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Case 2.1
Nasal Chondromesenchymal Hamartoma

L. Santiago Medina and Sara M. Koenig

Fig. 2.1

Fig. 2.2

Fig. 2.3

Fig. 2.4
A 3-day-old neonate develops cyanosis during feeding. MRI reveals a large mass in the sinonasal region, calcifications, and erosion of adjacent bony structures.

Nasal chondromesenchymal hamartoma is very rare and benign ossifying fibromyxoid tumor, and it most commonly presents during infancy as a congenital condition, although it may present later in childhood. It is must be distinguished from other masses such as a dermoid teratoma, nasal glioma, and estheseioneuroblastoma as well as other chondroid, angiomatous, or lipomatous hamartomas. A hamartoma is a tumor-like formation that originates from excessive growth of tissues native to the site of origin, unlike a teratoma known to be caused by excessive growth of pleuripotential cells foreign to the site of origin. Additional presenting symptoms of nasal chondromesenchymal hamartomas include deficits or impairment of eye movement (unilaterally), asymmetry of the face, asymmetric maxillary swelling, difficulty or inability to breathe nasally, and protruding nasal polyps.

Histologically, a chondromesenchymal hamartoma consists of proliferative lobules of cartilage with contiguous spindle cells and myxoid areas of mesenchymal tissue, as well as extensive RER and Golgi complexes and microfilamentous bundles within the cells.

Treatment typically involves complete resection of the aberrant tissue. In this case, a septoplasty and right middle turbinectomy were also performed. Recurrence is common after an incomplete resection, but the tumor typically remains as a microscopic residual tumor. No adjuvant therapy is necessary.

Axial, sagittal, and coronal CT images show irregular broad-based mass located in the anterior and medial nasal fossa on the right with multiple calcifications, mass effect to the surrounding structures, and deviating the nasal septum to the left (Fig. 2.1a–c). Coronal T1-weighted and coronal and axial Fat Sat (FS) T2-weighted MR images demonstrate the mass being iso- to hypointense in a T1-weighted MR image and slightly hyperintense in a T2-weighted image with well-defined margins and calcifications better defined on CT. No apparent extension to the brain or orbits (Fig. 2.2a–c). T1-weighted FS coronal and axial images with contrast show homogeneous and intense contrast enhancement with adequate border delineation of the lesion without intracranial or intraconal extension (Fig. 2.3a, b). Biopsy specimen pathology slide confirmed the diagnosis (Fig. 2.4).

Acknowledgment

Acknowledgment to Dr. Raj Palani for their help on the preparation of this case.
Case 2.2
Pleomorphic Xanthoastrocytoma

Francisco Menor Serrano and María Jesús Esteban Ricós

Fig. 2.5

Fig. 2.6

Fig. 2.7

Fig. 2.8
An 11-year-old boy presents with sudden-onset focal left arm seizure.

Pleomorphic xanthoastrocytoma (PXA) is a rare, superficially located tumor arising from subpial astrocytes and often showing extensive involvement of the leptomeninges. Kepes et al. coined the term PXA to describe this tumor in 1979 and it was added to the WHO classification in 1993 as a grade II tumor. PXA is associated with a higher frequency of recurrence, anaplastic transformation, and death in comparison with other low-grade gliomas. Extent of primary resection is a significant factor in the prediction of recurrence-free survival. Response to chemo- and radiotherapy is uncertain. Isolated cases with widespread neuro-axis dissemination at diagnosis and some observations of PXA forming part of both ganglioglioma and dysembryoplastic neuroepithelial tumor have been reported. PXA is rarely diagnosed in infants, being discovered most commonly in adolescents and young adults. The most common single location of PXA is the temporal lobe (50%) and affected patients commonly present with seizures. PXA is uncommon in the basal ganglia, cerebellum, and spinal cord.

The classical, although nonspecific, appearance of PXA is a well-circumscribed superficial temporal solid-cystic mass. Solid components usually exhibit iso-attenuation in relation to gray matter on CT, iso or slightly hypo-intensity on T1-weighted images, iso or mildly hyper-intensity on T2-weighted images, and hyperintensity on FLAIR images and significant contrast enhancement. Calcification is variable and hemorrhage is rare. Large or small cysts are present in about 50% of cases. Surrounding vasogenic edema is usually minimal or absent. Leptomeningeal contrast enhancement is a distinctive finding, seen in more than two thirds of MRI studies.

