

Congenital brain abnormalities: Pictorial essay

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Abstract. Among all fetal anomalies, the central nervous system anomalies represent one of the most frequently involved structures with an estimated incidence of 1 per 100 births. It is mostly difficult to make an accurate diagnosis of congenital brain malformation, based on clinical findings; thus use of computed tomography (CT) or magnetic resonance imaging (MRI) is essential in these cases. The aim of this essay is to state the imaging findings of the main, most prominent congenital brain abnormalities and to present a practical classification of the entity.

Key words: Congenital abnormality, central nervous system, Imaging

1. Introduction

Among all fetal anomalies, the central nervous system anomalies (CNS) represent one of the most frequently involved structures with an estimated incidence of 1 per 100 births (1). Several studies have shown that malformations of cerebral cortical development are the cause of 23% to 26% of intractable epilepsies in children and young adults (2) and according to the Burdenko Institute of Neurosurgery of Russian Academy of Medical Sciences data, 22.6% children with hydrocephalus have additional brain malformations, and 11.8% have multiple malformations (3). This number emphasizes the point that cortical malformations must be ruled out in essentially every pediatric patient with developmental delay or epilepsy. It is very difficult to make a diagnosis of congenital brain malformation, based on clinical findings, and use of CT or MRI is essential in these cases.

The aim of this essay is to demonstrate a simplified classification of congenital anomalies affecting brain, and imaging findings of the common, prominent congenital anomalies.

2. Materials and Methods

2.1. Imaging Technique

A retrospective analysis of 16 congenital brain abnormalities through 152 cases, from November 2009 to May 2013, was conducted. The examinations were performed on a 1.5 T equipment (SIEMENS Medical Systems) with sequences SE T1WI (TR/TE, 468/8.7 ms), FSE T2WI (TR/TE, 4000/92 ms) with a flip-angle of 30 and a field of view of 230 mm.

2.2. Anatomy

The human nervous system consists of the central nervous system (CNS) and peripheral nervous system. The brain consists of soft, delicate, non-replaceable neural tissue. It is supported and protected by the surrounding skin, skull, meninges and cerebrospinal fluid (CSF). The skin and skull constitute a protective barrier against to physical damage of underlying tissues, invasion of hazardous chemical and bacterial substances. Three meninges are connective tissue membranes enclosing the brain and the spinal cord. Their functions are to protect the CNS and blood vessels, enclose the venous sinuses, retain the cerebrospinal fluid, and form partitions within the skull. The outermost meninx is the dura mater, which encloses the arachnoid mater and the innermost pia mater. CSF is a watery liquid similar in composition to blood plasma. It is formed in the choroid plexuses and circulates through the ventricles into the subarachnoid space, where it is returned to the dural venous sinuses and absorbed by the arachnoid villi. The main purpose of the CSF is to support and cushion of the brain and helping to nourish it.

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3. Results and Discussion

A number of classification systems have been proposed for congenital brain abnormalities, but none is universally accepted. We categorized the congenital malformations of brain into disorders of:

- Hindbrain malformations (Posterior fossa malformations and cysts)
- Hindbrain Herniations and Miscellaneous Malformations
- Malformations Of Cortical Development
- Disorders of Diverticulation and Cleavage

3.1. Hindbrain Cystic Malformations

3.1.1. Dandy-Walker Malformation

Classic Dandy-Walker malformation comprises complete or partial vermian agenesis, cystic dilatation of the fourth ventricle, and enlargement of the posterior fossa, with elevation of the transverse sinus, tentorium, and torcula (Fig. 1) (4). Nearly 70% of patients with DWM have associated supratentorial malformations, and up to 50% have extracranial anomalies.

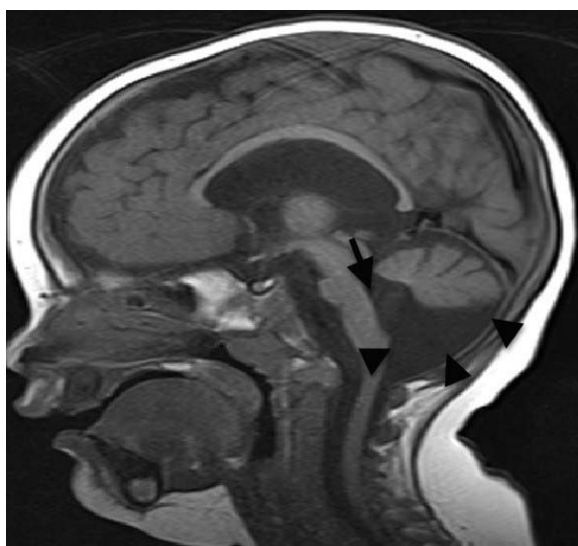


Fig. 1. Sagittal T1 weighted image. The floor of fourth ventricle (arrow) is normal, but dorsally fourth ventricle opens into a large CSF filled cyst (arrowheads). Vermis is not visualized.

