ELSEVIER

Seminars in
Pediatric
Neurology

# A 15-Month-Old Girl Presenting With Clitoromegaly and a Chest Mass

Courtney Darcy, PNP-BC, and Nicole J. Ullrich, MD, PhD

**A 15-month-old girl was initially referred for endocrine evaluation for clitoromegaly and subsequently found to have an adrenal mass that tracked along the paravertebral region and was associated with increased vascular markings along the skin. Neurologic examination was normal. Magnetic resonance imaging of the chest mass demonstrated a serpiginous lesion along the intercostal margins. Initial differential diagnosis included neuroblastoma, ganglioneuroblastoma, vascular lesion, or nerve sheath tumor. Biopsy was consistent with plexiform neurofibroma. Subsequent examination revealed features consistent with neurofibromatosis type 1. A review of clinical features of NF1 is provided in the case report.**
**Semin Pediatr Neurol ▮:▮▮▮-▮▮▮** 

## Case

A 15-month-old girl initially presented for endocrine evaluation regarding congenital clitoromegaly. She was healthy except for puffy genitalia noted at birth. Clinical examination was suggestive of possible testosterone excess with clitoromegaly, and she also had an increased anogenital ratio. For these reasons, pelvic ultrasound and laboratory studies were performed. Pelvic ultrasound showed normal uterus and ovaries and question of soft tissue abnormality in the right suprarenal region. Clinical workup and short-term interval follow-up ultrasound were recommended. Serum electrolytes, leutinizing hormone, follicle stimulating hormone, estradiol, and testosterone were performed and were normal; testosterone level was not measureable. She had a normal karyotype. These results were felt to rule out the most common forms of congenital adrenal hyperplasia that causes masculinization of female fetuses.

Short-interval abdominal ultrasound was performed several weeks later and demonstrated a somewhat amorphous, heterogeneously hypoechoic soft tissue structure in the right suprarenal area, without discernible internal vascularity. Subsequent magnetic resonance imaging (MRI) of the abdomen demonstrated a predominantly high T2 signal lesion in the region of the right adrenal gland extending across the midline and into the expected location of the left adrenal gland. The lesion additionally was thought to track along the celiac and superior mesenteric arteries. An additional high T2 signal abnormality was identified along the bilateral thoracic paravertebral regions, right greater than left, in the expected location of the sympathetic chain that extends bilaterally along the ribs, and into the right paravertebral soft tissues (Fig. 1). Postcontrast images demonstrated mild heterogeneous enhancement. Given the constellation of findings, a neoplastic process was suspected. Differential diagnosis at that time included neuroblastoma or ganglioneuroblastoma. Additionally, lymphoma was considered, especially given the finding of a mass crossing the midline. Given the tubular-appearing T2 structures within the right paravertebral soft tissues, the differential diagnosis also included an atypical vascular anomaly. On more detailed review of the existing imaging, plexiform neurofibroma was added to the differential.

Subsequent positron emission tomography scan showed mild fludeoxyglucose (FDG) uptake corresponding to the focal soft tissue nodularity in the right paraspinal region at T6 and more laterally in the sixth right intercostal space. The retroperitoneal soft tissue mass anterior to the crura at the level of T12 and L1, seen on computed tomography and MRI, showed very low-level uptake of FDG, similar to blood pool. Soft tissue nodularity that had been seen on MRI, in the left thoracic paraspinal region, tracking along the lower left intercostal spaces and in the lower right intercostal spaces, did not show definite increased FDG avidity. Overall, the constellation of findings on positron emission tomography, in conjunction with MRI and computed tomography from the same day,

Department of Neurology, Boston Children's Hospital, Boston, MA.
Address reprint requests to Nicole J. Ullrich, MD, PhD, Department of Neurology, Boston Children's Hospital, 300 Longwood Ave., Boston, MA 02115. E-mail: nicole.ullrich@childrens.harvard.edu

http://dx.doi.org/10.1016/j.spen.2017.03.020
1071-9091/11/

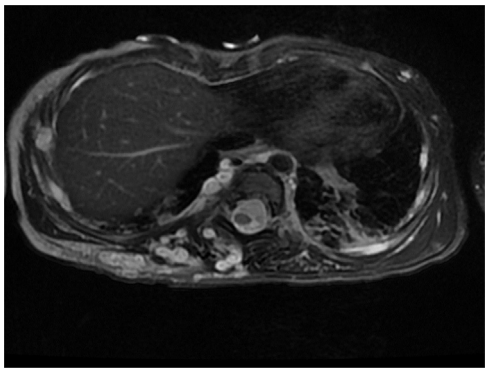

**Figure 1** Axial MR imaging through the abdomen demonstrates high T2 signal abnormality along bilateral thoracic paravertebral regions along the sympathetic chain, which extend bilaterally along the ribs, right greater than left and into the right paravertebral soft tissues. MR, magnetic resonance.

suggested the possibility of nerve sheath tumor without convincing evidence of malignant degeneration or FDG avid malignancy.