Axial SE T1-weighted (Fig. 2.5) and T2-weighted MR images (Fig. 2.6) show a right, predominantly solid temporal lobe mass with small peripheral cysts surrounded by edema. The solid component is slightly hypointense on T1-weighted MRI and mildly hyperintense on T2-weighted MR images compared to gray matter. On coronal FLAIR MR images, the tumor exhibits greater hyperintensity, being difficult to make it out from surrounding vasogenic edema; note the small peripheral cysts being hyperintense in comparison to the ventricles (Fig. 2.7). Coronal post-contrast image demonstrates intense contrast enhancement of both the solid tumoral component and adjacent leptomeninges (Fig. 2.8). Reprinted with permission of Editorial Médica Panamericana; Menor F. Imagen en Oncología 2009.)

Figure 2.5
Figure 2.6
Figure 2.7
Figure 2.8
Case 2.3
Desmoplastic Infantile Ganglioglioma

María I. Martínez León
A 27-month-old boy presents with a single epileptic seizure episode. On physical examination, the infant had a protruding forehead on the left side.

Desmoplastic Infantile Ganglioglioma (DIG) is a rare, benign intracranial neoplasm of early childhood with involvement of the superficial cerebral cortex and leptomeninges. They are usually large, predominantly cystic tumors located in the frontal or parietal lobes. DIGs are classified as a benign WHO grade I tumor of infancy and consist of an uncommon variety of ganglioglioma that occur exclusively in infants. Seizures are the most common clinical symptom. Also, a rapidly enlarging head size may be seen.

With CT, a heterogeneous mass containing both a solid and cystic component is identified. With MR T1-weighted imaging, the solid portion of the tumor is isointense relative to normal brain parenchyma and demonstrates significant contrast enhancement. The cystic component has a low signal intensity on T1-weighted MR images and a high signal intensity on T2-weighted MR images. MR spectroscopy shows a lower NAA/creatine ratio, a higher choline/creatine ratio, and no significant change in myoinositol/creatinine ratio. This study may aid in narrowing down the diagnosis.

The differential diagnoses, based on the neuroimaging findings, are primarily, cystic supratentorial astrocytomas, and secondly, high-grade astrocytomas, PNETs, and ependymomas. If the leptomeningeal component of the tumor is large, meningioma and meningeal sarcoma are other possible considerations.

Total resection of the tumor may be curative, eliminating the need for chemotherapy or radiation.

Axial T2-weighted MR image revealed a large supratentorial, predominantly cystic tumor in the left cerebral hemisphere, displacing midline structures to the right. Additionally, the left lateral ventricle is effaced and displaced (Fig. 2.9). Coronal T1-weighted MR image with contrast shows a large cystic component with strong enhancement of a solid mural portion. Contrast enhancement is not seen in the walls of the cyst and the solid component is widely attached to the dura (arrow) (Fig. 2.10). MR diffusion-weighted imaging shows no restriction of the solid or cystic components (Fig. 2.11). MR venography was done before surgical intervention to highlight the absence of longitudinal superior sinus involvement (Fig. 2.12). Tumor was completely resected with surgery and the histological diagnosis was DIG. No recurrence was documented on follow-up examinations.
Case 2.4
Dysembryoplastic Neuroepithelial Tumor of the Septum Pellucidum (DNET SP)

María I. Martínez León and Bernardo Weil Lara

Fig. 2.13

Fig. 2.14

Fig. 2.15

Fig. 2.16
A 3-year-old girl presents with headache.

DNET SP are low-grade neoplasms arising at the midline, in the region of the septum pel-lucidum, with many of the histological features of the DNET. Imaging shows tumors extending into the lateral ventricles from the septal region and obstructing the foramen of Monro causing varying degrees of hydrocephalus. The lesions are lobular, well-delineated, internally septated, hypointense to gray matter on T1-weighted MR images, and hyperin-tense on T2-weighted MR images. There is usually no mass effect nor is there edema. Diffusion is not restricted and ADC map is high (may be attributable to the presence of large extracellular spaces and their low cellularity). DNET SP is usually non-enhancing or shows only minimal peripheral contrast uptake.

