Magnetic Resonance Imaging of Congenital Brain Malformations

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Abstract

Brain malformations constitute an important cause of morbidity and mortality in the pediatric age group. Magnetic resonance imaging (MRI) has revolutionized the depiction of these malformations by providing excellent anatomical inforamtion. This paper reviews briefly the important MRI features of common clinically encountered brain malformations.

Key words: *Brain malformations, MRI; agenesis of the corpus callosum, MRI; holoprosencephaly, MRI; Arnold-Chiari malformation, MRI; Dandy-Walker malformation, MRI.*

Central nervous system (CNS) malformations constitute an important part of genetic disorders that now number more than 2,000. They occur in almost one percent of all births and represent at least 10% of all systemic malformations. Approximately 75% of fetal deaths, 40% of infant deaths and .05% of deaths in children under one year of age are caused by CNS malformations. The most severe forms of malformations are incompatible with life and are usually aborted. The ones that are clinically encountered are relatively mild in severity.

Magnetic resonance imaging (MRI) has revolutionized the evaluation of neurological diseases and provides exquisite neuroanatomical detail. Magnetic resonance imaging basically comprises placement of the patient in a magnet that results in alignment of the protons in the North to South axis of the magnet. By employing a radiofrequency that varies with strength of the magnet the protons are disturbed from their axis for a short moment. As soon as the radiofrequency application is terminated, the protons move back to regain their original position. In the process they emit a radiofrequency that is picked up by an antenna. This information is then used by computers to generage an image.

This paper is intended to briefly review the application of MRI to the investigation of CNS malformations[1]. I shall discuss only the most commonly encountered malformations and illustrate their salient MRI features.

Dysgenesis of the corpus callosum

The corpus callosum is the largest commissure of the brain that allows crosstalk between each cerebral hemisphere. The commonly used designation "agenesis of the corpus callosum" is incorrect. Corpus callosum is never totally absent. Longitudinal fibers are always present. The ones that are usually absent or dysgenetic are the ones that cross horizontally from one cerebral hemisphere to the other. The correct terminology therefore is dysgenesis of the corpus callosum.

In developmentally disabled individuals its incidence has been reported at 2.3%.[2] It may be present alone or in combination with other malformations. Since the corpus callosum develops from front to back, the partial dysgenesis usually occurs at middle to posterior part of the corpus callosum.

On MRI, the sagittal images provide the best perspective. In cases where there is absence of the horizontal fibers that cross from one cerebral heimsphere to the other, the genu, the body and the splenium of the corpus callosum cannot be recognized on sagittal images (Figures 1). The gyri on the medial surface of the cerebral hemisphere converge toward the site of the absent corpus callosum due to inrolling of the cingulate gyrus.[3-5] The third ventricle is placed higher than normal due to its maintaining a position that exists in the fetal brain prior to development of the corpus callosum at 16 to 20 weeks of

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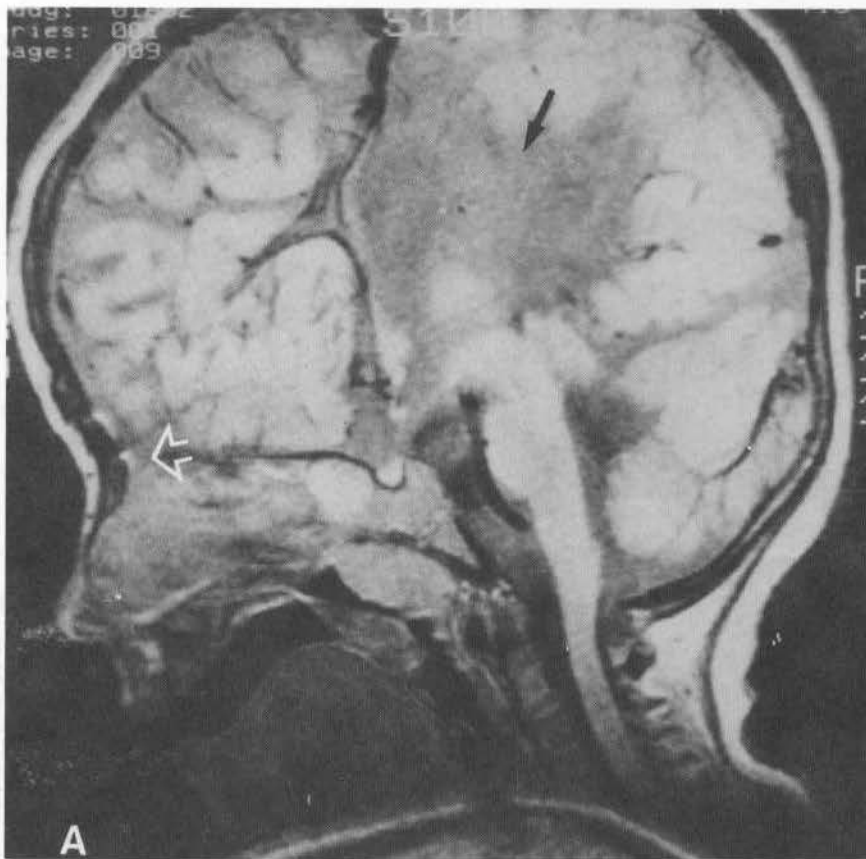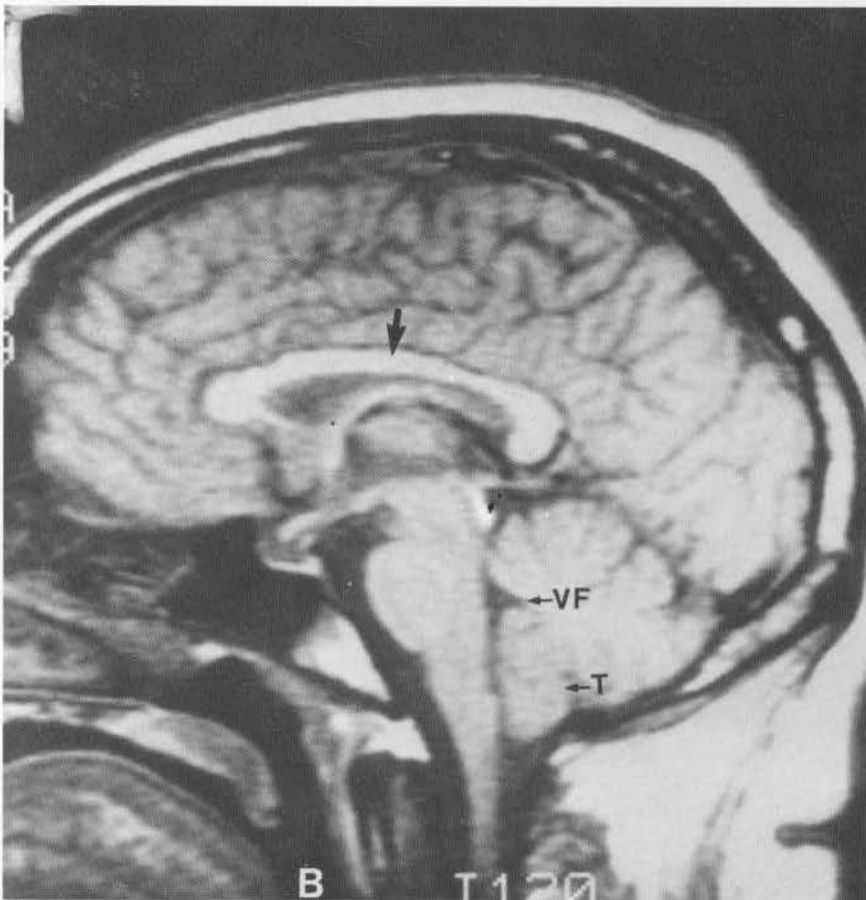*1740 West Taylor Street*
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Figure 1. Dysgenesis of the corpus callosum (DCC) and nasal encephalocele. A 3-year-old female with plagiocephaly, psychomotor retardation and cerebrospinal fluid rhinorrhea caused by nasal encephalocele.

