The Radiological Society of North America 86th Scientific Assembly and Annual Meeting

Image Interpretation Session: 2000

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Abbreviations: BCS = Budd-Chiari syndrome, CNS = central nervous system, HPV = human papilloma virus, LCH = Langerhans cell histiocytosis, PNH = paroxysmal nocturnal hemoglobinuria, PVNS = pigmented villonodular synovitis

Index terms: Angioma, 452.3141 • Appendix, neoplasms, 751.31222 • Aspergillosis, 60.2056 • Brain, abnormalities, 10.1452, 10.1839 • Brain, diseases, 134.253 • Bronchi, neoplasms, 671.319 • Budd-Chiari syndrome, 81.659 • Histiocytosis, 60.66 • Intestinal neoplasms, 74.3194 • Intussusception, 751.73 • Kidney, diseases, 81.659 • Knee, neoplasms, 452.3141 • Larynx, neoplasms, 271.369 • Lipogranulomatosis, 452.669 • Neurofibromatosis, 60.1472 • Papilloma, 271.369, 671.319


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Case 1

History and Findings
A 17-year-old boy presented with a painful, swollen right knee that had been symptomatic for the past 3 months. Plain radiography of the knee revealed a large, anteromedial cortical erosion of the tibia (Fig 1a) and a smaller erosive process near the joint line on the lateral aspect of the tibia (Fig 1b). No soft-tissue calcifications or mineralization were identified.
A sagittal T1-weighted magnetic resonance (MR) image obtained through the lateral knee showed the erosive changes in the tibia, both anteriorly and posteriorly (Fig 2a). A sagittal fast spin-echo T2-weighted image with fat suppression obtained through the lateral knee revealed that the erosive areas contained intermediate-signal-intensity soft tissue (Fig 2b). A prominent vascular component with one large vessel was seen extending inferiorly from the anterior erosion.

Surgery was performed to remove an intraarticular mass.

**Diagnosis:** Synovial hemangioma.

**Discussion**

The radiographs demonstrated a process that involved the tibia both medially and laterally; therefore, the finding most likely represented an intraarticular lesion. The MR images helped confirm the intraarticular location.

The differential diagnosis for an intraarticular process with underlying bone involvement includes pigmented villonodular synovitis (PVNS), synovial osteochondromatosis, rheumatoid arthritis, gout, and, less likely, synovial hemangioma and lipoma arborescens. In an otherwise healthy 17-year-old patient, rheumatoid arthritis and gout can be excluded. On MR images, PVNS has a characteristic appearance of low-signal-intensity hemosiderin deposits lining the synovium and scattered throughout the mass. The MR imaging findings in this case, however, were not characteristic for PVNS. Synovial lipomatosis has fatty deposits that are easily identified with MR imaging, but they were not seen in this case. Synovial osteochondromatosis typically has multiple discrete loose bodies (not seen in this case), but it may also manifest with loose bodies that are so tightly packed that they appear to be one large mass. This case could be such an example. However, the engorged vessels leading from the mass in this case make synovial hemangioma the most likely diagnosis.

Synovial hemangiomas are uncommon vascular lesions that primarily affect children or young adults, with a predilection for females. The knee is the most commonly affected joint. Most patients complain of a painful, swollen joint. The imaging characteristics of synovial hemangiomas are well demonstrated by this case. An intraarticular lesion with bone erosions and little mass effect is typical. Phleboliths can be present and are virtually pathognomonic. The presence of large vascular channels with or without fatty septa make the diagnosis of a synovial hemangioma straightforward.

**Suggested Readings**


**Case 2**

**History and Findings**

A 44-year-old woman presented with a 2-week history of primarily right-sided abdominal discomfort, characterized by baseline cramping pains with intermittent episodes of sharp pain. The patient also reported a change in bowel habits, including several episodes of diarrhea, without obvious bright red blood from the rectum. Contrast material–enhanced computed tomography (CT) showed a tubular structure with a target appearance along the course of the ascending colon (Fig 3). At the leading edge, located at the hepatic flexure, a cystic mass was seen.
**Diagnosis:** Intussusception caused by a mucinous cystadenoma of the appendix with a mucocele (a <1-cm carcinoid tumor was found at surgery).

**Discussion**
Mucocele of the appendix is a descriptive term for distension of the appendiceal lumen resulting from the abnormal accumulation of mucus, regardless of the underlying cause. These causes encompass disorders of mucosal proliferation, including both mucinous cystadenoma and cystadenocarcinoma, as well as processes that cause proximal obstruction of the appendiceal lumen. Mucoceles of the appendix may also be formed by accumulation of thick mucinous matter in patients with cystic fibrosis. Occasionally they may calcify.

Initially described and named “hydrops processus vermiformis” by Rokitansky in 1866, a mucocele may serve as a lead point for an intussusception, as in this case. The other major complication of mucoceles, depending on their underlying cause, is that of appendiceal perforation, with spillage of mucinous contents into the peritoneal cavity and leading to pseudomyxoma peritonei.

