

Neurofibromatosis

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Section: Abdominal imaging

Case Type: Clinical Cases

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DAY

Patient: 17 years, female

Clinical History:

A 17-year-old girl with chronic complaints of abdominal discomfort and swelling was referred for a CT scan of the abdomen. Laboratory findings were within normal limits.

Imaging Findings:

Fig. 1-2: Contrast-enhanced CT scan of the abdomen with axial (Fig. 1) and coronal (Fig. 2) reformatted images at the level of the celiac trunk shows an ill-defined low-attenuation soft tissue mass with involvement of the root of the mesentery. The coronal reformatted image (Fig. 2) better depicts the extent of the mass along the course of the vessels into the liver.

Fig. 3-5: MR examination of the abdomen with axial (Fig. 3) and coronal (Fig. 4) T2 HASTE fat-suppressed images at the level of the celiac trunk and kidneys show a hyperintense mass surrounding the mesenteric axis and extending into the liver. The coronal image shows numerous small hyperintense nodules with hypointense center (target sign) in the mesentery and subcutaneous fat. The coronal image (Fig. 5) at the level of the kidneys shows bilateral extensive elongated fusiform masses with high signal intensity, extending along the course of the lumbosacral plexus.

Fig. 6-7: On the fat-suppressed axial SE T1 MR-images before (Fig. 6) and after (Fig. 7) intravenous gadolinium contrast administration, the lesion is of low signal intensity and there is only very subtle peripheral contrast enhancement.

Discussion:

Neurofibromatosis type 1 (NF-1) or von Recklinghausen disease is the most common neurocutaneous disorder and affects approximately 1 in 3000 people. It has an autosomal dominant inheritance pattern. Common findings in NF-1 include café au lait spots, neurofibromas and plexiform neurofibromas, freckling in the axilla or groin, optic gliomas, Lisch nodules and skeletal dysplasia. Plexiform neurofibromas are considered pathognomonic of the disease. Gastro-intestinal involvement is relatively rare and affects 10-25% of patients. Abdominal neoplasms in NF-1 can be divided into five basic categories: neurogenic tumors (neurofibroma, plexiform neurofibroma, malignant peripheral nerve sheath tumor), neuroendocrine neoplasms (carcinoid, pheochromocytoma, paraganglioma, gangliocytic paraganglioma), non-neurogenic gastrointestinal mesenchymal tumors (GIST, leiomyoma, leiomyosarcoma), embryonal tumors (rhabdomyosarcoma, neuroblastoma, Wilms tumor) and miscellaneous tumors (gastrointestinal – pancreatic and biliary adenocarcinoma). Clinical presentation depends on lesion size and the site of involvement. Most small mesenteric plexiform neurofibromas are asymptomatic. Plexiform neurofibromas in gastrointestinal involvement arise possibly from either serosal (Auerbach's plexus) or

mesenteric nerves. They consist of a fingerlike tumoral network extending in a serpentine or ropelike fashion along a nerve and its branches. They may either extend from the root of the mesentery to the wall of the intestine or may originate primarily in the intestinal wall and appear as intraluminal or intramural masses.

Involvement of the liver and bile ducts is very uncommon. Plexiform neurofibromas may manifest as focal liver lesions or involve the periportal spaces and extend throughout the liver adjacent to portal vein branches.

On computed tomography (CT) neurofibromas and plexiform neurofibromas are hypoattenuating masses. The attenuation values range from 20 to 25 HU on non-enhanced scans and 30 to 50 HU after intravenous contrast administration. The low attenuation of neurofibromas has been attributed to a combination of factors including lipid-rich Schwann cells, adipocytes, cystic degeneration, myxoid and mucinous stroma.

On MRI neurofibromas show a low signal intensity on T1-weighted images and heterogeneous high signal intensity on T2-weighted images. Plexiform neurofibromas have a characteristic ringlike or septated pattern that is best depicted on T2-weighted images and gadolinium-enhanced T1-weighted images. MRI is more accurate than CT in defining precise lesion extension.

The differential diagnosis varies according to the anatomic location. In the abdomen NF-1 may mimic adenopathy such as seen in lymphoma, tuberculosis and metastatic disease. The main differential diagnosis is mesenteric lymphangioma. In NF-1 there are often associated small cutaneous and abdominal neurofibromas, a finding that is not seen in mesenteric lymphangioma.