3.1.2. Dandy-Walker Variant

The most common form of this anomaly demonstrates partial dysgenesis of the vermis (mild vermian hypoplasia) and remnant fourth ventricle that communicates with retrocerebellar cyst (Fig. 2). Posterior fossa is of normal size (5).

3.1.3. Mega Cisterna Magna

In this variant, fourth ventricle, vermis, and cerebellar hemispheres are normal. A large cisterna magna is present and may extend above

the vermis to the straight sinus (Fig. 3). Occasionally scalloping of occipital bone is seen (6,7).

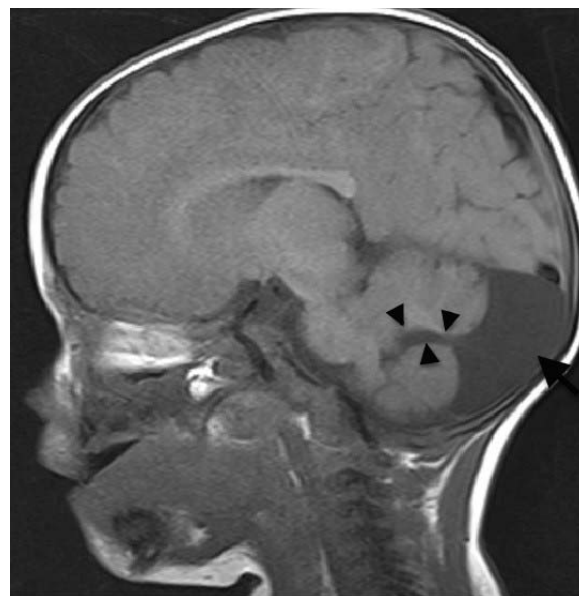


Fig. 2. Sagittal T1 weighted image. There is communication (arrowheads) between fourth ventricle and cisterna magna through enlarged vallicula, with a posterior fossa cyst (arrow).



Fig. 3. Axial T2 weighted image showing mega cisterna magna (arrows). Fourth ventricle, vermis and cerebellar hemispheres are normal.

3.1.4. Arachnoid Cyst

An arachnoid cyst is a congenital extracerebral mass that contains cerebrospinal fluid (CSF) encircled by walls composed of arachnoid membrane. Internal and external walls of the cyst consist of thin layers of arachnoid cells and join

unchanged arachnoid membrane at the margins. CT and MRI are the main diagnostic tools for arachnoid cysts. They reveal straightforward homogeneous masses isodense and isointense with CSF in all sequences and they do not show contrast enhancement and perifocal edema (Fig. 4). Arachnoid cysts of the posterior fossa may originate from the cisterna magna, in which case they are called inferior retrocerebellar cysts. Retrocerebellar cysts, pervading posterior fossa structures, cause compression of CSF pathways and markedly dilate the ventricular system (3).



Fig. 4. Axial T2 weighted image shows extraaxial CSF-like arachnoid cyst in posterior fossa with displacement of structures beside (arrow).

3. 2. Hindbrain Herniations and Miscellaneous Malformations

3.2.1. Chiari I

The Chiari malformations are a group of disorders involving mainly the posterior fossa. Chiari type I, a hindbrain dysgenesis, is caudal displacement of the cerebellar tonsils via the foramen magnum into the vertebral channel (Fig. 5), and it is the most common cerebellar abnormality identified on MRI (3,5). Tonsils that are 3–6mm below the foramen magnum are indeterminate and their significance needs to be correlated with a clinical symptomatology. Tonsils more than 6 mm below the foramen magnum are considered definitely abnormal and compatible with Chiari type I malformation. This malformation is more frequently isolated, but

concomitants hydrocephalus and hydromyelia, craniocervical dysgenesis, platibasia, basilar impression, occipitalisation of the atlas and Klippel-Feil malformation are not infrequent in these patients (3).