This neoplasm presents with the histological features of DNET, including the “specific glioneuronal element,” a histopathological hallmark characterized by axon bundles that form columns lined by small oligodendrogial-like cells.

First line of treatment is surgical resection and adjuvant chemotherapy or radiotherapy is not commonly needed.

On the basis of both neuroimaging and histopathology, DNET-like lesions should be considered as a differential diagnosis of midline, intraventricular tumors in children and young adults. Differentiating these tumors from more aggressive neoplasms is essential because of the benign evolution DNET SP.

There is a mass located in the anterior recesses of the third ventricle. Sagittal T1-weighted MR images without contrast and axial, T1-weighted MR images with contrast show its location with caudal extension to the suprachiasmatic recess and cranial extension to the intraventricular midline. Signal intensity is slightly increased in relation to CSF in T1-weighted MR images and there is no enhancement with contrast (Fig. 2.13 a, b). A slightly high signal similar to CSF can be appreciated on FLAIR sequences (Fig. 2.14), along with secondary ventricular dilatation due to obstruction of the foramen of Monro. T2-weighted MR image shows a signal similar to that of the CSF. Note that the vessels are encased by the tumor without alteration (Fig. 2.15). No restriction on DWI is identified (Fig. 2.16). According to the location, signal intensity, and behavioral pattern, the findings are indicative of DNET SP. There is histological confirmation of the radiological diagnosis.

Figure 2.13
Figure 2.14
Figure 2.15
Figure 2.16
Case 2.5
CNS Langerhans Cell Histiocytosis

Diego Alcaide Martín and María I. Martínez León
A 15-year-old patient was sent to the endocrinology department for assessment of diabetes insipidus.

Langerhans cell histiocytosis (LCH) is a rare condition that especially affects children and displays a wide variety of clinical manifestations. The most common features are bone lesions. There is limited knowledge about extra-osseous affectations of LCH. Examples of targeted systems include skin (55%) and the CNS (35%).

Approximately 25–35% of children with LCH, especially those who show multisystem manifestations, have CNS involvement. Two patterns have been described: granuloma formation and degenerative changes.

Granulomas can develop anywhere in the CNS, the most frequent location being the hypothalamic–hypophysary axis. MRI shows a loss of normal T1 signal from the neurohypophysis due to a decrease in storage of vasopressin, which leads to diabetes insipidus, a distinctive characteristic of the condition. MRI also displays an abnormal thickening and increased contrast enhancement of the hypophysis due to histiocytic infiltration.

Degenerative changes tend to occur in the cerebellum, especially in the dentate nuclei in a bilateral, symmetrical manner. Less often, the basal ganglia and brainstem are affected. These lesions cause inflammatory diffuse axonal damage, which leads to demyelination and, ultimately, atrophy. MRI shows hypointense lesions in T1-weighted MR images and iso or hyperintense lesions in T2-weighted MR images, which enhance with contrast proportionally to their degree of activity.

Lateral radiographs of the skull show multiple geographic lytic lesions of the bone with well-defined, non-sclerosed margins (Fig. 2.17). The MR T1-weighted image shows loss of the normal high signal from the neurohypophysis (Fig. 2.18). Bilateral, symmetric lesions of the white matter that are hypointense in T1-weighted images (not shown) and hyperintense in T2-weighted images characterize the cerebellar involvement (Fig. 2.19). After administering contrast, the hypophysis shows a normal uptake (not seen here) and the lytic lesions show a significant enhancement (Fig. 2.20). On the other hand, the cerebellar lesions do not present contrast uptake, which signifies demyelination and gliosis.

Figure 2.17
Figure 2.18
Figure 2.19
Figure 2.20
Case 2.6
Hemangioma of Infancy

Cristina Bravo Bravo and Pascual García-Herrera Taillefer

Fig. 2.21

Fig. 2.22

Fig. 2.23

Fig. 2.24
A 2-month-old girl presents with a rapidly growing bluish tumor on the right mammary gland that had appeared at approximately 2–3 weeks of age.