The rate of reported malignant transformation is unclear. Young age at diagnosis, multiple tumors and large tumor load may be in favor of a higher risk of malignant transformation. Surgery is the treatment of choice but is rarely possible because of the infiltrative nature of the plexiform neurofibromas.

Differential Diagnosis List: neurofibromatosis type 1

Final Diagnosis: neurofibromatosis type 1

References:

Levy AD, Patel N, Dow N, Abbott RM, Miettinen M, Sobin LH (2005) From the Archives of the AFIP. Abdominal neoplasms in patients with Neurofibromatosis Type 1: Radiologic-Pathologic Correlation. *Radiographics* 25:455-480. (PMID: [15798063](#))

Zacharia TT, Jaramillo D, Poussaint TY, Korf B (2004) MR imaging of abdominopelvic involvement in neurofibromatosis type 1: a review of 43 patients. *Pediatr Radiol* 35:317-322. (PMID: [15517232](#))

Hartley N, Rajesh A, Verma R, Sinha R, Sandrasegaran K (2008) Abdominal manifestations of neurofibromatosis. *J Comput Assist Tomogr* 32:4-8. (PMID: [18303281](#))

Karcaaltincaba M, Haliloglu M, Akpınar E, Akata D, Özmen M, Ariyurek M, Akhan O (2007). Multidetector CT and MRI findings in periportal space pathologies. *Eur J Radiol* 61: 3-10. (PMID: [17123763](#))

Parizel PM, Geniets C (2006). Tumors of peripheral nerves. In: De Schepper AM (Ed.), Vanhoenacker F, Gielen J, Parizel PM (Co-Eds.): *Imaging of Soft Tissue Tumors*. Springer Berlin Heidelberg New York (2006): pp. 325-353.

Figure 1

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Description: Contrast-enhanced CT scan of the abdomen with axial (Fig. 1) reformatted images at the level of the celiac trunk and the superior mesenteric artery. **Origin:**

Figure 2

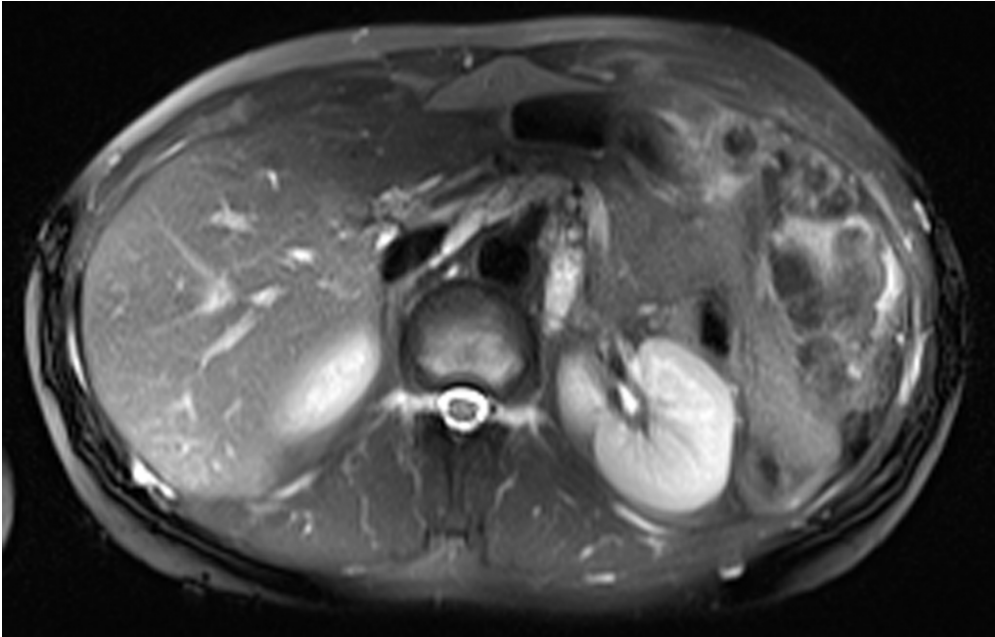
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Description: Contrast-enhanced CT scan of the abdomen with coronal (Fig. 2) reformatted images at the level of the celiac trunk and the superior mesenteric artery. **Origin:**