A
Midline sagittal MRI. There is no corpus callosum (straight arrow). A small nasal encephalocele (open arrow) can be seen at site of absent crista galli.

B
Normal midline sagittal MRI of another patient to show the normal appearance of the corpus callosum (arrow). VF = Normal fourth ventricle. T = Normal cerebellar tonsil.
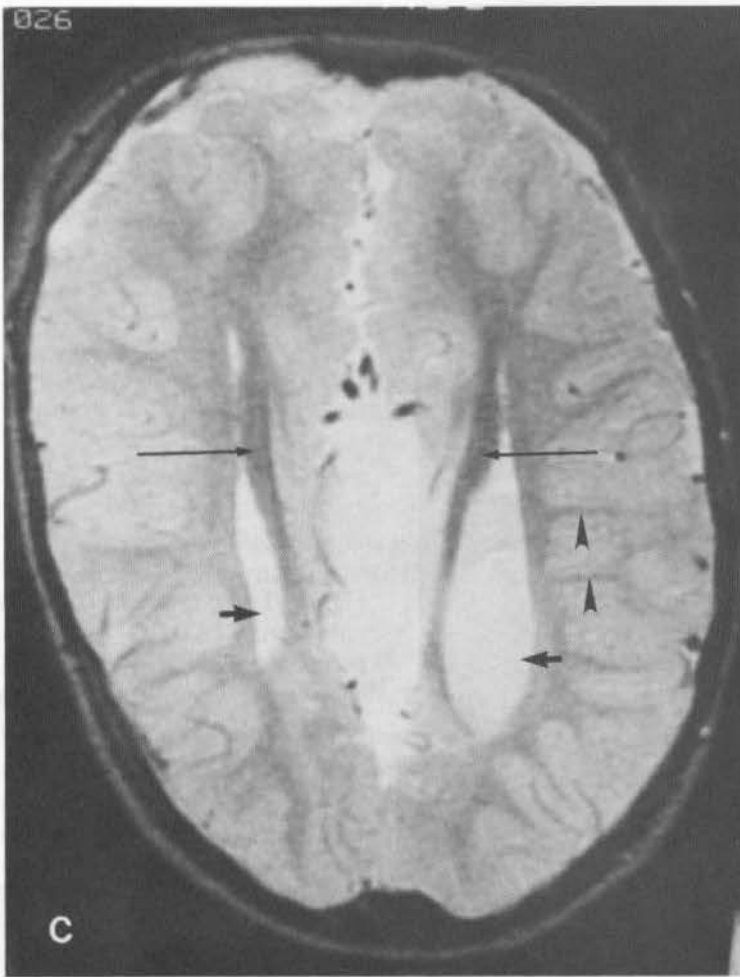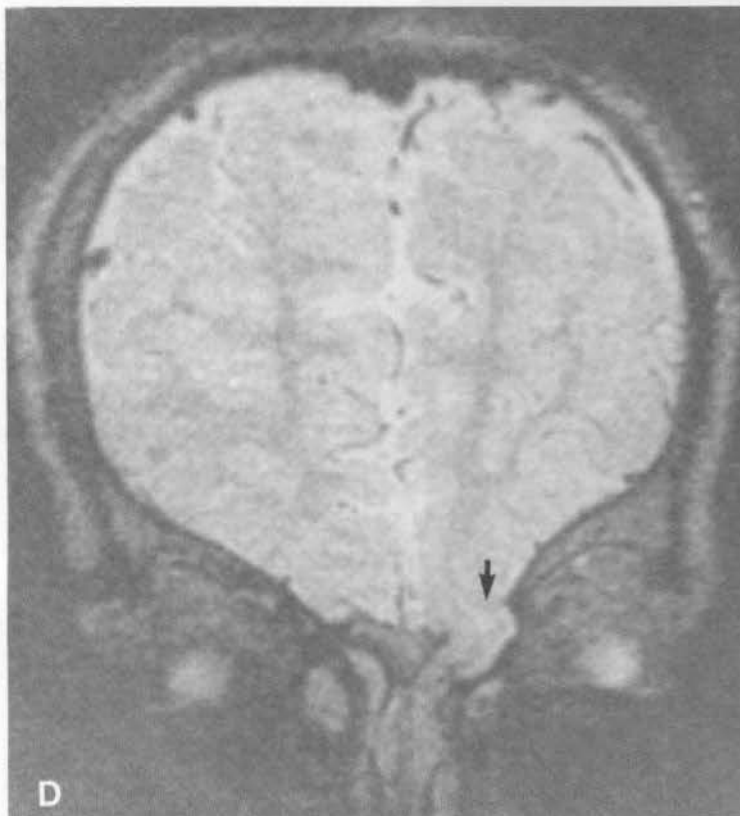
Figure 1.

C

Axial MRI to show features of DCC. The lateral ventricles (short arrows) are abnormally separated from each other. The occipital horns are disproportionately larger than other parts of the lateral ventricles. Probst bundles (long arrows) are clearly shown. These bundles are the longitudinal components of the dysgenetic corpus callosum. The horizontal bridging corpus callosum fibres have not formed. Thin white matter radiations (arrowheads at two) represent generalized white matter hypoplasia that is an important characteristic of this malformation.

D

Coronal MRI clearly showing the herniation of cerebral tissue (arrow) into nasal cavity (nasal encephalocele).
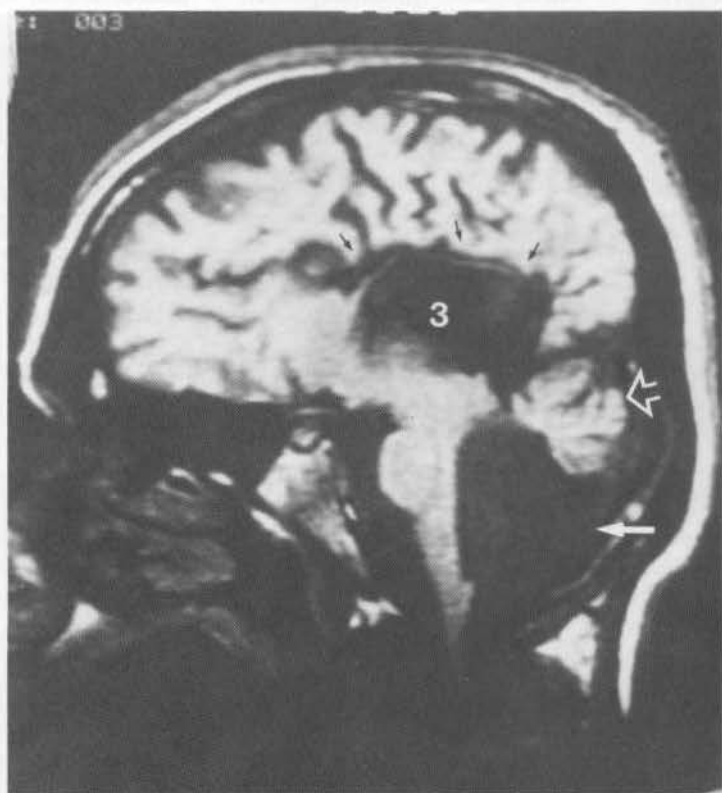
Figure 2. A 21-year-old male with ataxia. Sagittal MRI shows Dandy-Walker malformation (white arrow). The superior and anterior part of the vermis is present (open arrow). There is dysgenesis of the corpus callosum as well. Note the gyri converging at the site of absent corpus callosum (black arrows). Third ventricle (3) is enlarged and its roof located higher than normal.

