Abdominal Pain: A Unique Presentation of Neurofibromatosis-1

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ABSTRACT

Neurofibromatosis type 1 (NF-1) is a common autosomal dominant neurocutaneous disorder affecting 1 in 3000 people. It often presents with a myriad of cutaneous features including, neurofibromas, Lisch nodules, café-au-lait macules, axillary freckling, and plexiform neuromas (PFN). Many other non-cutaneous manifestations have been observed in NF-1. Gastrointestinal (GI) stromal tumors, malignant peripheral nerve sheath tumors, and adrenocarcinoma are commonly found in the GI tract of NF-1 and can manifest as a complaint of abdominal pain. Here we present a unique case of NF-1 with an initial presenting symptom of abdominal pain caused by PFN located outside the gastrointestinal tract.

CASE PRESENTATION

A 13-year-old female was admitted for investigation of non-radiating abdominal pain in the right upper quadrant. The patient did not have a significant medical history or family history of NF-1; however, her mother was found to have multiple café au lait macules. Physical examination revealed numerous café-au-lait macules on her limbs (Fig. 1) and torso, axillary freckling and three subcutaneous nodules were noted on the neck and face. Further inspection revealed bilateral Lisch nodules (Fig. 2).

Cervical MRI revealed PFNs from the foramen magnum to T1 extending into the neuroforamina bilaterally. Imaging studies of the cervical, thoracic, and lumbar regions revealed innumerable tumors around the margins of multiple transverse processes, posterior ribs, and neural foramen. Plexiform neurofibromas were also found extending into the retroperitoneum, pelvis, and illoposa muscles. Imaging of the bowel was negative for gastrointestinal tumors (Fig. 3). (Fig.4)

No signs of cord compression or neurological symptoms were found although countless neurofibromas were located. Other structural symptoms such as disc atrophy, intramedullary signal abnormality, or hemorrhage were not present. The chief complaint of abdominal discomfort was likely caused by PFN’s compressing structures in the abdomen since gastrointestinal tumors were not found on MRI or esophagogastroduodenoscopy (EGD) with biopsy.

There was no acute neurological intervention warranted. It was recommended that she have an MRI of the cervical/thoracic/lumbar spine in 1 year, or sooner if she should have any acute changes. The patient was released from the hospital and is being followed by dermatology, ophthalmology, and neurosurgery services.

CLINICAL PHOTOS

Table 1- NIH diagnostic criteria for NF-1

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<th>Cutaneous Manifestations</th>
<th>Common Age of Onset for Patients with Neurofibromatosis Type 1</th>
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<tr>
<td>Café-au-lait spots</td>
<td>Birth through age 12</td>
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<td>Axillary freckling and Lisch nodules</td>
<td>Age 3-adolescence</td>
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<td>Subcutaneous and cutaneous neurofibromas</td>
<td>Adolescence</td>
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REFERENCES