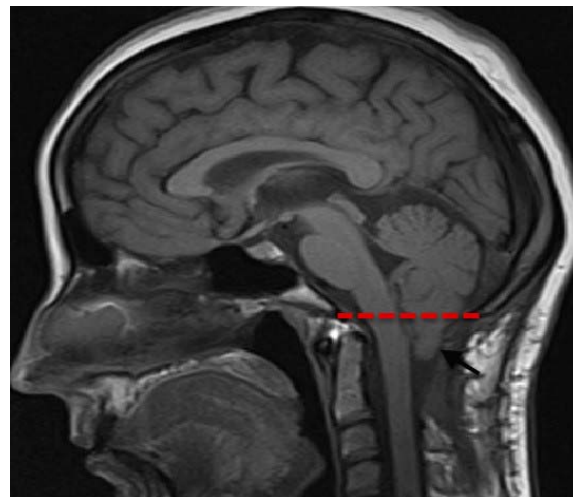


Fig. 5. Sagittal T1 weighted MR image showing caudal protrusion of the cerebellar tonsils (arrow) below the foramen magnum (spaced lines).

3.2.2. Chiari II

Chiari type II is a complex of malformations of the hindbrain, vertebral column and supratentorial structures. Almost all patients with Chiari type II have congenital myelomeningocele (Fig. 6 A, B) (3). Although this spinal dysraphism may involve any part of the canal, it is most common in the lumbar region (5). The cerebellum is indented superiorly by the tentorium and inferiorly by the foramen magnum. The pons and fourth ventricle are stretched inferiorly and narrowed in their anterior-posterior diameters. The medulla is also stretched inferiorly and extends below the foramen magnum (2). The upper medulla kinks at the attachment with a dentate ligament; this is called a “cervico-medullary kink”. The dorsal midbrain in these patients demonstrates fusion of the superior and inferior colliculi making the tectum appear beaked. The corpus callosum may be hypoplastic or absent. In these cases the white matter of the forceps major is deficient resulting in dilatation of the atria and occipital horns of the lateral ventricles (colpocephaly) (5).

3.2.3. Chiari III

The Chiari III malformation is a rare anomaly. Affected patients have an occipital or high cervical encephalocele (Fig. 7) in association with the classic findings seen in Chiari II malformations (8).



Fig. 6. Sagittal T1 weighted image (A) shows the inferiorly stretched pons and fourth ventricle (arrows), the beaked tectum (arrowhead), enlarged massa intermedia (M), a small posterior fossa (long arrows) and enlarged lateral ventricles (L). Axial T2 weighted image (B) shows spinal myelomeningocele (arrows).

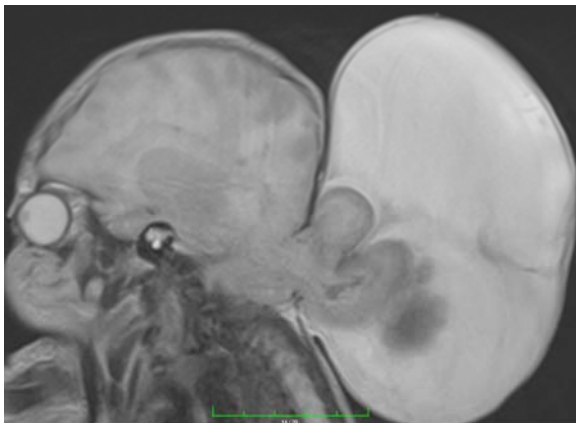


Fig. 7. Sagittal T2 weighted image. Herniation of posterior fossa contents through occipital cephalocele is seen with Chiari II malformation.

3.2.4. Corpus Callosum Agenesis

Corpus callosum forms from anterior to posterior except for the rostrum, which is formed last. Primary agenesis occurs in the 12th week of fetal life as a consequence of vascular or inflammatory damage of commissural lamina and may be isolated or concomitant with other malformations (3). Callosal agenesis may be complete or partial. In partial agenesis, splenium and rostrum are the missing parts. In complete agenesis the entire corpus callosum and the cingulate sulcus and gyrus are absent (6). Complete callosal agenesis shows high riding third ventricle with spoke-like orientation of gyri

around it. Lateral ventricles are widely separated, parallel and non-converging. Colpocephaly is commonly present. Lateral ventricles are indented superomedially by the longitudinal white matter bundles (Probst bundles) (Fig. 8 A, B) (6).

3.2.5. Joubert Syndrome

Joubert's syndrome is a rare condition representing one of the developmental defects of the cerebellar vermis associated with episodic hyperpnea and apnea, abnormal eye movements and mental retardation (5). Fourth ventricle is enlarged and has a typical "bat-wing" or "umbrella" appearance. Pontomesencephalic junction is narrow. Midbrain has the typical "molar tooth" appearance (Fig. 9 A, B) (6). A cleft is seen between the two cerebellar hemispheres (5). Hydrocephalus is not a feature of this syndrome.