Figure 3. A newborn with massive head enlargement. There is an occipital meningocele (arrow) that contains a hugely dilated fourth ventricle (4) with the Dandy-Walker malformation as its contents.

gestation. There may be associated polymicrogyria and gray matter heterotopia. On axial images the longitudinal fibers of the corpus caccosum (Probst bundle) can be exquisitely recognized (Figure 1C). The anterior commissure may or may not be present and occasionally may be enlarged to provide an avenue of communication from one cerebral hemisphere to the other. Similarly the fornix may be hypoplastic. The dysgenesis of the corpus callosum should be considered as a manifestation of generalized white-matter hypoplasia. This aspect accounts for generalized enlargement of the ventricular system, especially of the occipital horns. There usually is hypoplasia of the brainstem as well, accounting for enlargement of the fourth ventricle and of the subarachnoid spaces.

The other disorders that can accompany dysgenesis of the corpus callosum include interhemispheric arachnoid cyst and midline lipomas that usually calcify.

The patients with dysgenesis of the corpus callosum may show psychomotor retardation and seizure disorders.

Dandy-Walker malformation

Dandy-walker malformation is characterized by a triad of a) vermian hypoplasia or aplasia, b) continuation dorsally of the dilated fourth ventricle with a cyst and c) hydrocephalus. It occurs before 12 weeks of gestation. The anterior and susperior part of the vermis is almost always present. The defect always ocurs at the inferior medullary velum (Figures 2, 3) and then involves the remaining vermis to a varying degree. Whereas hydrocephalus may not be present at birth, it is usually recognized by age two months. The description in older textbooks and literature that suggests this malformation is characterized by atresia of outlet foramen of the fourth ventricle (foramen of Magendie and foramina of Luschka) is a myth. There is at least some patency of one or more outlet foramina of the fourth ventrcle. These patients exhibit a variable degree of cerebellar dysfunction. Unless there is concomitant malformation of the cerebral hemispheres, these patients may not show any psychomotor retardation.

It is not uncommon to see prominent CSF space at the level of the inferior medullary velum with enlargement of the vallecula; it is difficult in such cases to state whether this situation is a mild form of Dandy-Walker malformation or a normal variant (Figure 4). This aspect of the malformation requires further research correlating the morphologic features as shown by MRI and the clinical presentation of the patient.

Holoprosencephaly

This term implies a malformation of the pro-

sencephalon (prosencephalon = telencephalon and diencephalon). The malformation is characterized primarily by failure of the telencephalon and diencephalon to cleave and occurs in the early part of the first trimester. Thus, depending on the severity of the malformation, the cerebral hemispheres (telencephalon), thalamus, and hypothalamus (diencephalon) show a variable degree of fusion. There is a frequent association of this malformation with malformations of the face and the severity of face dysmorphology commonly correlates with severity of brain malformation. This association can be traced to their closely related embryologic origin. The prechordal mesoderm, located at the cephalic end of the notochord, gives origin to face structures and lies in close proximity to the developing prosencephalon. Though prechordal mesoderm and prosencephalon each have a mutually inductive influence on growth of the other, this relationship is heavily dominated by the prosencephalon.

There are three varieties of holoprosencephaly complex: a) alobar (Figure 5), b) semilobar, and c) lobar. In Alobar Holoprocencephaly, which is the most severe form, there are no cerebral hemispheres; interhemispheric fissure, falx and olfactory bulbs are absent. There is a single ventricle. In Semilobar Holoprosencephaly there is fusion of frontal and parietal lobes accounting for absence of the interhemispheric fissure anteriorly. The olfactory bulbs are usually absent or hypoplastic. In Lobar Holoprosencephaly the cerebral hemispheres are either normally formed or show continuity of some gyri deep within the interhemispheric fissure. There is some dysmorphology of the ventricular system characterized by bulbous appearance of the frontal horns, absence of the septum pellucidum or both. The olfactory bulbs are aplastic or hypoplastic.

The facial anomalies are classified as: a) cyclopia, b) ethmocephaly, c) cebocephaly, d) medial cleft lip with hypotelorism, and e) intermaxillary rudiment with hypotelorism. Common to all these facies is aplasia or hypoplasia of the medial facial skeleton of frontonasal prominence origin. Hypotelorism is virtually present in all cases.

Holoprosencephaly can be associated with other brain malformations that include dysgenesis of the corpus callosum, encephalocele and Dandy-Walker malformation. Its familial occurence and concurrence with chromosomal aberrations (Trisomy 13-15) have been reported. Most patients with alobar holoprosencephaly die in infancy. Longer survival is possible in milder forms of the disorder.

Septo-optic dysplasia (de Morsier syndrome)