Percutaneous needle biopsy was performed on the soft tissue mass along the right posterior chest wall. Pathologic evaluation revealed schwannian proliferation and tactoid bodies, consistent with plexiform neurofibroma. The lesion involved a neural ganglion. The patient was then referred for evaluation of possible neurofibromatosis type 1 (NF1).

At the subsequent specialist evaluation at 19 months of age, the family recounted the presence of at least 1 birthmark and several areas of hypopigmentation, which were more apparent when she was angry. There was no history of vision or hearing concerns. She was described as a cranky baby until 10 months of age. She had no reflux or feeding difficulties; however, they did report that she spit out food and tended to pocket solid food. There were no concerns of pain, and there was no additional past medical history outside the current medical workup except for an episode of gastroenteritis.

Developmentally, she did not crawl until 1 year of age and walked at 18 months. Parents described frequent falls, especially with greater distances. At the age of 19 months, she babbled and repeated and had at least 20 words. She also identified several body parts. There were no other family members with café au lait macules, birthmarks, or nerve-based lesions. There was no other significant family history.

On examination, height and weight were in the 44th and 90th percentiles, respectively. General physical examination was notable for very pale skin, more than 6 café au lait macules measuring $>0.5$ cm, several areas of hypopigmentation, and no skin fold freckling. She also had an area of relative hyperpigmentation with increased vascular markings over the posterior aspect of the ribs with hair growth that matched the region of pigmentation (Fig. 2). Thoracic scoliosis was noted. There was no spinous tenderness to palpation. There was slight bowing of the tibia on outward examination but no joint tenderness or swelling. Labia were slightly darker in pigmentation with increased soft tissue bulk. Her mental status examination revealed slight speech disarticulation; she could identify body parts. Cranial nerves were intact. Deep tendon

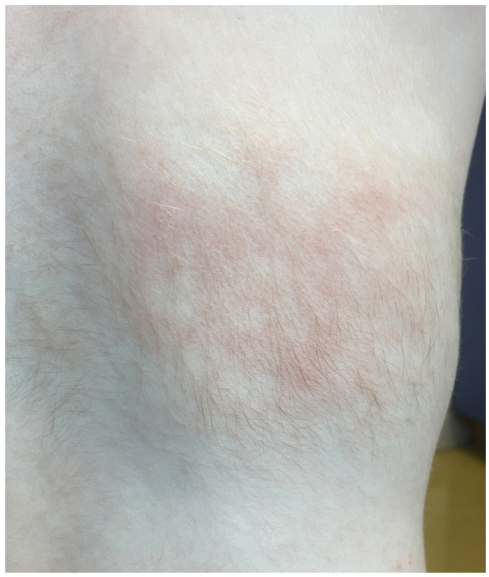

**Figure 2** Patient was noted to have an area of hyperpigmentation, subcutaneous nodularity, and increased hair growth over her upper back, which are skin changes typical of plexiform neurofibroma. (Color version of figure is available online.)

reflexes were 2+ and symmetric. She had globally decreased muscle tone throughout without any motor asymmetries noted. Sensation and coordination were intact. She walked with assistance.

Additional diagnostic workup was performed, which included bilateral tibial films. These revealed moderate apex anterolateral bowing of the tibial shafts, left greater than

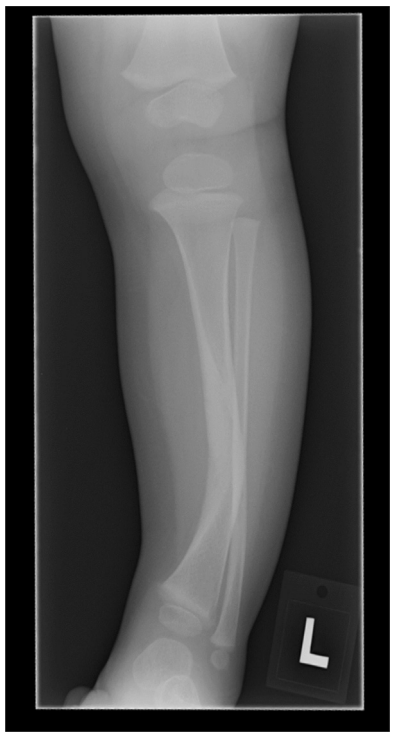


**Figure 3** Plain X-ray demonstrates anterolateral bowing of the tibia without evidence for fracture. Mineralization is normal.