Hemangiomas are the most common soft-tissue tumors of infancy. They are usually absent at birth and appear between the second and sixth week of life. Hemangiomas show a characteristic clinical evolution: a phase of rapid proliferation (3–9 months) followed by a period of relative stability and finally, a phase of slow involution (18 months up to 10 years of age). Most are diagnosed clinically and do not require further diagnostic studies or treatment. The GLU-T1 immunohistochemical marker serves to differentiate the hemangioma of infancy from congenital hemangiomas and vascular malformations. Ultrasound and MRI are indicated in atypical cases and in lesions that are large in size in order to evaluate the extent of compromise and its relation with neighboring structures. Furthermore, imaging may aid in assessing associated abnormalities such as lumbar hemangiomas, spinal dysraphisms, segmented facial hemangiomas, PHACE syndrome, multiple cutaneous hemangiomas, and diffuse neonatal hemangiomatosus.

Sonographically, these tumors are well-delineated, lobulated, and show variable echogenicity. On gray scale, US vascular structures are not usually identified; although, on occasion, peripheral supplying arteries can be seen. Doppler US reveals high vessel density with high systolic arterial velocities and a low resistance pattern. There is little or no evidence of arteriovenous shunting, and veins show a monophasic pattern. Diagnostic criteria for hemangiomas of infancy include the presence of five or more blood vessels by square centimeters of area and displacement of the systolic frequency by 2 kHz or more. During the involutive phase, the size of the lesion and the number of vessels decrease, but arterial velocities remain unchanged.

Possible differential diagnoses include vascular malformations and other soft-tissue tumors. If a lesion does not meet the diagnostic criteria for hemangioma, a biopsy must be taken.

Ultrasound shows a predominantly echogenic mass with heterogeneous echo-structure and peripheral blood vessels (Fig. 2.21). Color Doppler shows a high vessel density with occasional areas of turbulent blood flow (Fig. 2.22). Spectral Doppler (Fig. 2.23) displays a low-resistance vascular pattern with high systolic velocities and a pulsatile venous flow due to small arteriovenous fistulas (Fig. 2.24). These findings are consistent with a hemangioma of infancy in a proliferative phase.

Figure 2.21
Figure 2.22
Figure 2.23
Figure 2.24
Case 2.7
Vascular Lesion of the Face
Sara M. Koenig and Juan E. Gutiérrez

Fig. 2.25

Fig. 2.26

Fig. 2.27

Fig. 2.28
A 5-year-old female presents with a left-sided facial vascular malformation and history of prior surgical interventions.

Capillary hemangiomas and venous malformations are each typically benign lesions of vascular channels. Hemangiomas are benign endothelial cell neoplasms that commonly occur in children, especially under the age of 12 months. A red-colored lesion with a lobulated appearance appears on the skin, from which rapid growth may occur within the first 12 months of life. Angiography reveals a capillary lesion with well-demarcated dense opacification throughout, and with substantial blood flow arising from dilated arteries and dilated venous drainage. These benign lesions are typically harmless and only pose a cosmetic defect that typically stabilizes within a year of age and, in some cases, regress within a few years. In some circumstances, hemangiomas may cause functional impairment that requires aggressive treatment. Functional impairments may include impairment of vision development, feeding patterns, or language due to location on the eyelid, lips, or inside the mouth. Other defects may include hemorrhage or airway defects due to obstruction. Treatment typically consists of surgical resection, laser coagulation, or embolization, whereas endovascular interventions are only used in extreme cases that involve thrombocytopenia and bleeding diathesis.

Arteriovenous malformations vary from hemangiomas in that they are a benign growth of vascular channels with little and poorly demarcated opacification during angiography. Direct percutaneous injection of contrast typically optimizes opacification for imaging. Arteriovenous and venous malformations are typically treated conservatively, although complications such as hemorrhage, infiltration, or osseous involvement may require surgical resection or endovascular treatments.

Axial unenhanced T1-w (Fig. 2.25), enhanced T1-w (Fig. 2.26), axial T2-w (Fig. 2.27) and coronal and sagittal T2-w (Fig. 2.28) MR images exhibit the large, complex lesion with cystic components and avid enhancement involving the right side of the face (and posterolateral aspect of the neck ending at the right posterior triangle of the neck). This mass involves the oral cavity, masticator compartment, parotid space, and submental regions. The imaging characteristics of this lesion are compatible with a large venous malformation.