Septo-optic dysplasia consists of a) primary hypoplasia of the optic nerves, b) absence of the septum pellucidum (Figure 6) and c) hypothalamic –
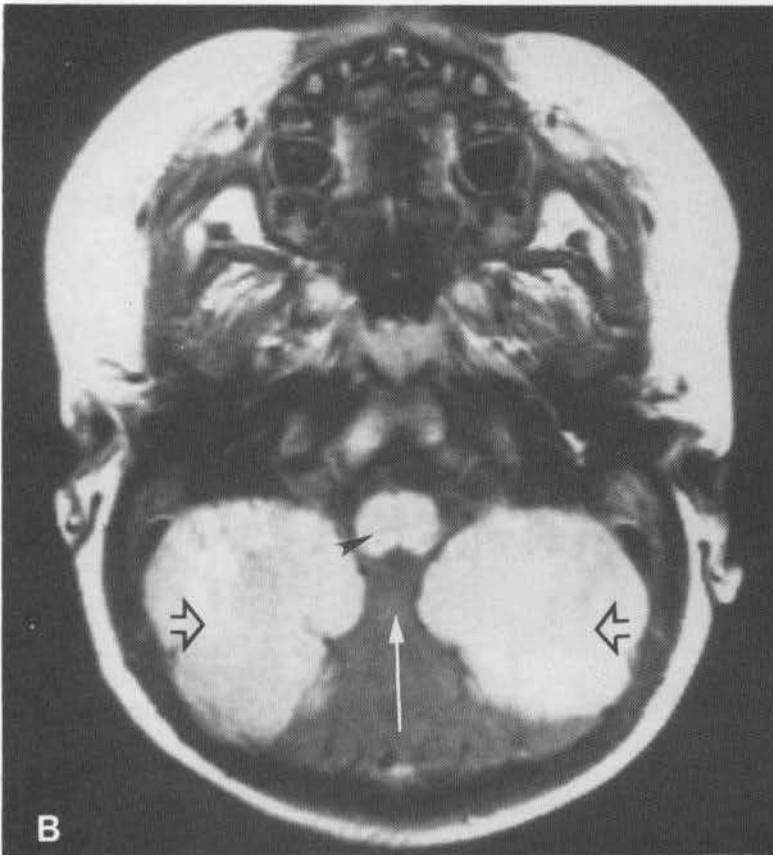
Figure 4. A one-year-old male with microcephaly, developmental delay, seizures and mild facial dysmorphology. A) sagittal and B) axial MRI. There is a cystic space that starts at the inferior medullary velum – foramen of Magendie level (arrows) and continues inferiorly under the vermis and posteriorly between the cerebellar hemispheres. It is difficult to state whether this is a normal anatomical variant or a mild form of Dandy-Walker malformation. Arrowhead = Brainstem. Open arrows = Cerebellar hemispheres.
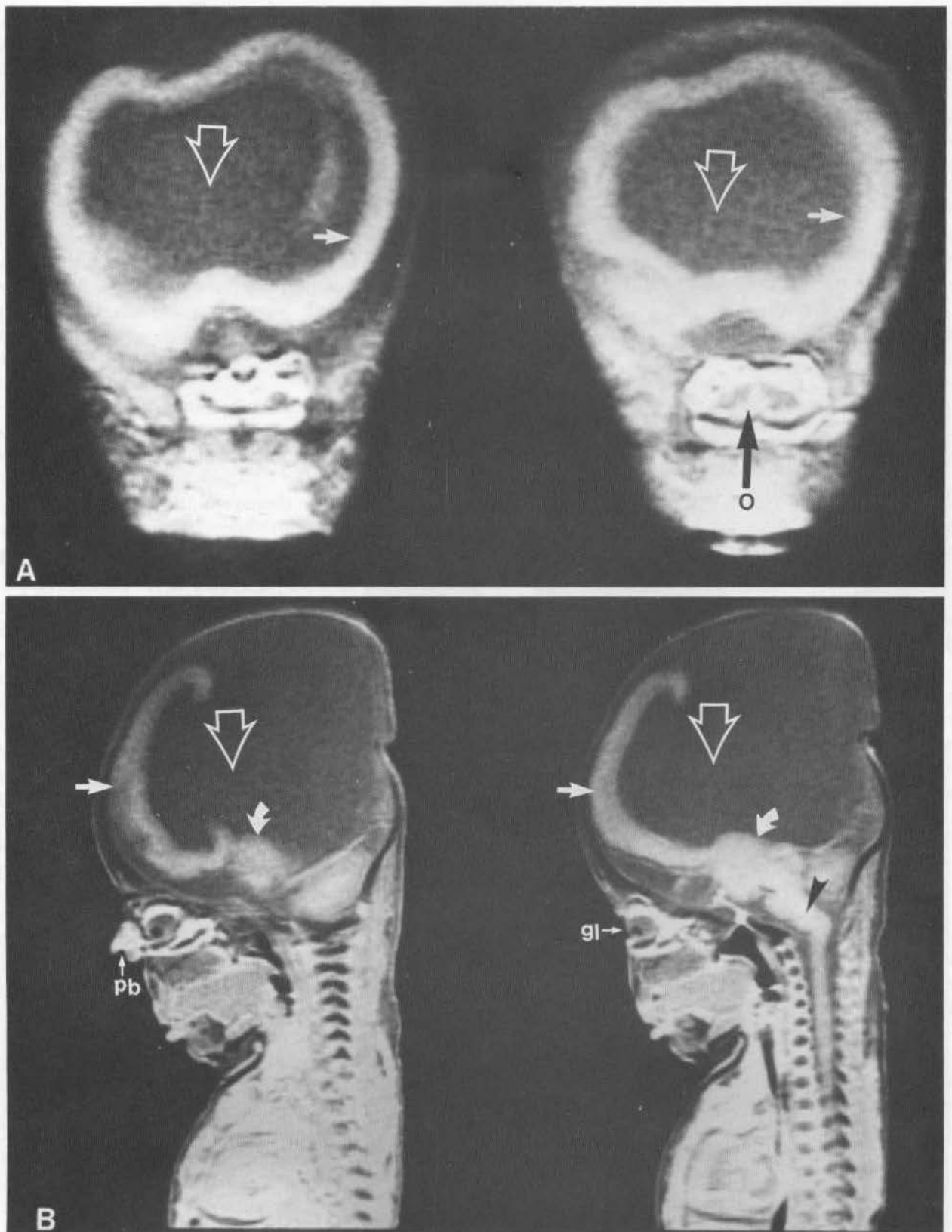
Figure 5. A stillborn cyclops with alobar holoprosencephaly. Coronal (A) and sagittal (B) MRI. The cerebral hemispheres are respresented by a smooth strip of neural tissue that is present only in the frontal region (closed arrows). The lateral ventricles and the third ventricle cannot be identified as such and are replaced by a CSF containing cystic structure (open arrows). The basal ganglia (curved arrowhead) are represented by a globular dysmorphic neural tissue. The pituitary gland and the sella turcica have not formed. The brainstem (arrowhead) is hypoplastic and shows somewhat normal form. In coronal MRI the orbits (O) are closely placed together and contain rudimentary globe and eye muscles. In sagittal image the proboscis (PB) and rudimentary globe (gl) can be seen.
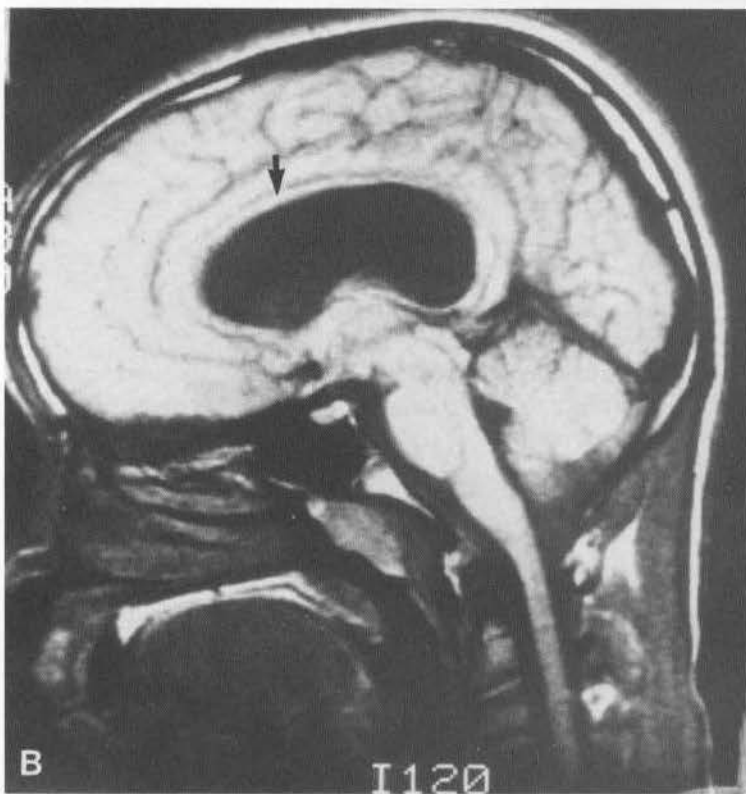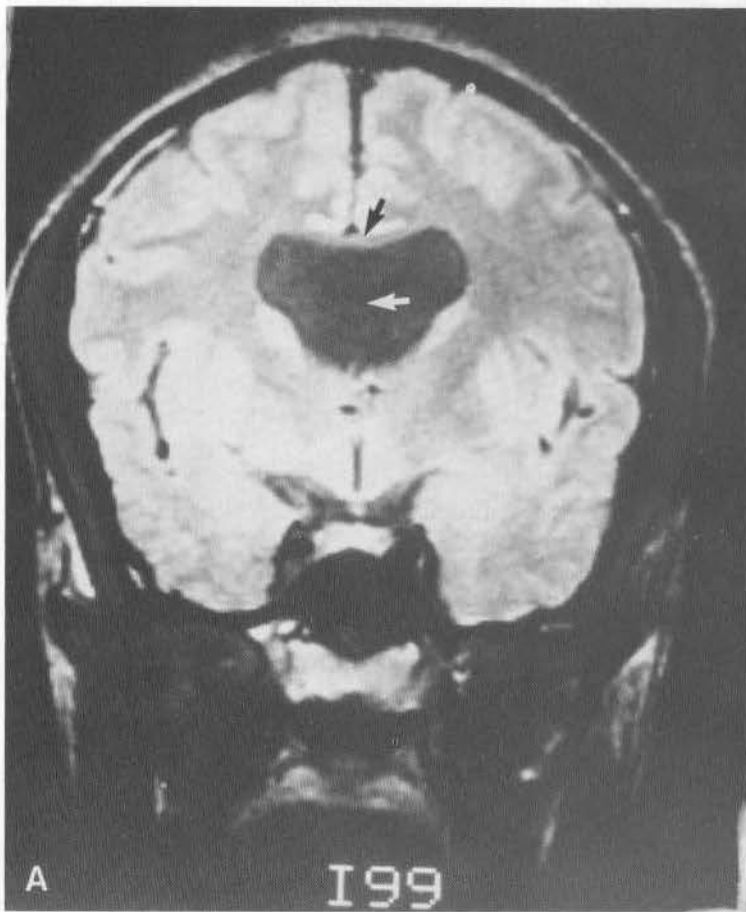
Figure 6. Probable septo-dysplasia. A 15-year-old male with mental retardation and behavior disorder. Coronal (A) and sagittal (B) MRI. There is absence of the septum pellucidum (white arrow) and the roofs of the dilated lateral ventricles are flat. The corpus callosum (black arrow) is hypoplastic.