Figure 3

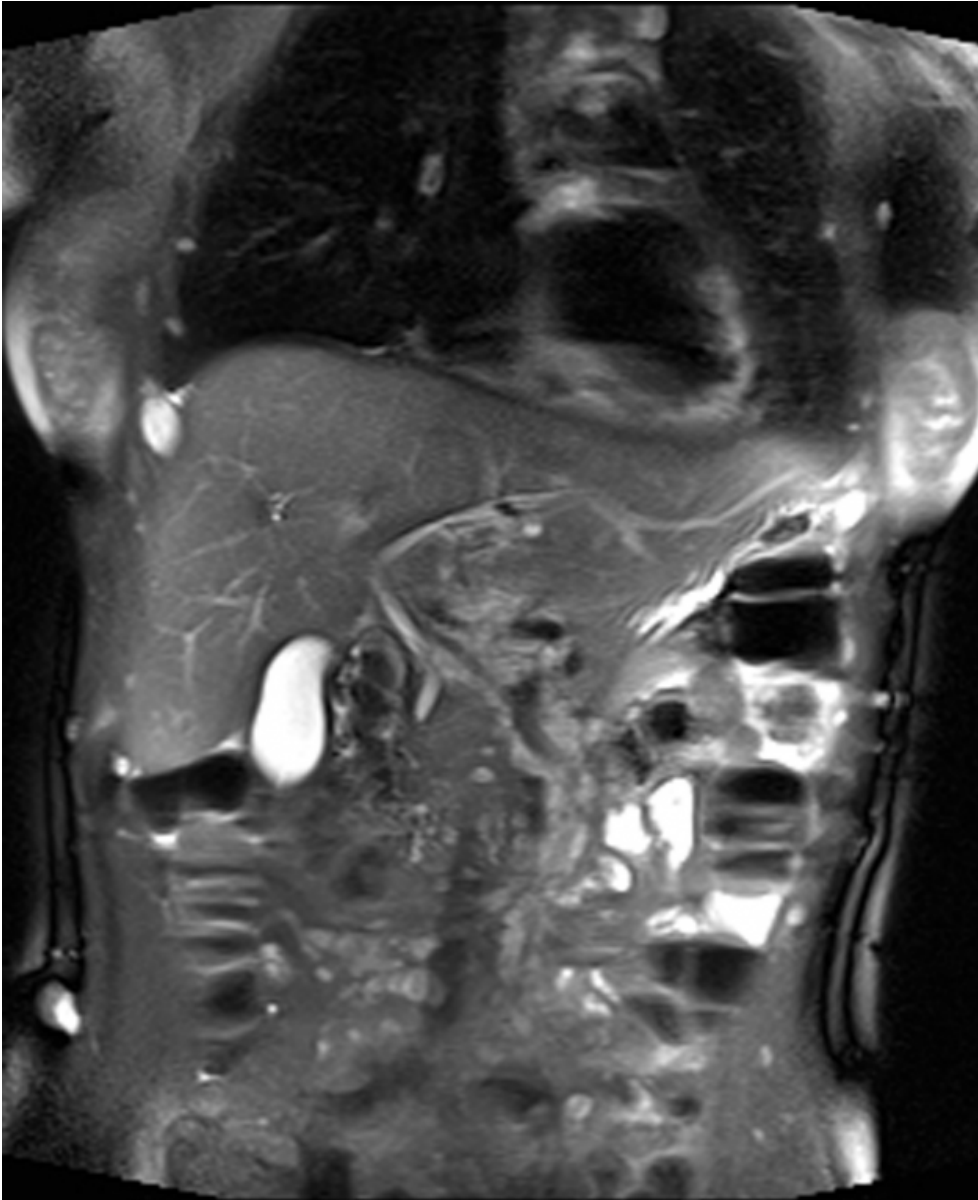
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Description: MR examination of the abdomen with axial (Fig. 3) and coronal (Fig. 4-5) T2 HASTE fat-suppressed images at the level of the celiac trunk and kidneys. **Origin:**

