optimum management of these unfortunate conditions. It is as important for every radiologist to be familiar with basic imaging findings of common congenital anomalies, as it is for the paediatrician.

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Modern neuroimaging tools allow unprecedented opportunities for understanding brain neuroanatomy and function in health and disease. Each available technique carries with it a particular balance of strengths and limitations, such that converging evidence based on multiple methods provides the most powerful approach for advancing our knowledge in the fields of clinical and cognitive neuroscience. The scope of this book is not to provide a comprehensive overview of methods and their clinical applications but to provide a "snapshot" of current approaches using well established and newly emerging techniques.

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