pituitary dysfunction, especially deficiency of the growth hormone.

The malformation is considered a very mild form of holoprosencephaly complex, probably resulting from an abnormal induction of the prechordal mesoderm. It occurs at about the fourth to sixth week of gestation.

Clinically, these patients are blind and show searching nystagmus. Though it has been reported mainly in the young, it has been recorded in older patients as well. Dwarfism is related to growth hormone deficiency. Diabetes insipidus may follow hypothalmic dysfunction.

Arnold–Chiari malformation

This is a complex malformation that must include in its mildest form at least herniation of the inferior cerebellum below the foramen magnum to more than 3mm.[6] It is classified into three types depending upon herniation caudally of posterior fossa structures and the presence of associated anomalies of the mesencephalon and telencephalon.

Type I is characterized by displacement only of the inferior cerebellum into the upper cervical canal.[7-9] It is usually seen in older children and adults and can be associated with skeletal anomalies at the cranio–cervical junction.

Type II (Figure 7) includes the features in Type I plus a caudal displacement of the fourth ventricle and of medulla oblongata. The medulla oblongata buckles at its juction with the spinal cord. A myelomeningocele almost invariably accompanies this malformation. Other features include mesencephalic (tectal) beaking, large massa intermedia, absence of the septum pellucidum, disproportionate enlargement of the occipital horns, beaking of the frontal horns, overlapping by the cerebellum of the mid to upper brainstem, hypoplasia of falx cerebri and tentorium, widening of the tentorial incisura and cranialacunia. Syringohydromyelia is relatively a common concurrence. This type is the commonest and is seen in infants. These patients can live up to three decades or longer and curiously may not show much intellectual impairment unless infection has occured of the ventricular shunt that is required for hydrocephalus.

Type III consists of displacement of the medulla oblongata, the fourth ventricle and almost all of the cerebellum into an occipital and high cervical encephalomeningocele.

Hydrocephalus in Arnold-Chiari malformation usually cannot be explained by a single factor. A multitude of factors probably operate in various combinations. These include: a) an aqueductal obstructive lesion that is usually stenosis[10], b) compression of the subarachnoid spaces at the foramen magnum and at the tentorial incisura, c) obstruction of the outlet foramina of the fourth ventricle, and d) compression of the pliable venous sinuses of the posterior fossa leading to increased venous pressure and consequent CSF absorption impairment.

Neurocutaneous syndromes

This group of inherited disorders is characterized primarily by skin and CNS lesions. They are also known as phakomatoses, in reference to the associated skin lesions, a relationship that is explained by the common origin of ectoderm and neuroectoderm. Visceral lesions (mesodermal dysplasia) also may be present. These disorders are autosomally dominant and show a great propensity to develop neoplasia.

The following is a list of important phakomatoses:
1. Neurofibromatosis (Von Recklinghausen disease)
2. Sturge-Weber syndrome (encephalofacial angiomatosis)
3. Tuberous sclerosis (Bourneville disease)[11]
4. Von-Hippel-Lindau disease
5. Ataxia-telangiectasia (Louis-Bar syndrome)

Published reports have not yet emerged that document the MRI features of phakomatoses. However I believe that just as in other neurological diseases, MRI will depict the features of these disorders far more exquisitely than CT (Figure 8).

Craniofacial anomalies

Considering that the development of the mid-face from prechordal mesoderm and of the forebrain are so closely related embryologically, it comes as no surprise that we have encountered a varying dysmorphology of the brain in patients who show various face anomalies. These include; hypertelorism with or without associated cleft lip and palate (Figures 9, 10), cleft nose, various combinations of sutural synostoses, and facial skin lesions that do not fit into a known neurocutaneous disorder. I strongly suggest that any child that shows any peculiarity of the face must have an MRI, especially if there is concomitant psychomotor retardation. The recognition of a brain abnormality is important both for understanding the cause of psychomotor retardation and for genetic counseling.
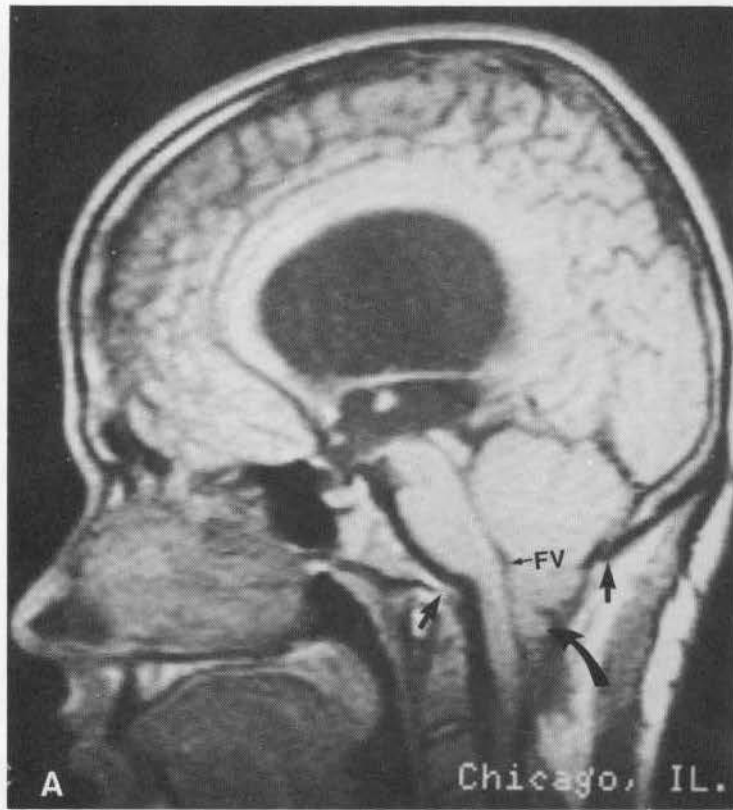
Figure 7. A 21-year-old male who had had repair of a lumbosacral myelomeningocele in infancy. There is associated Chiari II malformation and syringohydromyelia.