Appendiceal intussusception, itself an uncommon event, has been recognized since 1858, when it was first described by McKidd. In addition to mucoceles, a number of other causes of appendiceal intussusception have been described, including endometriomas, fecoliths, foreign bodies, polyps (e.g., juvenile polyps, inflammatory polyps related to Crohn disease), papillomas, adenoma or adenocarcinomas, and carcinoid tumors. There have also been reports of intussusception caused by a postappendectomy stump, as well as by an apparently normal appendix.
Presenting symptoms and signs may have an acute onset or be intermittent and chronic and most commonly include nausea and vomiting, abdominal pain, asymptomatic melena, or guaiac-positive stool. Other symptoms, such as an alteration in bowel habits (either constipation or diarrhea), fever, weight loss, or a palpable mass are encountered less often.

Several imaging modalities may demonstrate the classic “coiled spring” appearance of intussusception, related to telescoping of the intussusceptum into the intussuscipiens. Early on, an intussusception resembles a targetlike mass, associated with obstruction. As the wall of the outer intussuscipiens becomes more edematous, more thickening and layering can be seen. Ultimately, an amorphous mass associated with severe obstruction may develop as the bowel undergoes necrosis.

CT has been advocated as the most accurate and useful imaging study for demonstrating intussusception because it can demonstrate a central low-attenuation area and a surrounding layered or stratified pattern, caused by interspersed stripes of low and high attenuation. CT findings may also suggest the underlying cause, which is particularly important in adults. Although adults account for only 5% of all cases of intussusception, 70%–90% of adult cases are associated with an underlying lesion, including malignancy in 20%–50%. On the other hand, in infants and young children, the overwhelming majority of cases are idiopathic, without any definite lead point.

CT features suggestive of an unruptured mucocele, as was seen in this case, include a homogeneous, round or oval, low-attenuation structure with a thin uniform wall as the lead point. This appearance, however, could also be seen with intussusception caused by an enteric duplication cyst or a Meckel diverticulum. Helical CT with multiplanar reformation or MR imaging with off-axis image acquisition may allow the diagnosis of intussusception to be made with greater certainty, based on the evaluation of several planes of imaging. An additional theoretic benefit of imaging with the newer multidetector helical CT scanners is the ability to image a transient event rapidly during a single breath hold, before the intussusception either changes position or resolves.

The coiled spring appearance can also be seen at ultrasonography (US) and air contrast barium enema studies and, in the latter case, may be associated with nonfilling of the appendix. US features of a mucocele vary depending on the echogenicity of its contents.

Because intussusception is associated with a high rate of malignancy in adults, when it is detected, nonoperative reduction should not be performed. In fact, most sources agree that surgical intervention is necessary in all cases of adult intussusception, with treatment most often consisting of resection and primary re-anastomosis (depending on the location of the lesion). It is also important to avoid intraoperative reduction of the mucocele to reduce the chance of rupture or spillage into the peritoneal cavity.

**Suggested Readings**


Case 3

History and Findings
A 20-month-old girl was admitted for evaluation because of failure to thrive and jaundice. A CT scan obtained through the upper abdomen demonstrated periportal low-attenuation zones (Fig 4a). A slightly caudal image did not show a mass in the region of the pancreas nor dilatation of the common bile duct (Fig 4b). A chest radiograph demonstrated diffuse interstitial lung disease with multiple cysts, mimicking a honeycomb appearance (Fig 5). A CT section obtained through the upper chest revealed an enlarged thymus with multiple cysts and septations (Fig 6).

Diagnosis: Langerhans cell histiocytosis (LCH).

Discussion
LCH encompasses a ubiquitous group of diseases linked by the distinguishing feature of monoclonal proliferation of histiocytes. The Langerhans cell, an epidermal dendritic cell, was discovered in 1868 by Paul Langerhan. However, it was not until 1965 that its unique ultrastructural hallmark, the Birbeck granule, was seen with the aid of electron microscopy and described by Basset (Nezelof and Basset, 1998).

Langerhans cell histiocytosis encompasses a broad spectrum of diseases that at one end includes a predominantly lethal, fulminant disorder of infants known as Letterer-Siwe disease and, at the other end, solitary indolent bone lesions.
known as eosinophilic granulomas. An intermediate form of this diverse disease process, known as Hand-Schüller-Christian disease, classically manifests with diabetes insipidus, proptosis, and multifocal lytic bone lesions. Hashimoto-Pritzker disease, a self-limited congenital form of the disease, has also been described.

The clinical manifestations of this enigmatic disease vary greatly. Lesions associated with LCH have been reported in the lungs, skin, bone, pituitary gland, liver and biliary tract, lymphatic system, thyroid, and testicles. An association between thymic abnormalities and LCH has also been described. Pulmonary infiltrates are often described as cystic, but interstitial and micronodular changes have also been reported. Cutaneous abnormalities are common and consist of petechiae and yellow-brown papules. Osseous lesions are classically described as “lytic” or “punched out” and may be a solitary manifestation or associated with widespread disease. Involvement of the pituitary gland often manifests as diabetes insipidus with or without other associated hormonal imbalances. Hepatic involvement is often associated with hepatomegaly and has been linked to sclerosing cholangitis in children. Lymph node enlargement and splenomegaly also commonly occur.