3.2.6. Cerebellar Cortical Dysplasia

Focal cerebellar cortical disorganization or dysplasias are often observed in healthy newborns. The mechanisms of such dysplasias as well as their influence in appropriate positioning of components of cerebellar cortex, have been previously investigated, but are not clearly understood. Cerebellar cortical dysplasia may be associated with widespread cerebral malformations and cases with isolated cerebellar abnormalities are rare. The most relevant MR appearance of cerebellar cortex includes: defective, large, or vertical fissures; irregular

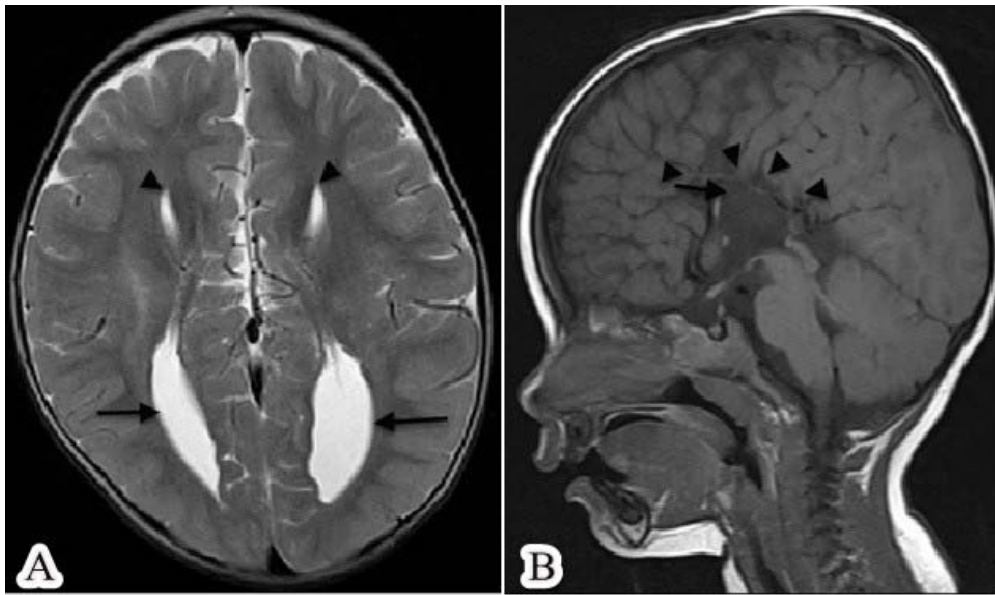


Fig. 8. Axial T2 weighted image (A) shows widely separated, parallel lateral ventricles with colpocephaly (arrows), and small and pointed frontal horns (arrowheads). Sagittal T1 weighted image (B) shows agenesis of corpus callosum and the cingulate sulcus and gyrus, high riding third ventricle (arrow) with spoke-like orientation of gyri around it (arrowheads).