A
Sagittal MRI. The cerebellar tonsil (curved arrow) is below the foramen magnum (straight arrows). The fourth ventricle (FV) is narrowed and is located more caudally than normal. (See Figure 1B for normal posterior fossa structures). There is hydrocephalus and polymicrogyria.
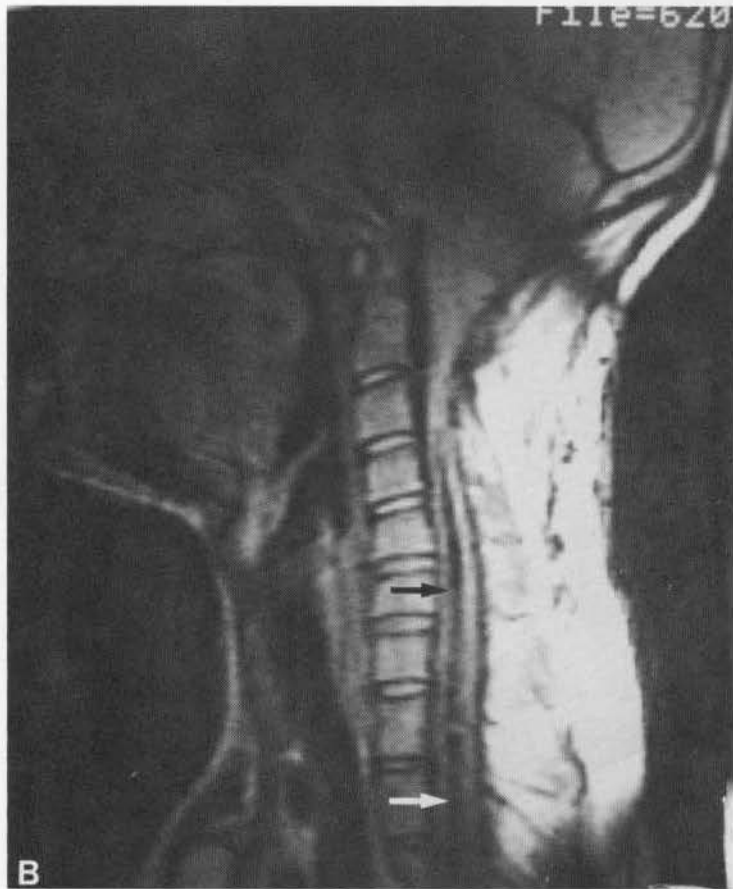
B
Sagittal cervical MRI. The cervical cord shows a long cavity lesion (arrows) indicating syringohydromyelia. This is a relatively common association of Chiari II malformation.
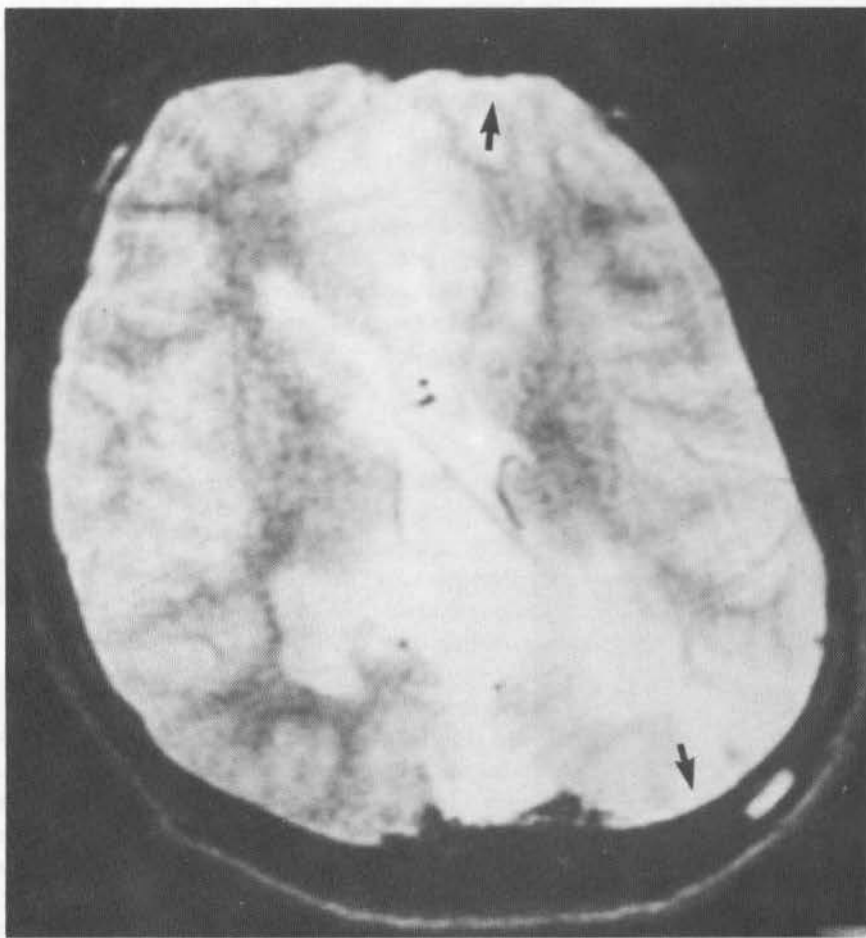
Figure 8. A 25-year-old male with facial stigmata of tuberous sclerosis, seizure disorder and severe mental retardation. Axial MRI. Note loss of gray-white matter differentiation (arrows) in left frontal and parietal lobes that is pathologically so characteristic of this disorder but had remained elusive to neuroimaging until the advent of MRI. Observe the normal appearance on the other side. The patient has dysgenesis of the corpus callosum as well.
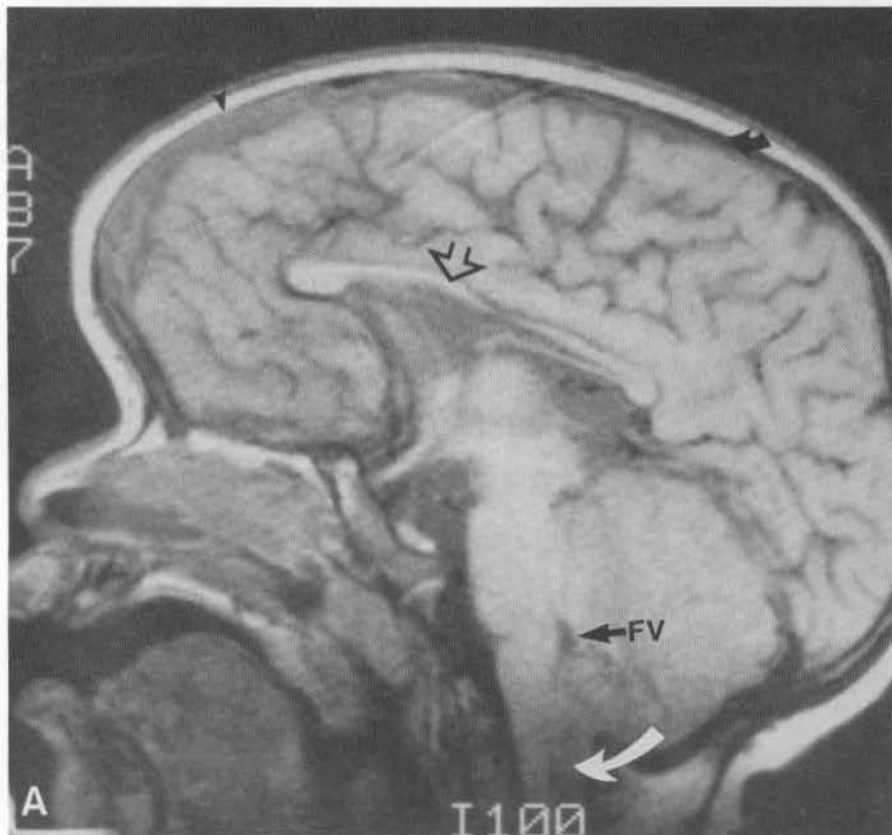

