

## Letters to the Editor

### 13 cm GIST in 19-year-old patient, is it the first manifestation of Carney triad?

*Key words: Gigant GIST. Carney. Condroma. Paraganglioma.*

Dear Editor,

Gastrointestinal stromal tumors have a median age of presentation at diagnosis of 60 years (1) but, what we must think about if our patient is 19-years old?

We present the case of a 19-year-old woman, without any relevant personal clinical background. She was requested an abdominal ultrasound because an abdominal mass was detected during physical examination. Radiologist decided to perform a

CT, demonstrated multiple gastric wall dependent masses that were extended until a diameter of 13 cm (Fig. 1A). We carried out an upper endoscopy by which we found an ulcerated submucosal lesion, 5 cm in diameter, in the fundus. From the middle gastric body and covering all its circumference there were multiple submucosal lesions alternating with extrinsic compression, which reduced the diameter of the stomach, some of them with a central ulceration (Fig. 1B). Biopsies were taken that were diagnostic for GIST with high positivity to C-KIT.

We did extension studies that demonstrated the absence of metastases and other lesions. After the diagnosis, the case was presented to the tumors committee. They decided the administration of imatinib previous to surgery, with the intention of reducing the tumor mass and performing a less aggressive surgery. During the next six months, there was no modification in the size of the mass and, after an episode of upper gastrointestinal bleeding that was successfully controlled, surgery was decided; the patient underwent total gastrectomy (Fig. 1C).

We did a molecular study after surgery that was negative for the C-KIT exons mutations 9,11,13 and 17 and also was negative for the PDGFRA exons 12 and 18, that are the most common mutations found in the sporadic GIST.

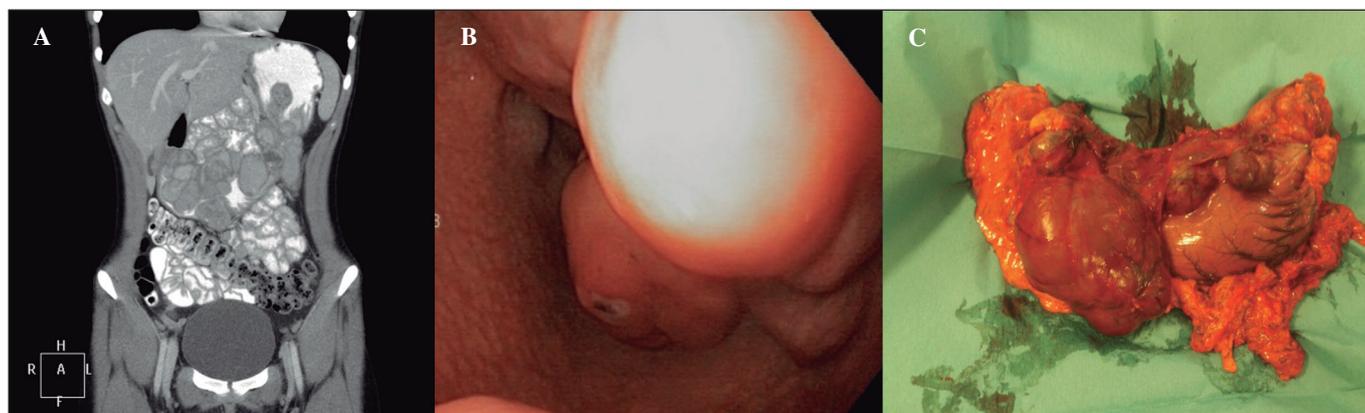


Fig. 1.

## Discussion

Gastrointestinal stromal tumors are characterized with a median age of 60 at diagnosis, male predominance and a median size between 5 and 8 cm (1). Our patient did not present any of these characteristics, so we did a genetic study that was negative for the dominant autosomic familiar form. We also discarded neurofibromatosis type I (2). Then, what other causes could explain a giant GIST with such an early age of presentation? Carney triad.

The Carney triad is the combination of gastric stromal sarcoma, pulmonary chondroma, and extra-adrenal paraganglioma (3). It is an extremely rare syndrome, with about 120 documented cases, some of them incomplete forms (4). This disorder affects mostly young women (5). The first tumor appears around the age of 20 in 45 % of the patients (6). At the moment of diagnosis, only one tumor is present and the first tumor identified usually is a gastric GIST. There is often a lapse of time above 5 years between the diagnosis of the first and the second tumor, and sometimes it could be higher than 20 years (4). Because of this, all the pediatric GIST or GIST under 20 year of age should be considered as a potential Carney, especially if the patient is female and with multiple gastric tumors (4).

GIST in Carney triad is multiple and with a large size, it does not have the common mutations of C-KIT and PDGFRA that appears in sporadic GIST. Also it does not respond well to the treatment with imatinib especially in females (5).

Our patient met all the characteristics described previously for the Carney's GIST. We discarded the presence of condromas and paragangliomas; however, as GIST is the first tumor that usually appears, we must think about the possibility of developing it in the future, as a potential Carney, and because of this, we

must perform an close monitoring and follow-up of the patient during years.

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