Figure 4

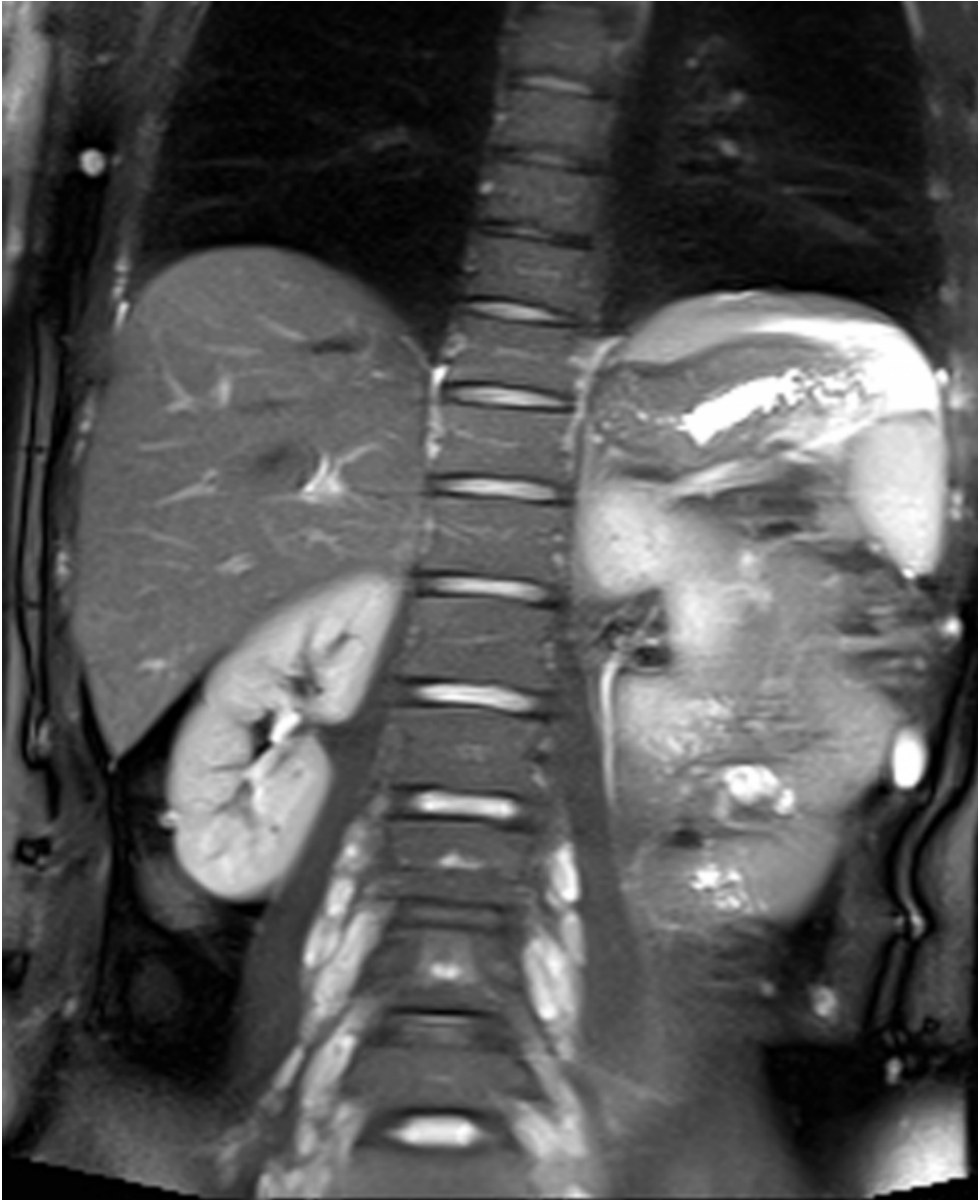
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Description: MR examination of the abdomen with axial (Fig. 3) and coronal (Fig. 4-5) T2 HASTE fat-suppressed images at the level of the celiac trunk and kidneys. **Origin:**