Despite recent advances in the understanding of the monoclonal nature of this disease, the pathophysiologic mechanism remains largely unsettled, with ongoing debate as to the predominantly reactive or neoplastic nature of LCH. The occurrence of spontaneous remissions and lack of aneuploidy favor a reactive origin, whereas infiltration of organs by aberrant cells and monoclonality are reminiscent of a truly neoplastic process.

Typically, evaluation of a patient with suspected LCH may include blood cell count, coagulation studies, liver function tests, tests of urine osmolarity, chest radiography, and radiographic skeletal survey. A small bowel series or biopsy may be considered if malabsorption or failure to thrive is encountered. MR imaging of the hypothalamic–pituitary axis is often reserved for patients with hormonal, visual, or neurologic disturbances.

Disorders to consider in the differential diagnosis will vary widely with the clinical manifestations present in each patient. This patient presented at 19 months with failure to thrive and dyspnea. The chest radiograph demonstrated diffuse cystic lesions that were better characterized on subsequent CT scans. The CT images elegantly demonstrated the thin-walled lung cysts and also depicted low-attenuation changes within an enlarged multicystic thymus. LCH and lymphoid interstitial pneumonitis should be considered as possible diagnoses in children with cystic lung disease. On the other hand, enlargement or infiltration of the thymus strongly suggest LCH, human immunodeficiency virus–associated changes, and lymphoma for the differential diagnosis. The low-attenuation periportal abnormalities in this case were attributed to periportal edema. However, similar findings might also be associated with sclerosing cholangitis related to LCH. In summary, the diffuse cystic lung lesions, thymic abnormalities, and periportal regions with low attenuation render LCH as the most likely diagnosis.

**Suggested Readings**


Case 4

History and Findings
A 57-year-old female smoker presented with a recent onset of confusion, weight loss, and complex partial seizures. MR imaging performed with fluid-attenuated inversion recovery (FLAIR) pulse sequences showed homogeneously high signal intensity involving the left hippocampus, left parahippocampal gyrus, and amygdala, as well as diffuse enlargement of these structures (Fig 7). Positron emission tomography (PET) performed with fluorine-18 fluorodeoxyglucose (FDG) showed markedly increased tracer uptake within the medial cortex of the left temporal lobe, corresponding to the areas of signal abnormality seen on MR images (Fig 8).

Diagnosis: Limbic encephalitis associated with small cell carcinoma of the lung.

Discussion
Limbic encephalitis is a syndrome manifested by development of severe memory loss and behavioral abnormalities over the course of many weeks and is often accompanied by alterations of mood, delusions, and seizures. In the past few decades, an association between limbic encephalitis and cancer has become well established. The disorder almost always occurs in patients with an underlying malignancy, with small cell carcinoma of the lung (as in this case) being the most common (Fig 9). However, limbic encephalitis has been reported in patients with other malignancies, such as non-small cell carcinoma of the lung,
thymoma, testicular carcinoma, and breast carcinoma. On occasion, clinical features of limbic encephalitis can precede discovery of an underlying neoplasm. In rare instances, an underlying malignancy is never found.

Establishing the diagnosis of limbic encephalitis can be exceedingly difficult on the basis of clinical and laboratory findings alone, particularly in cases in which a neoplasm has not been previously diagnosed. The clinical features can be mimicked by a wide spectrum of metabolic, infective, and neoplastic conditions. Laboratory findings are often normal and, when abnormal, are nonspecific. The presence of antineuronal autoantibodies within cerebrospinal fluid can be a helpful finding.

Histologic changes associated with limbic encephalitis include an inflammatory reaction with a perivascular lymphocytic infiltrate, neuronal loss, gliosis, and, subsequently, Wallerian degeneration. Gray matter structures are more commonly affected than white matter structures. The cause of the histologic and clinical findings has not been fully determined, but an autoimmune phenomenon directed against tumor antigens bearing a similarity to antigens expressed by cells of the nervous system is a leading consideration.

The presence of antineuronal antibodies in many cases is taken as evidence supporting the autoimmune hypothesis, and they may play a role in pathogenesis by reaction with brain tissue.

Imaging studies have become increasingly important in the diagnosis of limbic encephalitis; in the appropriate clinical setting, imaging studies can preclude the need for brain biopsy. CT findings are often normal but occasionally reveal a low-attenuation lesion in the medial aspect of one or both temporal lobes. T2-weighted MR images have shown abnormal areas of high signal intensity in one or both temporal lobes in the acute phase of limbic encephalitis. In the acute and subacute periods, PET imaging has been reported to show increased FDG uptake in the temporal lobe (Fig 8), whereas in the chronic phase, it has shown decreased temporal lobe uptake.

Treatment of the underlying neoplastic process appears to have more effect on the neurologic outcome than treatment with immune modulation. However, only a minority of cases has shown neurologic improvement with either treatment. In rare cases, the encephalopathy resolves after successful treatment of the underlying malignancy.