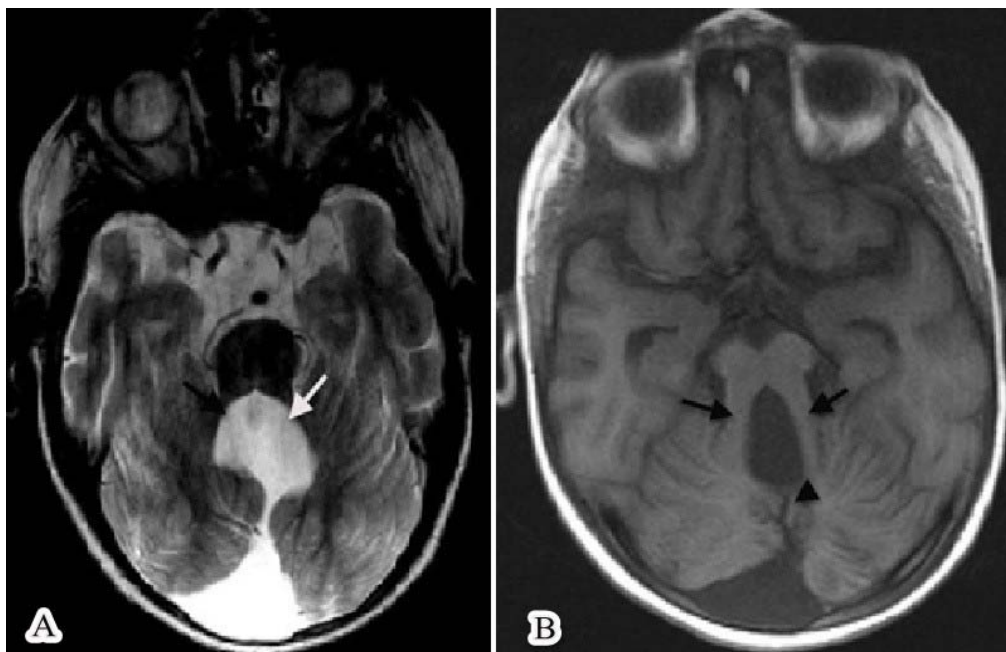


Fig. 9. Axial T2 weighted image (A) shows enlarged fourth ventricle and its typical "umbrella" appearance (arrows). Axial T1 weighted image (B) shows the typical "molar tooth" appearance of midbrain (arrows) and small vermis (arrowhead).

gray/white matter junction; lack of normal arborization of the white matter; and heterotopia within cerebellar hemispheres (Fig. 10) (9).

3.2.7. Unilateral Cerebellar Hypoplasia

Unilateral cerebellar hypoplasia is a relatively rare malformation. On the basis of MRI findings, cerebellar malformations can be divided into

those associated with hypoplasia and those with dysplasia; each type can show either focal or diffuse malformations (Fig. 11). Focal cerebellar hypoplasia can be further subdivided into isolated vermis hypoplasia or hypoplasia of one cerebellar hemisphere. Unilateral cerebellar hypoplasia may be an incidental finding in a patient with no

previous evidence of neuromuscular or metabolic disease and no past history of trauma or anoxia. However, cases with unilateral cerebellar hypoplasia can present with severe grand mal seizures, persistent headache, or with psychomotor retardation without cerebellar symptomatology (10).

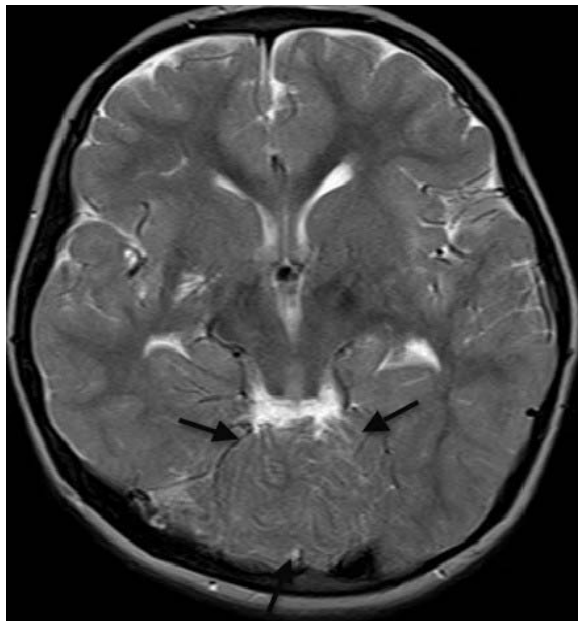


Fig. 10. Axial T2 weighted image shows cerebellar cortical disorganization; irregular gray/white matter junction, lack of normal arborization of the white matter.

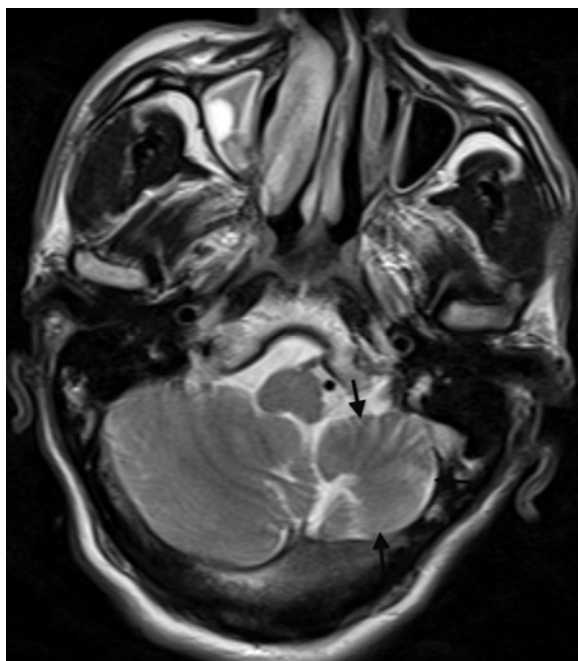


Fig. 11. Axial T2 weighted image shows hypoplasia of the left cerebellar hemisphere with asymmetry of the posterior fossa (arrows).

