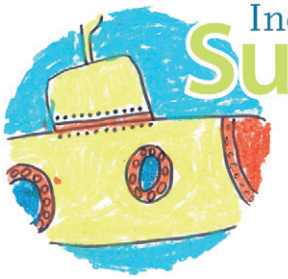

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4 Worsening Abdominal Distention in a 2-year-old Boy

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PRESENTATION

A previously healthy 2-year-old boy presents to the emergency department with a history of abdominal distention that began 1 week ago and has since been worsening. He is toilet-trained and his family mentions that he has been having intermittently decreased urination for the past month. Some days he only urinates once or twice. He has had no fever, emesis, joint pain, anorexia, weight loss, or decreased activity. The boy's medical history and family history are unremarkable.

On physical examination, he has normal vital signs. He appears well and has normal physical examination findings, with the exception of his gastrointestinal evaluation. His abdomen is soft but with prominent swelling in the right upper quadrant and epigastric regions. In the area of swelling, there is a smooth, nontender, palpable mass without overlying erythema. A liver edge is not palpable because the mass is overlying that area, but a spleen tip is noted at the left costal margin.

Initial laboratory results include:

- White blood cell count, 14,000/μL (14.0 $\times$ 10^9/L)
- Absolute neutrophil count, 5,100/μL (5.1 $\times$ 10^9/L)
- Absolute lymphocyte count, 7,900/μL (7.9 $\times$ 10^9/L)
- Blood smear revealing occasional anisocytosis
- Hemoglobin, 11.1 g/dL (111 g/L)
- Platelet count, 329 $\times$ 10^3/μL (329 $\times$ 10^9/L)
- Aspartate aminotransferase, 47 U/L (0.78 μkat/L) (reference range 0-37 U/L [0-0.62 μkat/L])
- Alanine aminotransferase, 31 U/L (0.52 μkat/L) (reference range 0-78 U/L [0-1.30 μkat/L])
- Lactate dehydrogenase, 727 U/L (reference range 100-240 U/L)
- Uric acid, 4.2 mg/dL (249.84 μmol/L) (reference range 3.5-7.2 mg/dL [208.20-428.29 μmol/L])
- Urea nitrogen, 18 mg/dL (6.3 mmol/L)
- Creatinine, 0.28 mg/dL (24.8 μmol/L)

The remainder of the basic electrolyte panel, the coagulation profile, and urinalysis are interpreted as normal. The patient is hospitalized and additional evaluation leads to the diagnosis.

The Case Discussion and Suggested Readings appear with the online version of this article at http://pedsinreview.aappublications.org/content/38/1/49.

DISCUSSION

Abdominal ultrasonography revealed a large heterogeneous mass suspected to originate from the liver. Abdominal magnetic resonance imaging (MRI) further defined the mass to be largely involving the left hepatic lobe (Fig). Additional laboratory results included ferritin measuring 38 ng/mL (85.39 pmol/Lng/mL) (reference range 22-322 ng/mL [49.43-723.53 pmol/Lng/mL]) and negative urine vanillylmandelic acid and homovanillic acid. Biopsy of the mass established the diagnosis of an embryonal rhabdomyosarcoma. The patient underwent a gross total resection of the mass and had multiple lymph node biopsies and a bone marrow biopsy that were negative. He received 22 weeks of chemotherapy with vincristine, dactinomycin, and cyclophosphamide. He has been off therapy for 12 months and is doing well.

Differential Diagnosis

A palpable abdominal mass is 1 of the more common findings of malignant solid tumors in children. Neuroblastoma and Wilms tumor are the 2 most common intra-abdominal tumors. For children younger than age 2 years, neuroblastoma is the most common extracranial malignant tumor. Wilms tumor is the most common renal malignancy and two-thirds of cases are diagnosed before age 5 years. Leukemia, lymphoma, hepatoblastoma, and soft-tissue sarcomas are more prevalent in older children.

The Condition

Sarcoma is a relatively rare malignant tumor that arises from embryonic mesenchymal cells and has the ability to differentiate into striated skeletal and smooth muscle, bone, cartilage, and adipose tissue. Rhabdomyosarcoma (RMS) is the most common soft-tissue sarcoma, accounting for 3% of all pediatric tumors. Given its origin in the embryonal mesenchyme, this disease has the potential to arise anywhere in the body. Most cases are sporadic. Several environmental factors have been implicated with an increased risk for RMS, including cigarette smoking, advanced maternal age, radiation exposure in utero, and antibiotic use. Inherited familial syndromes, such as neurofibromatosis and Li-Fraumeni syndrome, also have some association with RMS.