**Suggested Readings**


Case 5

History and Findings
A 57-year-old man presented with intermittent cough and fever. Chest radiography revealed air-filled cysts in both lung apices (Fig 10a). Some of these cysts contained nodular opacities. Additional findings included acute angulation of the right posterior third rib, narrow right posterior third and ninth ribs, short segment dextroscoliosis of the superior thoracic spine, and multiple scattered well-defined subcutaneous nodules (Fig 10). CT showed bilateral apical cavities with thin irregular walls and rounded masses in dependent position (Fig 11).

Diagnosis: Neurofibromatosis complicated by cystic lung disease and aspergillomas.

Discussion
Neurofibromatosis type 1 (von Recklinghausen disease) is the most common of the phakomatoses, occurring with an incidence of 1:2000 to 1:3000. Common clinical findings of the disease include “café au lait” spots and subcutaneous neurofibromas. The central nervous system (CNS) is prominently involved with a variety of tumors; however, seizures and frank mental retardation occur in less than 10% of patients. Respiratory symptoms are usually mild, with dyspnea upon exertion being the most common. When results of pulmonary function tests are abnormal, they usually indicate the presence of obstructive disease but restrictive disease may be present. Diffusion capacity may be decreased. Various other symptoms have been described.

The thorax is involved in multiple ways that are evident radiologically. Linear areas of opacity with lower lung predominance are the usual radiographic finding, and honeycomb formation may be seen. Pulmonary fibrosis has been seen on chest radiographs in up to 20% of patients over the age of 30 years with neurofibromatosis type 1, but it is uncommon in younger patients. Large, thin-walled bullae with asymmetric upper lobe predominance almost always accompany pulmonary fibrosis. However, bullae without fibrosis may also be seen.
Subcutaneous neurofibromas often appear as well-defined, rounded nodules on chest radiographs. When these masses project over the lung, they may be mistakenly interpreted as pulmonary nodules. Plexiform neurofibromas may cause erosion of rib margins or separation of rib interspaces, and they may also involve the mediastinum. Neurofibromas involving the nerve roots may enlarge the neural foramina and may extend intra- and extraspinal, causing a dumbbell shape. Malignant degeneration of these neurogenic tumors has been reported in 1%–29% of patients with neurofibromatosis type 1. Imaging findings suggestive of malignant degeneration include a sudden change in size of the preexisting mass or development of heterogeneous attenuation or signal intensity on CT or MR images. Bone deformity, including short segment scoliosis and angulation and narrowing of ribs, may be related to primary dysplastic changes or pressure from adjacent tumors. Posterior scalloping of the thoracic vertebral bodies may be caused by dural ectasia. Lateral meningoceles may form, protruding through the enlarged neural foramina, and appear as a large mediastinal mass.

It is not surprising that fungus balls infected the apical bullae in this patient with neurofibromatosis, since they may form in a cavitary space of any cause. One of the most common causes of cavities, however, is post primary tuberculosis: 20%–55% of patients with a fungus ball have a history of tuberculosis, and 17% of patients with tuberculosis develop an aspergilloma. Other disorders that may form cavities permitting formation of fungus balls include sarcoidosis, bronchiectasis, abscess, lung trauma or surgery, ankylosing spondylitis, other causes of pulmonary fibrosis, pulmonary carcinoma, and bullous emphysema.

A fungus ball is composed of fungal hyphae, mucus, and cellular debris located within a lung cavity. A fungus ball is usually caused by the Aspergillus species, especially A. fumigatus, in which case it may be called an aspergilloma. Aspergillomas are often mobile within the cavity and rarely invade the wall. Similar clinical and radiographic appearances may result from other organisms, including Candida albicans, Pseudallescheria boydii, Coccidioides immitis, Nocardia species, Actinomyces species, and others.

Patients are frequently asymptomatic; however, cough and expectoration may occur. Up to 95% of patients will experience hemoptysis of varying amounts at some time. Life-threatening hemoptysis is the major serious sequela of this disease.

Radiologic appearance of fungus balls in the appropriate clinical setting is often sufficient for diagnosis. Transthoracic needle aspiration biopsy or bronchial washings may allow cytologic or culture confirmation; however, isolation from culture is often difficult and contamination may be a problem. Supportive serologic studies include a positive precipitin test to Aspergillus antigen, elevated Aspergillus-specific IgE, IgG-ELISA, and polymerase chain reaction identification of Aspergillus.

The classic appearance at chest radiography and CT is a round or oval mass located in the dependent portion of a lung cavity. Interval thickening of a cavity wall may indicate impending formation of a fungus ball. The air outlining the nondependent margin of the fungus ball has a crescent shape (air crescent sign). Mobility is confirmed by imaging the patient in a second position such as decubitus or prone. Large mycetomas may fill the entire cavity and obliterate the air crescent. CT scans may demonstrate aspergillomas not seen on radiographs. At CT, a sponge-like appearance of the aspergilloma may be seen.