3.3. Malformations of Cortical Development

3.3.1. Schizencephaly

Schizencephaly is an uncommon congenital disorder of cerebral cortical development, defined as a gray matter-lined cleft extending from the pial surface to the ventricle (Fig. 12). The spectrum of cleavage ranges from a thread of CSF connecting the subarachnoid space to the ventricle, which is encircled by abnormal gray matter (Type I, closed-lip schizencephaly), to a wide communication between the subarachnoid space and the ventricle with edges of abnormal cortex (type II, open-lip schizencephaly) (11).

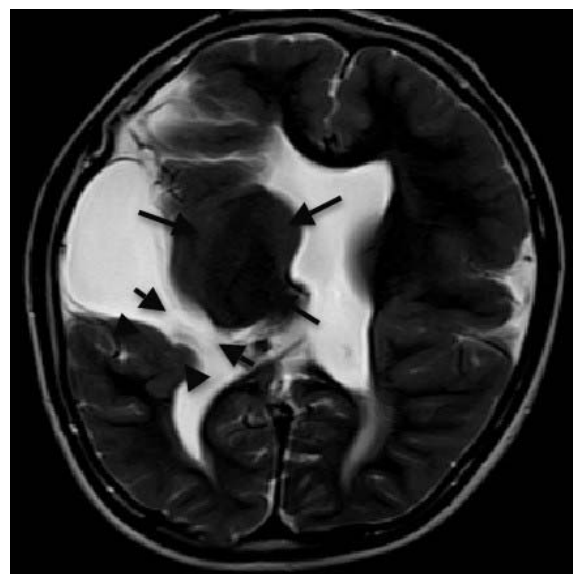


Fig. 12. Axial T2 weighted image shows a large cleft in continuity with lateral ventricle (short arrows). Continuity of gray matter through cleft (arrowheads) and the focus of cortical dysplasia are clearly shown (long arrows).

3.3.2. Lissencephaly

Lissencephaly belongs to the neuronal migrational disorder group. The syndrome is characterized by no or few cerebral gyri and sulci. Lissencephaly can be divided into two main groups such as type I (classic form) and type II (5). Type I lissencephaly shows the typical figure eight configuration of brain with colpocephaly, thickened cortex, smooth gray-white matter interface, flat broad gyri and shallow sylvian fissures (Fig. 13). Type II lissencephaly on MR shows thickened cortex having polymicrogyric appearance (6).

3.3.3. Cortical Heterotopia and Dysplasia

Heterotopic gray matter is caused by arrested migration of neurons from periventricular germinal zone to cortex. It can be inherited or acquired. It can be band-like or nodular. Nodular

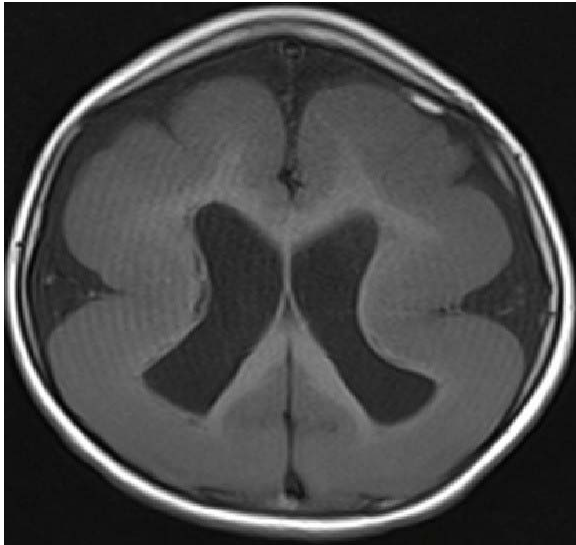


Fig. 13. Axial T1 weighted image shows a smooth outer cortical layer with near absent gyri, taking on a figure-8 appearance. Cerebral white matter is underdeveloped and the lateral ventricles are dilated.

heterotopias can be focal or diffuse. Subependymal heterotopias are the most common, seen as nodules indenting the ventricles. Heterotopic gray matter is isodense with gray matter on CT and isointense with gray matter on MRI (Fig. 14A) (6).