The presentation depends on the site of the tumor and whether it involves any metastasis. The head and neck region is the most common site, accounting for about one-third of total cases, followed by the genitourinary (GU) tract (25%) and extremities (20%). Sites that are involved within the head and neck include the orbit and parameningeal sites (middle ear, nasopharynx, paranasal sinuses) and can present with proptosis or obstruction of the parameningeal region. GU tract tumors can involve the bladder, prostate, or vagina and present with hematuria, outflow obstruction, or urinary frequency. Extremity involvement typically presents with obvious swelling and has a higher likelihood of spreading to regional lymph nodes.

Initial evaluation involves plain radiographs of the area surrounding the mass, followed by computed tomography (CT) scan and/or MRI. Laboratory evaluation includes complete blood cell count with smear, electrolytes, liver function tests, uric acid, and calcium. Definitive diagnosis requires biopsy and special staining performed by a pediatric pathologist. If there is suspicion of metastasis based on the site of the primary tumor, further imaging should be performed with CT scan or radionuclide bone scan.

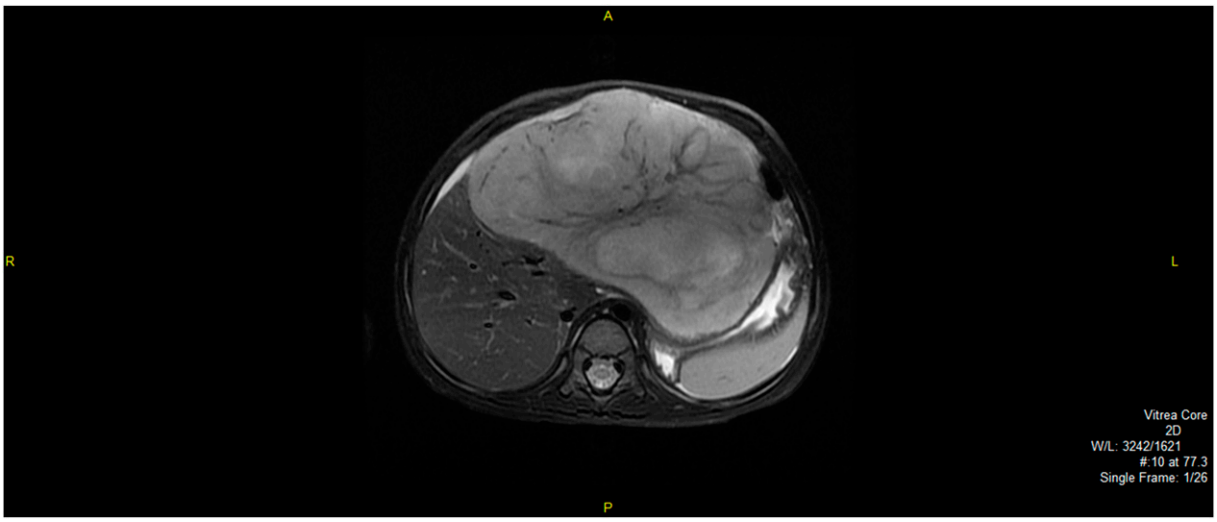

Figure. Abdominal magnetic resonance imaging shows a mass largely involving the left lobe of the liver.

Management

Current treatment is based on the recommendations of the Intergroup Rhabdomyosarcoma Study Group (IRSG), which began in 1972. The most current protocol is the IRSG-V, which achieves a cure rate of 70% in children with localized disease. Complete surgical excision is recommended for local disease if it is possible to achieve appropriate functional and cosmetic results. Tumors that are in areas such as the orbit, vagina, or bladder may require induction chemotherapy to decrease the tumor burden to allow for acceptable excision. Following surgical excision, radiation therapy enhances the treatment of residual microscopic disease. All patients with RMS should subsequently undergo chemotherapy with the gold standard regimen of vincristine, dactinomycin, and cyclophosphamide. Patients who relapse after the initial treatment have a poor prognostic outcome.

Lessons for the Clinician

- Rhabdomyosarcoma should be in the differential diagnosis for children with enlarging masses anywhere in the body.
- Plain films are the appropriate initial imaging study, followed by computed tomography scan and/or magnetic resonance imaging.
- Biopsy is required to obtain the definitive diagnosis.
- Prompt therapy should be initiated with a combination of surgical excision, chemotherapy, and radiation therapy, depending on the tumor size and affected area.

Suggested Readings

McCarville MB, Spunt SL, Pappo AS. Rhabdomyosarcoma in pediatric patients: the good, the bad, and the unusual. *AJR Am J Roentgenol.* 2001;176(6):1563–1569

Perez EA, Kassira N, Cheung MC, Koniaris LG, Neville HL, Sola JE. Rhabdomyosarcoma in children: a SEER population based study. *J Surg Res.* 2011;170(2):e243–e251

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