Figure 2.25
Figure 2.26
Figure 2.27
Figure 2.28
Case 2.8
Retinoblastoma

Juan E. Gutiérrez and Sara M. Koenig
A 14-month old male presents with an abnormal fundoscopic exam. His mother has a history of bilateral retinoblastoma. Calcifications appear in the soft tissue of the left eye.

Retinoblastoma (RB) is the most common intraocular malignancy in children. Of all retinoblastoma cases 70–80% are in infants less than 2 years old, and these tumors arise from retinal tissue. The most common presentation of retinoblastoma is leukocoria in early childhood, or a whitening of the retina seen on fundoscopic exam.

The most common mutation associated with RB is in the RB1 tumor suppressor gene on chromosome 13 controlling progression of the cell cycle, and greater than 200 mutations have been found. Most cases are sporadic; however, 10% are heritable as an autosomal dominant disease. Hereditary RB is often bilateral (rather than unilateral), and among all cases of retinoblastoma approximately 30% are bilateral and 30% multifocal. “Trilateral RB” occurs in approximately 4–7% of individuals with bilateral retinoblastoma, where a small cell intracranial tumor concurrently develops. These individuals often present at an earlier age than those with unilateral or sporadic retinoblastoma, have a higher likelihood of hereditary retinoblastoma, may develop additional tumors in the pineal, suprasellar, or fourth ventricular regions, and have a poor prognosis.

Imaging studies usually start with US. On CT scan revealing a high-density mass with calcifications arising from the retina, although margins may vary from well delineated to very unclear. Calcification within these tumors is considered a primary factor in the radiological diagnosis of RB. Retinal detachment is often seen due to the local mass effect of the tumor, and extension of the tumor often follows the optic nerve or the lymphatics of the orbit. MRI should be used in patients with suspected intracranial spread of the tumor or with bilateral retinoblastoma, and increased attention should be given to areas mentioned above: the pineal, suprasellar, and fourth ventricular regions. MR images are more sensitive to the spread of the tumor along the optic nerve and, with contrast, illustrate a well-enhanced intraocular mass. Unenhanced T1-and T2-weighted MRI show a mass at approximately the same intensity as normal gray matter.

CT without and with contrast, show of the left orbit revealing retinal high density enhancing mass with calcifications (Figs. 2.29 and 2.30). MRI axial Fat-Sat post-contrast image reveals left retinal detachment due to a solid mass with homogenous enhancement (Fig. 2.31). Fundoscopic appearance of the lesion (Fig. 2.32).
Case 2.9
Tuberous Sclerosis

Ana Alonso Murciano and María I. Martínez León

Fig. 2.33
Fig. 2.34
Fig. 2.35
Fig. 2.36
Young boy presents with known congenital syndrome and uncontrolled seizures.

Tuberous Sclerosis (TS) is an autosomal dominant neurocutaneous syndrome characterized by the presence of benign congenital tumors in multiple organs. The diagnosis is usually established on the basis of major and minor diagnostic criteria applied to physical or radiological findings. The classical triad of epilepsy, mental retardation, and sebaceous adenoma is rare. TS is caused by a mutation of two tumor-suppressing genes known as TSC1 and TSC2. Mutation in TSC2 tends to result in a more severe form of the disease and a higher number of cortical tubers (CTs). Neurological involvement is seen in 95–100% of cases and includes CTs, subependymal nodules (SNs), subependymal giant-cell astrocytomas (SGCAs), and white matter abnormalities. Other common manifestations are renal angiomyolipomas (AMLs) (55–75% of cases) and cardiac rhabdomyomas (50–65% of cases).