Focal cortical dysplasia is the name given to a malformation in which the cortex has abnormal lamination, and abnormal cells are seen in the

cerebral cortex and underlying white matter, often extending all the way to the wall of the lateral ventricle (Fig. 14B) (12). Neuroimaging studies are quite characteristic. When the involved region of brain is large, the conical gyral pattern will be abnormal, with broad gyri and large, irregular sulci (13).

3.4. Disorders of Diverticulation and Cleavage

3.4.1. Holoprosencephaly

It is a spectrum of congenital structural forebrain anomalies and is the most common malformation involving face and brain together. Its hallmark is monoventricle with non-cleaved frontal lobes. Also there is non-cleavage of diencephalon, and at times basal ganglia and thalami. Sylvian fissures are displaced anteriorly, resulting in increased sylvian angle (6).

It is a spectrum with three forms, alobar, semilobar and lobar. Alobar holoprosencephaly is the most severe form. Affected fetuses are often spontaneously aborted. On imaging, the brain is small and the hemispheres are completely fused as one entity. The basal ganglia and thalami are also fused. The ventricular system is seen as a single cavity and is in continuity with a large dorsal cyst (Fig. 15) (5).

In semilobar holoprosencephaly facial anomalies are variable, rudimentary occipital horns of lateral ventricles are present and falx is partially present (14).

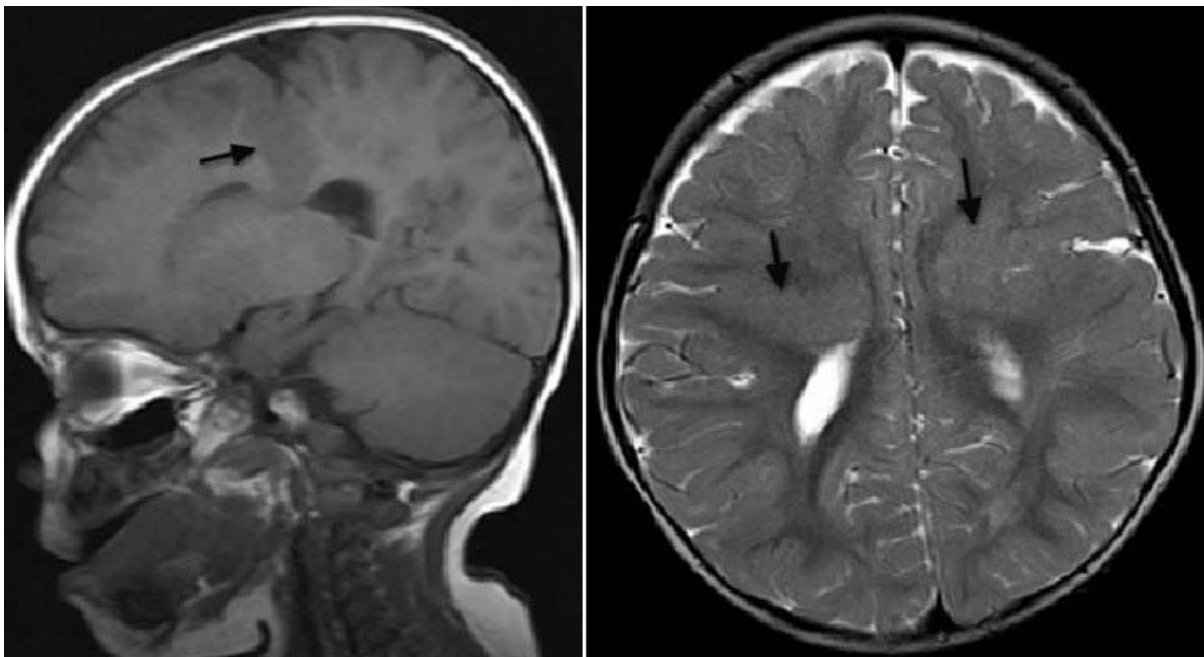


Fig. 14. Sagittal T1-weighted image (A) shows heterotopic gray matter (arrow). Axial T2 weighted image (B) through the parietal region shows heterotopic gray matter on right side and dysplastic cortex on left side, respectively (arrows).