An air crescent sign is most often associated with a fungus ball; however, this appearance can occur in other abnormalities, including invasive aspergillosis, echinococcal lung cyst, lung abscess with or without pulmonary gangrene, tuberculoma, blood clot in a tuberculous cavity, Rasmussen aneurysm, primary and metastatic lung carcinoma or sarcoma, bronchial adenoma, cystic hamartoma, and pulmonary hematoma.

Whether or not aspergillomas should be treated is controversial, although patients with moderate hemoptysis are at risk of severe hemorrhage, and definitive surgical resection should be considered. Patients with poor operative risk may receive conservative therapy including antifungal medication; however, this treatment has not always been successful. Success with an intracavitary infusion of medication such as amphotericin B or ketoconazole from percutaneous or bronchoscopically placed catheters is anecdotal. Overall, aspergillomas spontaneously resolve in 10% of patients; hence, it has been suggested that no therapy is needed for patients with no or mild symptoms.

**Suggested Readings**


Figure 12. Case 6.

Figure 13. Case 6.

Case 6

**History and Findings**

A 79-year-old man presented with pain of several years duration in both knees and lower legs. The anteroposterior plain radiograph of both knees revealed patchy sclerosis in the distal femurs and the proximal tibias that was diametaphyseal in distribution (Fig 12). The radionuclide bone scan showed markedly increased uptake bilaterally and symmetrically in the diametaphyseal regions around the knees and in the distal diametaphyses of the tibias (Fig 13). The proximal femurs demonstrated a lesser degree of increased uptake. T1-weighted (Fig 14a) and T2-weighted (Fig 14b) MR images of the femurs showed low signal intensity throughout the marrow, corresponding to the areas of abnormality seen on the radiograph and the areas of increased uptake on the bone scan.

**Diagnosis:** Lipogranulomatosis (Erdheim-Chester disease).

**Discussion**

The symmetric distribution of the disease process limits the possibilities in the differential diagnosis considerably. Metastatic disease and infection would not have a symmetric distribution, and the time course of the patient’s symptoms makes these possibilities unlikely as well. Infarcts might be considered on the basis of radiographic findings; however, the findings from bone scintigraphy and MR imaging should exclude infarcts. Less common entities to consider would include sclerotic myeloma. However, the bone scan appearance is not consistent with that of myeloma, and the symmetric distribution around the knees would be unusual. An uncommon process with this radiographic appearance that should be considered is Erdheim-Chester disease or lipogranulomatosis.
Erdheim-Chester disease is a non–Langerhans cell histiocytosis. Unlike the Langerhans type of histiocytosis (eosinophilic granuloma) that occurs almost exclusively in younger patients (<30 years of age), Erdheim-Chester disease occurs in older patients. It is a disseminated disease involving viscera, mesenchymal tissue, and bone. The long bones are involved primarily, with sparing of the axial skeleton. The epiphyses are usually spared, as in this case. The cause is unknown. Erdheim-Chester disease is characterized by involvement of multiple organs with slow progression and a relatively high mortality rate. Diagnosis can be made with imaging and confirmed from results of bone biopsy.

**Suggested Readings**


**Case 7**

**History and Findings**

A 25-year-old woman presented with complaints of fatigue, easy bruising, and right-sided abdominal pain. She also had a history of portal vein thrombosis. She underwent MR imaging of her abdomen before (Fig 15a, 15b) and after (Fig 15c) gadolinium-chelate administration. The images revealed ascites, a marked decrease in the signal intensity of the renal cortex, small or nondiscernible hepatic veins, and a mottled enhancement pattern in the periphery of the liver.

**Diagnosis:** Paroxysmal nocturnal hemoglobinuria (PNH) complicated by Budd-Chiari syndrome (BCS).

**Discussion**

PNH was first described by Paul Strubing in 1882 as a syndrome with hemoglobinuria after sleep. It is an acquired disorder of hemopoietic stem cells, characterized by an increased sensitivity to complement-mediated lysis, which affects erythrocytes, granulocytes, and platelets. Manifestations of the disease include intravascular
hemolysis without gross hemoglobinuria, pancytopenia or aplasia, increased susceptibility to infections, and venous thrombosis. Venous thromboses, a major cause of death in patients with PNH, form at uncommon sites such as the cerebral, mesenteric, and hepatic veins. Many patients with severe hemoglobinuria develop tubulo-interstitial nephritis, which may progress to fatal uremia.

MR imaging may be used to evaluate patients with PNH to demonstrate iron overload and assess the abdominal vasculature. In patients with PNH, the signal intensity of the renal cortex is usually low on T1- and T2-weighted images because of hemosiderin deposited in the proximal convoluted tubules secondary to intravascular hemolysis. In contrast to patients with other hemolytic anemias, patients with PNH usually have decreased iron concentration in the liver and spleen. However, patients with transfusional hemosiderosis may also have diffuse low signal intensity in the spleen and liver.