Figure 9. A 2-year-old female with oblique facial cleft and various brain anaomalies. A) midline sagittal;
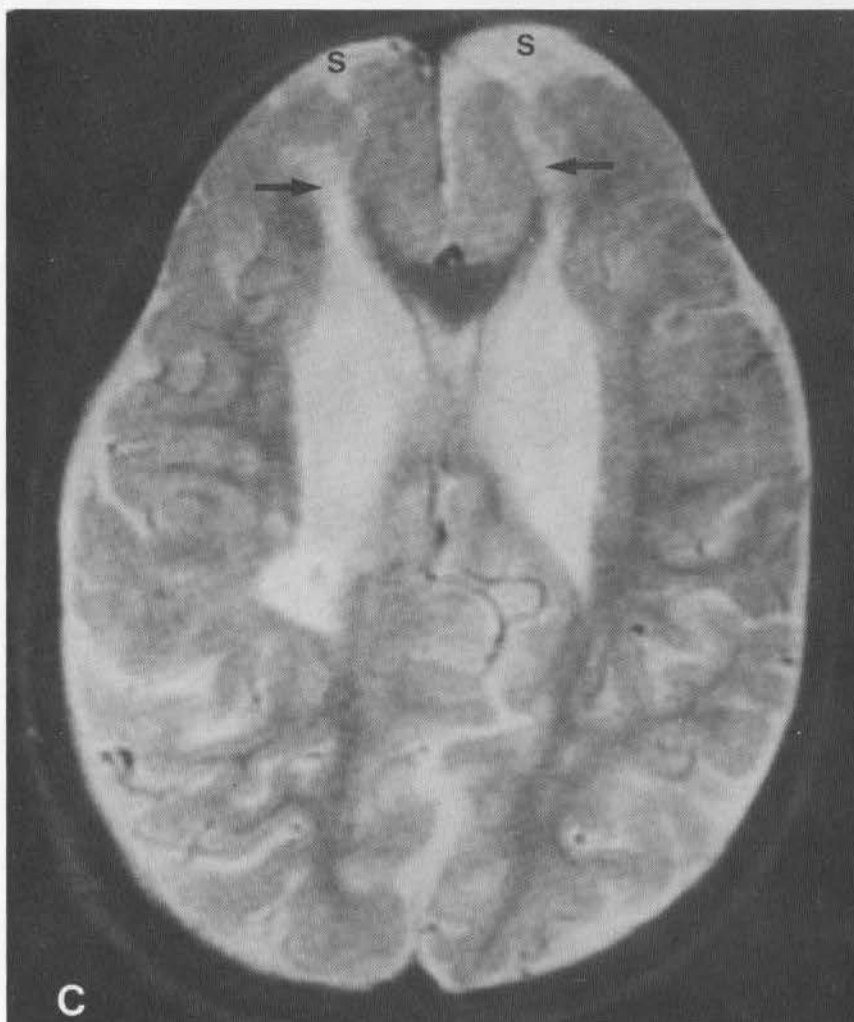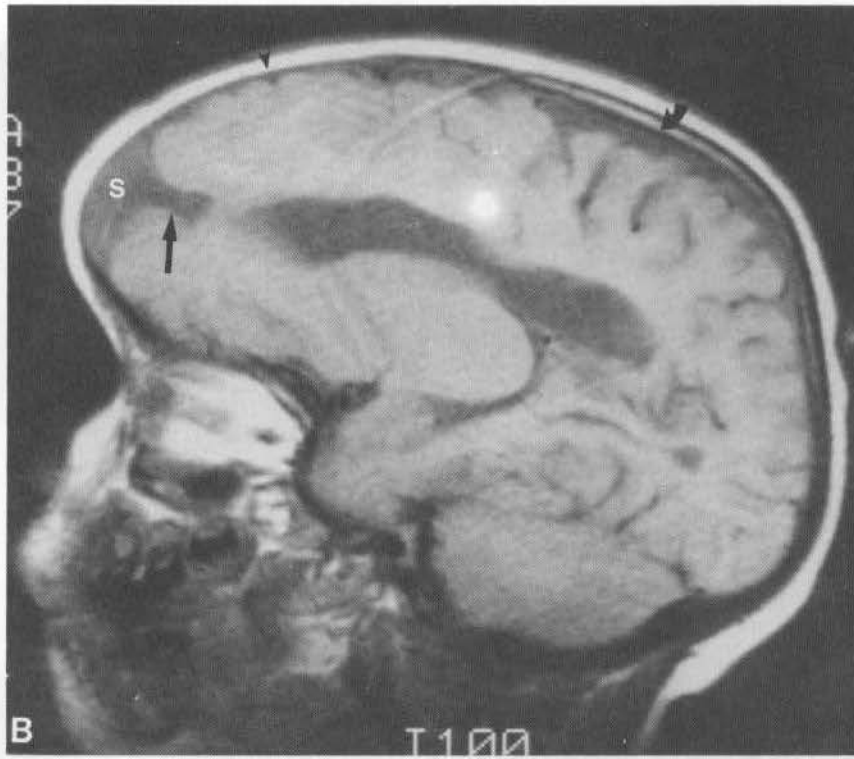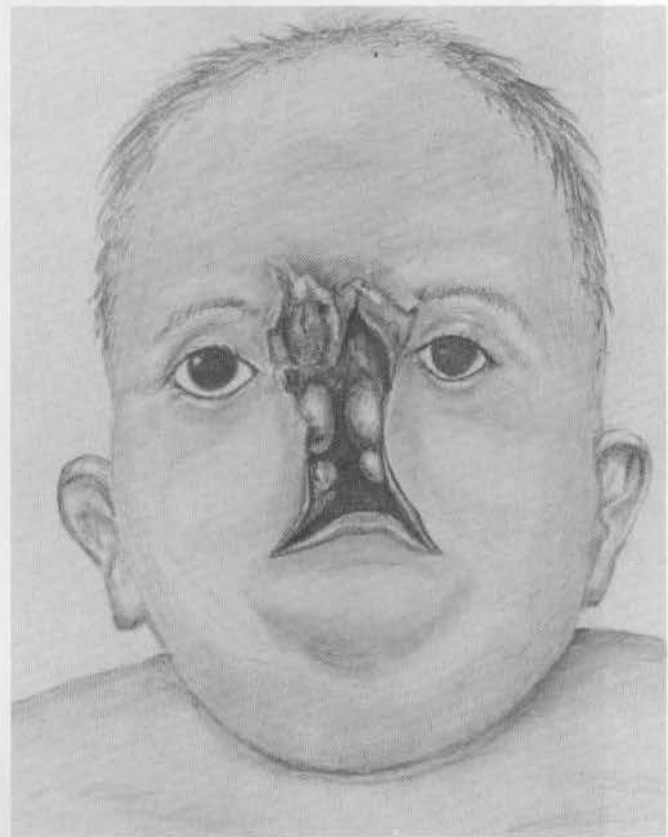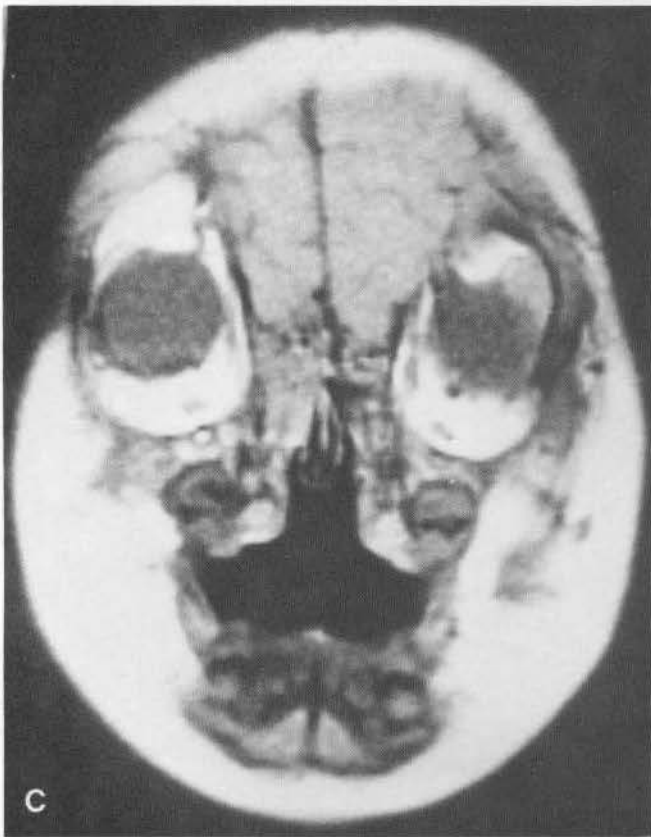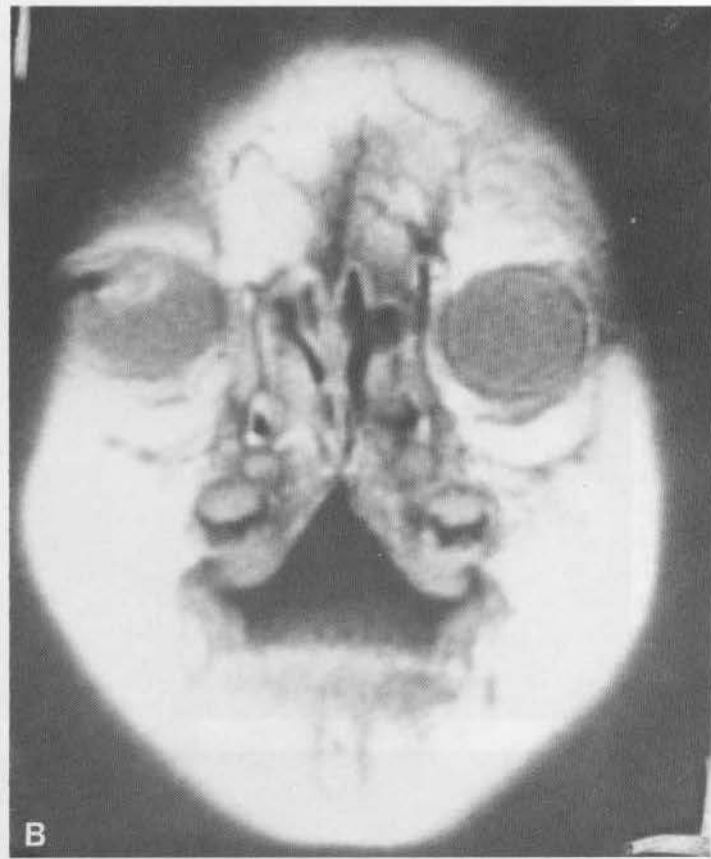
Figure 9.
B) parasagittal; and C) axial MRI. The anomalies are: 1) thin corpus callosum (open arrow); 2) bilateral frontal lobe schizencephalies (developmental clefts or porencephalies)(straight black arrows) that communicate with the subarachnoid space (S); 3) absence of the frontal bones (arrowheads)(curved black arrows are at normal