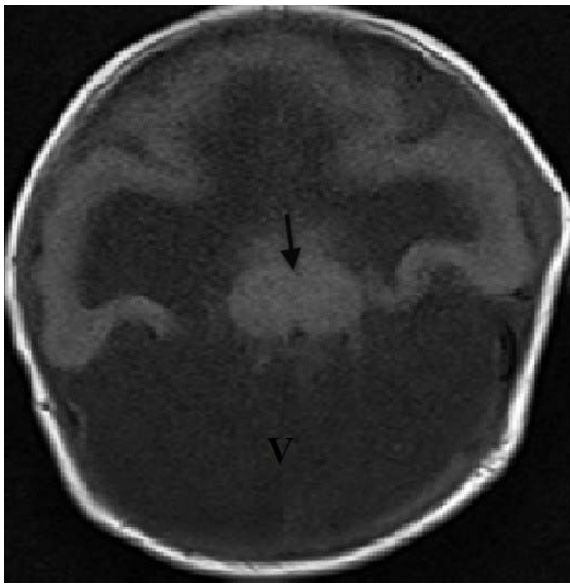


Fig. 15. Axial T1-weighted image shows a central monoventricle (V) with minimal residual frontal brain tissue with the fused thalami (arrow).

In lobar holoprosencephaly, the brain is generally of normal volume and shows almost complete separation into two hemispheres. The falx is often dysplastic anteriorly but otherwise normal. The ventricular system is well defined but may be dysmorphic. The septum pellucidum is absent. The corpus callosum is absent or dysmorphic (Fig. 16A, B) (15).

3.4.2. Septo-optic Dysplasia

Septo-optic dysplasia is a hypoplasia of optic nerves, combined with hypoplasia or absence of the septum pellucidum. It is thought that septo-optic dysplasia is a result of different genetic abnormalities and intrauterine ischemic events during the first two trimesters of pregnancy (3). It is characterized by hypoplastic optic nerves/tract, absence of septum pellucidum and hypothalamic-pituitary dysfunction (16). Diagnosis is confirmed by optic discs hypoplasia, absence of the septum pellucidum and optic nerves atrophy detection on CT and MRI (Fig. 17) (3).

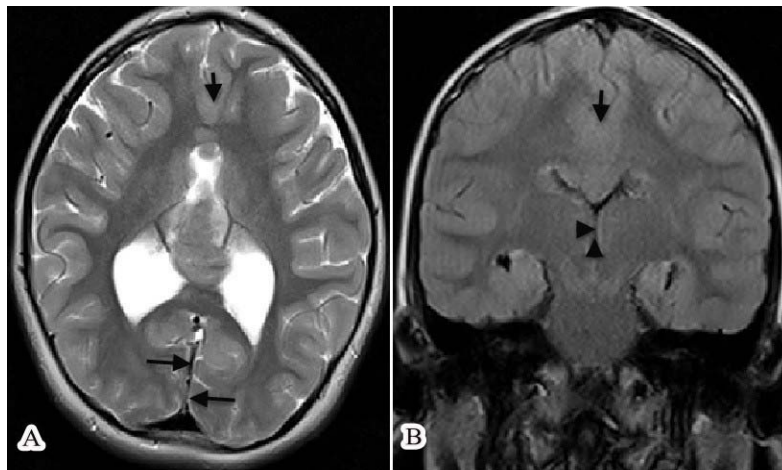


Fig. 16. Axial T2 (A) and coronal T1 (B) weighted images show fusion of the frontal lobes (short arrows), separated thalami (arrowhead) and presence of the falx (long arrows).

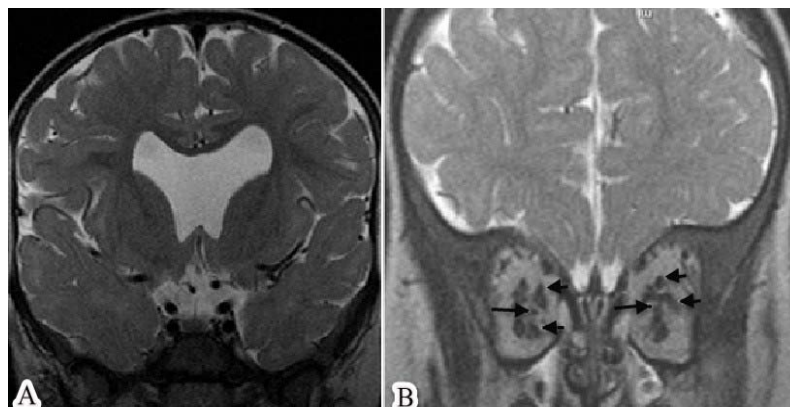


Fig. 17. Coronal T2 weighted image shows absent septum pellucidum with squared-off appearance of frontal horns with inferior pointing (A). Optic nerves (long arrows) and ocular muscles (short arrows) are hypoplastic (B).

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