PNH has been reported as a cause of BCS in up to 12% of cases. This syndrome of hepatic venous outflow obstruction was first described by Budd in 1845, and the associated histologic features were first reported by Chiari in 1899. Patients with acute BCS have hepatomegaly, right upper quadrant abdominal pain and ascites, whereas patients with chronic BCS present with signs and symptoms of portal hypertension. BCS occurs in men and women of all ages. It may be idiopathic or associated with the use of oral contraceptives. It may also occur as a complication of an illness associated with thrombosis, such as polycythemia rubra and PNH. Results of liver functions tests are nonspecific, with an elevated level of hepatic transaminases indicating hepatic injury.

Doppler US is the noninvasive imaging modality of choice for diagnosing BCS. Features on Doppler US include decreased hepatic venous blood flow and pulsatility, thrombosis of one or more hepatic veins or the inferior vena cava, and caudate lobe hypertrophy. Doppler US has a sensitivity of 85%–100% and a specificity of 85% for BCS. At hepatic venography, which is the standard of reference for diagnosing BCS, the major hepatic veins are not usually visualized and a characteristic spider web pattern of collateral and recanalized vessels is seen. Often, the inferior vena cava is markedly compressed from diffuse enlargement of the liver or from a hypertrophied caudate lobe with a resultant high pressure gradient between the infrahepatic and suprahepatic portions of the inferior vena cava.

MR imaging has sensitivity and specificity similar to those of Doppler US in the diagnosis of BCS. Its use is recommended when doubt exists after sonographic examination or when the patient’s habitus limits a complete US evaluation. The MR imaging examination includes use of T2-weighted fat-suppressed fast spin-echo pulse sequences and T1-weighted gradient-echo sequences, both before and after intravenous administration of a gadolinium chelate. In acute BCS, the peripheral liver parenchyma has moderately low signal intensity on T1-weighted images and moderately high signal intensity on T2-weighted images relative to the central portion of the liver. Early gadolinium-enhanced images show diminished and mottled peripheral enhancement of the liver. In chronic BCS, the differences in signal intensity between the peripheral and central portions of the liver are minimal on T1- and T2-weighted images. Enhancement differences of the liver parenchyma are also minimal in chronic BCS. Other MR imaging features include absence of flow within the hepatic veins, intrahepatic collateral vessels, enlargement of the caudate lobe, and ascites.

CT is a second line investigation for BCS. On contrast-enhanced CT scans, BCS has a characteristic “flip-flop” enhancement pattern. On early images, the central liver enhances prominently, while the peripheral liver enhances weakly. On delayed images, the periphery of the liver is enhanced, while contrast material has washed out of the central liver. The caudate lobe is characteristically enlarged and enhances normally because of its separate venous drainage into the inferior vena cava. If the cause of obstruction is suprahepatic, the caudate lobe may not be enlarged. Comma-shaped intrahepatic collateral vessels may be seen on CT or MR images (the comma sign).

Treatment of BCS may be medical or surgical. Nonsurgical approaches include the control of ascites with diuretics and sodium restriction, anticoagulation, thrombolytic therapy, angioplasty, and placement of a transjugular intrahepatic portosystemic shunt. Patients with cirrhosis or significantly impaired liver function or patients with acute BCS who have failed shunt therapy are candidates for orthotopic liver transplantation. There have, however, been a number of reports of recurrent BCS in patients with PNH following liver transplantation.
Suggested Readings

Case 8

History and Findings
A 12-year-old boy presented with rectal bleeding and complaints of difficulty in swallowing solid foods. An upper gastrointestinal series with small bowel follow-through demonstrated several, small (less than a centimeter) nodular filling defects throughout the small bowel (Fig 16). T2-weighted MR images obtained through the abdomen showed multiple high-signal-intensity lesions in the small bowel and in the left multifidus muscle (Fig 17). In the liver, an additional small high-signal-intensity lesion was seen in the anterior segment of the right lobe. CT (Fig 18) and T2-weighted MR (Fig 19) images obtained through the mandible demonstrated a 2-cm high-signal-intensity mass with a single calcification in the right submandibular region.

Diagnosis: Blue rubber bleb nevus syndrome.

Discussion
Blue rubber bleb nevus syndrome is a rare disorder characterized by distinctive cutaneous vascular malformations associated with similar lesions of the gastrointestinal tract and other viscera. Although Gascoyen first described an association between cutaneous vascular lesions and gastrointestinal bleeding in 1860, the term blue rubber bleb nevus syndrome was not coined until 1958 when Bean reported a case with similar findings.

The principal clinical manifestations of this syndrome include vascular hamartomas of the skin and visceral hemangiomas predominantly affecting the gastrointestinal tract. Visceral lesions have been reported in other tissues, including the liver, spleen, skeletal muscle, heart, lung, kidney, thyroid, eyes, and CNS. The cutaneous vascular malformations are distinctive in that they are red to deep blue, soft, typically painless, and readily evacuated with pressure (the remaining empty sac slowly fills again). These skin lesions are commonly present from birth but may increase in size and number with age. The majority of reported cases appears to be sporadic; however, an autosomal dominant form of the disease with variable expressivity has been reported. The gastrointestinal lesions are prone to spontaneous hemorrhage and often result in iron deficiency anemia. These lesions have also been associated with intussusception and volvulus.