Figure 5

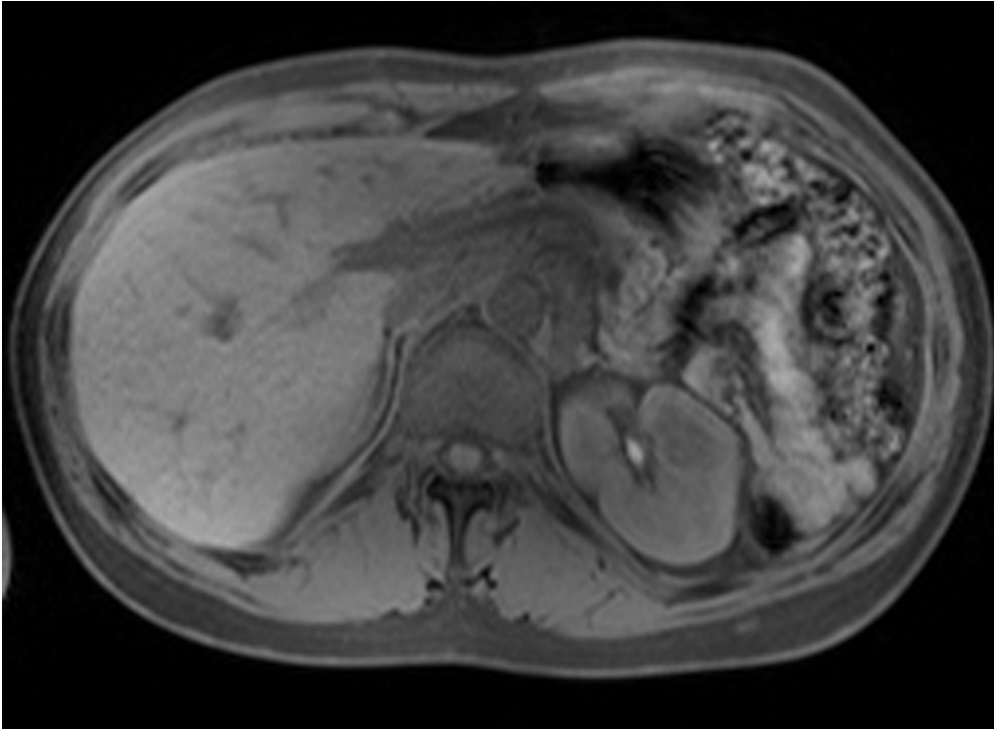
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Description: MR examination of the abdomen with axial (Fig. 3) and coronal (Fig. 4-5) T2 HASTE fat-suppressed images at the level of the celiac trunk and kidneys. **Origin:**

Figure 6

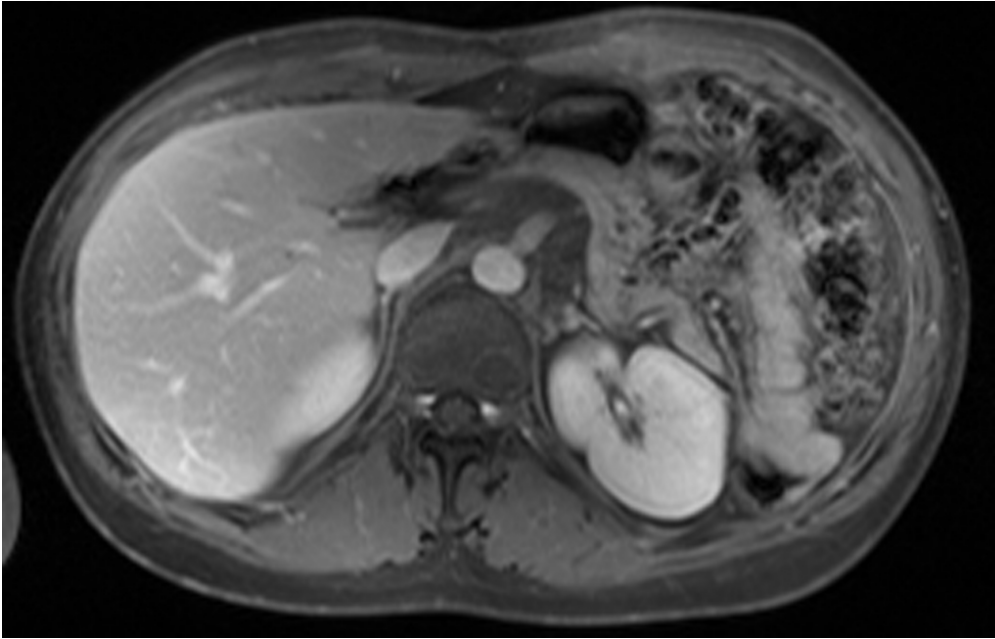
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Description: Fat-suppressed axial SE T1 MR-images before (Fig. 6) and after (Fig. 7) intravenous gadolinium contrast administration **Origin:**

Figure 7

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Description: Fat-suppressed axial SE T1 MR-images before (Fig. 6) and after (Fig. 7) intravenous gadolinium contrast administration **Origin:**