calvarium); and 4) downward herniation of cerebellar tonsil (curved white arrow) and of narrowed fourth ventricle (FV)indicating Chiari II malformation. (See normal fourth ventricle in Figure 1B).
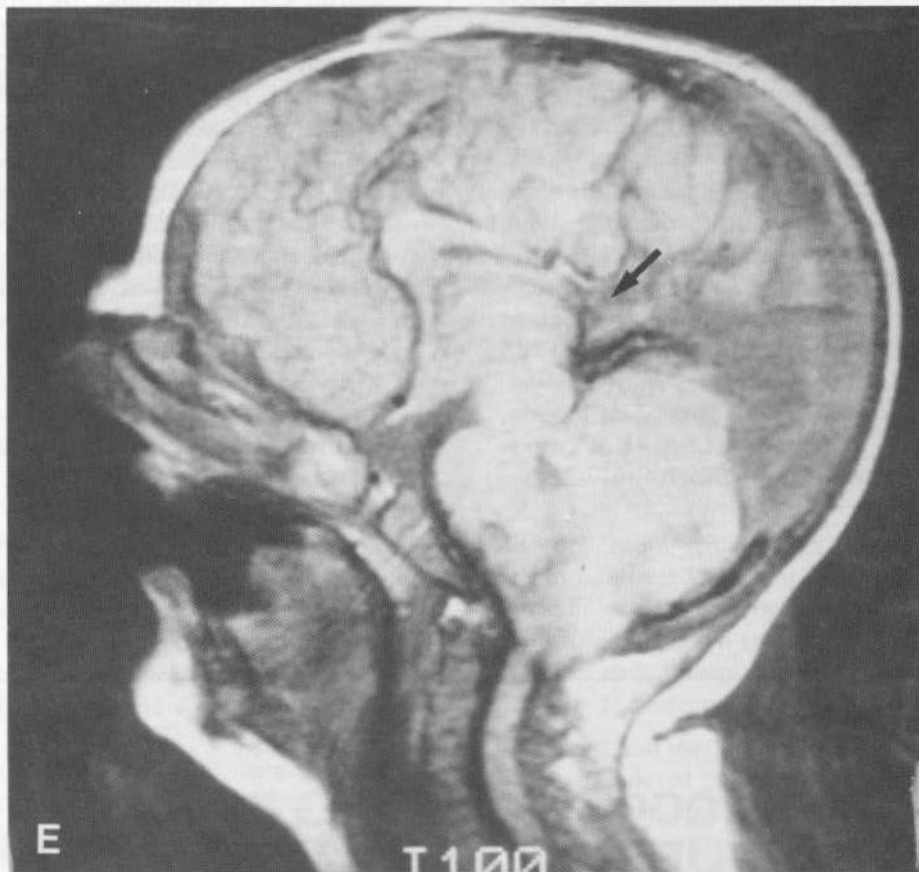
Figure 10. A 9-month-old male with a severe degree of cleft lip, cleft palate and cleft nose. Note that coronal images of MRI (A-C) depict the face dysmorphology quite faithfully. (D) shows a diagram of the face drawn from a picture of the patient's face. This patient's brain is hypoplastic and shows absence of the posterior part of the corpus callosum (arrow in E).

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