In addition to cutaneous and visceral vascular malformations, patients with blue rubber bleb nevus syndrome may suffer from orthopedic complications such as pressure erosion from a juxtaposed vascular lesion or osseous and soft-tissue hypertrophy secondary to associated hyerpervascularity.

Histopathologic evaluation of these lesions reveals a thin layer of connective tissue and a single layer of endothelial cells surrounding blood-filled ectatic vessels of variable size.

Although these vascular lesions may be evaluated with a variety of imaging modalities, MR imaging has been an effective tool for noninvasive assessment of the number and extent of both cutaneous and visceral lesions. The lesions typically demonstrate high signal intensity on T2-weighted MR images, an appearance that is attributed to a slow-flow phenomenon or thrombosis within the ectatic vessels.

Disease processes to be considered in the differential diagnosis include uncommon syndromes associated with cutaneous and soft-tissue vascular lesions and two of the polyposis syndromes. Maffucci syndrome is a disorder of dyschondroplasia and osteochondromas associated with vascular malformations. Klippel-Trénaunay-Weber syndrome includes port wine stain, vascular malformations, and secondary limb hypertrophy. Kasabach-Merritt syndrome is related to consumptive coagulopathy associated with large vascular malformations. Kaposi sarcoma often involves the skin and gastrointestinal tract but is not associated with true venous malformations. Peutz-Jeghers syndrome is a congenital polyposis syndrome associated with melanotic cutaneous lesions. Likewise, Gardner syndrome may be associated with soft-tissue tumors and sebaceous cysts.
Suggested Readings

Case 9

History and Findings
A 1-year-old girl with a history of complex partial seizures was found to have numerous hyperpigmented cutaneous lesions on her trunk. An unenhanced axial CT image demonstrated bilateral high-attenuation foci in the anterior temporal lobes and mild, diffuse high attenuation in the cerebellum (Fig 20). An unenhanced coronal T1-weighted MR image showed homogeneous, abnormal high signal intensity in the cerebellum (Fig 21). Both CT and MR images also revealed hydrocephalus and prominent cerebrospinal fluid spaces surrounding the cerebellum.

Diagnosis: Neurocutaneous melanosis with an associated Dandy-Walker variant.

Discussion
Neurocutaneous melanosis is a rare congenital syndrome characterized by the presence of large or multiple congenital melanocytic nevi and melanotic lesions of the CNS. The melanotic CNS lesions can arise from benign melanosis of the leptomeninges and parenchyma or from malignant melanoma, although the distinction between benign and malignant lesions is often not possible on the basis of imaging findings alone. Most patients present within the first 2 years of life with neurologic manifestations of increased intracranial pressure, seizures, ataxia, and occasionally cranial nerve VI and VII palsies. Less commonly, patients present in the 2nd or 3rd decade of life. The syndrome occurs sporadically with equal representation among the sexes. An
estimated 40%–60% of patients eventually develop CNS melanomas, and 5%–15% develop cutaneous melanomas. The course of the disease is usually one of rapid deterioration, with death occurring within 3 years of diagnosis in the majority of patients, due to either development of malignant melanoma or to complications of hydrocephalus. Most patients who undergo palliative surgery to relieve increased intracranial pressure experience only transient improvement, and antineoplastic chemotherapy does not significantly improve symptoms or alter disease course. The prognosis is grave even when there is no evidence of associated malignancy.

Melanotic cells (which include melanocytes and melanin-containing macrophages) are normally found in the basilar leptomeninges. In neurocutaneous melanosis, the concentration of melanotic cells in their normal location is markedly increased, with concomitant cell infiltration into the perivascular spaces. This abnormal abundance of melanocytes in the basilar leptomeninges has been attributed to abnormal migration of melanocyte precursors (which are neuroectodermal in origin), abnormal expression of melanin-producing genes within leptomeningeal cells, and rapid proliferation of melanin-producing leptomeningeal cells. CNS parenchymal melanosis is thought to be caused by either primary migration of melanotic cells early in development or subsequent secondary spread from the perivascular spaces. Because benign melanotic cells can show cellular pleomorphism, nestlike accumulations, and occasional atypia, differentiation of benign from malignant lesions is difficult even at histologic analysis.

The typical radiologic findings in neurocutaneous melanosis are foci of abnormally thickened leptomeninges, which have high attenuation on CT scans, high signal intensity on T1-weighted images, and low signal intensity on T2-weighted MR images. The appearances on CT and MR images are presumed to be caused by melanin pigments, which are relatively dense and contain oxygen-free radicals which have a paramagnetic effect. Contrast material enhancement on MR images is unusual but has been reported. Common locations for leptomeningeal melanosis are (a) on the inferior surfaces of the cerebellum and frontal, temporal, and occipital lobes and (b) at the ventral aspects of the pons, cerebral peduncles, and upper cervical spinal cord. These sites correspond to areas of physiologic melanocytic distribution. Parenchymal melanosis is less common than leptomeningeal melanosis but has the same imaging characteristics and tends to occur in the cerebellum and anterior temporal lobes, especially in the amygdala. When a frankly hemorrhagic, necrotic, or invasive mass is seen, the presence of malignant melanoma can be relatively confidently diagnosed. However, these imaging characteristics are usually not present, making it difficult to differentiate benign melanosis from malignant melanoma on MR images. Therefore, serial follow-up studies or biopsy is necessary to assess for malignancy.