1. CTs are characterized by the presence of dysmorphic neurons and large astrocytes. Patients with more than six CTs present with a greater difficulty to control seizures.
2. SNs and SGCAs represent hamartomatous changes in subependymal tissue. SNs are frequently calcified. SGCAs are typically located in the foramen of Monro and have a benign course. Nevertheless, due to their location, they may cause obstructive hydrocephalus.
3. White matter alterations include superficial white matter abnormalities associated with cortical tubers, radial white matter bands, and cyst-like lesions.
4. Cardiac rhabdomyomas are benign striated muscle tumors that are commonly located in the ventricular septum and may be single or multiple. Most of them do not cause clinical manifestations and spontaneous regression may occur.
5. AMLs are characterized by variable amounts of abnormal vessels and immature smooth-muscle and fat cells. In patients with TS, AMLs usually develops at a younger age and tends to be larger in size, bilateral, and multiple.

CT without contrast shows calcified subependymal nodules and frontal bilateral cortical tubers (arrows) (Fig. 2.33). Axial T2-weighted MR image depicts multiple cortical tubers and white matter abnormalities (Fig. 2.34). Coronal FLAIR MR image displays a left subependymal giant cell astrocytoma (Fig. 2.35). CT with contrast shows bilateral renal angiomyolipomas (Fig. 2.36).

Figure 2.33
Figure 2.34
Figure 2.35
Figure 2.36
Case 2.10
Neurofibromatosis Type 1

Inés Solís Muñiz
A 12-year-old boy with known neurofibromatosis type 1 presents with multiple *café-au-lait* spots, visual disturbances, mild mental retardation, and scoliosis.

Neurofibromatosis type I (NF-1), formerly known as von Recklinghausen disease, is a relatively common (1/3,000 live births) autosomal dominant genetic disorder classified as a neurocutaneous syndrome or phakomatosis. Diagnosis is usually established in childhood based on a series of well-known major and minor criteria.

Apart from the dermatological manifestations of the condition (*café-au-lait* spots, axillary freckles, Lisch nodules of the iris), neurological abnormalities such as myelin vacuolization (40–90%), and optic tract (30%) and cerebral (1–3%) gliomas can also be identified. Dermal neurofibromas are seen in 90% of cases and plexiform neurofibromas in approximately 30% of patients. While neurofibromas are usually considered benign nervesheath tumors, the plexiform variation has shown malignant transformation in up to 10% of cases. Other abnormalities include bone dysplasia (5%) and scoliosis. Patients with NF-1 also have a higher risk of developing genetically related tumors such as rhabdomyosarcomas and neuroblastomas. Close monitoring is required due to their increased tendency to develop both benign and malignant neoplasms.

Imaging studies, specifically MRI, play an important role in the detection, extension assessment, and follow-up of the aforementioned neurological and non-neurological manifestations of the disease.

Surgical resection of symptomatic tumors is currently the first line of treatment.

Axial FLAIR (Fig. 2.37) and coronal T2-weighted (Fig. 2.38) MR images show multiple focal hyperintense lesions of the cerebellar white matter, brainstem, and bilateral thalami. No mass effect or contrast enhancement is observed. These findings are consistent with myelin vacuolization. Coronal T1-weighted MR image shows a predominantly left-sided volume increase of the optic chiasm consistent with glioma (Fig. 2.39). Coronal STIR MR image of the thorax and superior abdomen exhibits a large number of paravertebral, intercostal, and bilateral subcutaneous tumors. Additionally, a large mass can be seen on the left hemithorax with hyperintense lobulations and a central, target-like loss of signal, typical of neurofibromas (Fig. 2.40).

Figure 2.37
Figure 2.38
Figure 2.39
Figure 2.40
Further Reading

Books

Barkovich AJ (2005b) Pediatric Neuroimaging. Lippincott Williams & Wilkins, Philadelphia

Web Links

http://www.childrenshospital.org/az/Site979/mainpageS979P0.html
http://www.searchmedica.com/search.html?q=pleomorphic%20xanthoastrocytoma
http://www.radiographics.org
http://journals.lww.com/ajsp/pages/default.aspx
http://scielo.isciii.es/scielo.php
www.issva.org (International Society for the Study of Vascular Anomalies)

Articles

Further Reading

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Learning Pediatric Imaging
100 Essential Cases
Martínez-León, M.I.; Ceres-Ruíz, L.; Gutierrez, J.E.
2011, XIX, 243 p. 400 illus., 164 illus. in color., Softcover
ISBN: 978-3-642-16891-8