Other imaging findings of neurocutaneous melanosis in the CNS include hydrocephalus, posterior fossa cysts, cerebellar hypoplasia, Dandy-Walker malformation, syringomyelia, intraspinal arachnoid cysts, and intraspinal lipoma. These associated abnormalities suggest a defective ectodermal-mesodermal interaction as the common etiologic mechanism for both abnormal melanocytic accumulation and developmental anomalies of the hindbrain and spinal canal. The frequent presence of hydrocephalus may be attributable to obstruction of cerebrospinal fluid flow, decreased absorption of cerebrospinal fluid by thickened meninges, or the Dandy-Walker complex.

**Suggested Readings**


**Case 10**

**History and Findings**

A 37-year-old woman with a chronic illness complained of a recent increase in sputum production and intermittent fever. Previously, she had undergone left lower lobectomy and tracheostomy as part of her treatment. Representative
posteroanterior and lateral radiographs showed a tracheostomy tube in good position and evidence of the prior lobectomy with compensatory hyper-inflation of the remaining left lung (Fig 22). Extensive, thick, branching tubular opacities suggestive of mucoid impaction were present in the right lower lobe.

Despite intermittent antibiotic treatment, she continued to have recurrent pneumonias, so she was admitted for intravenous administration of antibiotics. CT demonstrated large oblong and round cystic lesions with air-fluid levels that proved to be saccular bronchiectasis (Fig 23a). Thin-section (3 mm) CT through the trachea demonstrated a 5-mm nodule on the nondependent, anterior surface of the trachea (Fig 23b).
**Diagnosis:** Tracheobronchial papillomatosis.

**Discussion**

Tracheobronchial papillomatosis is a rare complication of laryngeal papillomas, occurring in only 5%–10% of a large series, with extension to the lung in less than 2% of cases. Most of these cases occur in childhood and are rarely manifested for the first time in the adult years. When bronchopulmonary disease occurs, it appears an average of 10 years after the initial diagnosis of laryngeal papillomas. Laryngeal papillomas are often acquired at birth from exposure to the human papilloma virus (HPV) during passage through the birth canal. HPV 6 and 11 are the most commonly associated forms, although other strains have been identified in rare cases. Most often, these lesions are discovered because of symptoms of airway compromise. Treatment consists of laser fulguration and debulking, as was performed in this case. Occasionally, a tracheostomy is required to maintain airway patency.

It is generally accepted that tracheobronchial papillomatosis results from aerial dissemination of laryngeal disease. The need for recurrent laser treatment or tracheostomy appears to be associated with a higher rate of distal spread. This is thought to be caused by repeated trauma, with transmission of infected material through the airways. Whether these cases also include patients with higher viral loads is unclear. The proximal bronchi and trachea usually deal with disseminated material through the action of the mucociliary transport system. In airways with few or no cilia, however, cells may lodge in the bronchi and proliferate. Other theories on the development of tracheobronchial papillomatosis have included contiguous extension of disease, diffuse viral contamination, and multicentric origination of papillomas. These theories are not well supported by clinical data and the temporal progression of the disease.

Tracheobronchial papillomatosis should be suspected when a patient with prior laryngeal papillomas presents with chronic cough, hemoptysis, or recurrent pneumonias.

Chest radiographs may be normal with laryngeal papillomas. Occasionally, nodules may be evident within the airways on high kilovoltage studies. With dissemination of disease to the lower airways, findings include solid or cavitory nodules. With increased growth, blood supply is interrupted, resulting in central cavitation. Endobronchial growth can eventually lead to saccular bronchiectasis and mucus plugging, as was seen in this case. Obstructive atelectasis is unusual.

CT demonstrates the lesions to better advantage and may reveal them even when the chest radiograph appears normal. Cavitary nodules tend to have thin walls (2–3 mm), and they may have air-fluid levels if superinfected. Over time, the nodules may coalesce and form larger cavitory masses. Bronchiectasis is also better delineated with CT.

The overall prognosis for patients with tracheobronchial papillomatosis is poor. Although interferon may occasionally be effective, there is no optimal treatment once the disease becomes disseminated. Progression of disease eventually leads to respiratory failure and death. Spontaneous regression has not been seen. Longer survival has been seen in patients with fewer lesions. Patients with tracheobronchial papillomatosis are also at increased risk for squamous metaplasia and development of squamous cell carcinoma. This risk is more frequently associated with HPV 11 and 16 but has also been seen with the more common HPV 6.

**Suggested Readings**

Fraser RS, Müller NL, Colman N, Pare PD. Fraser and Pare’s diagnosis of diseases of the chest. 4th ed. Philadelphia, Pa: Saunders, 1999; 1262–1263.

