gangliomas extraadrenales malignos tienden a presentar márgenes irregulares con densidad heterogénea[9]. La gammagrafía con metayodobenzilguanidina, un análogo de las catecolaminas captado en más del 90% de los paragangliomas, puede ser utilizada para su localización y tratamiento. Esta prueba es específica, pero en ocasiones carece de sensibilidad[10].

El tratamiento de elección es la resección quirúrgica, ya que existe una correlación positiva entre la exéresis completa y el pronóstico. La quimioterapia se utiliza en pacientes con enfermedad no resecable o metastásica y como tratamiento neoadyuvante en grandes tumores para facilitar la resección quirúrgica[7]. En las recurrencias tumorales también está indicada la resección quirúrgica. No existen reportes de metástasis a ganglios linfáticos ni a órganos cercanos o distantes[6].

Los paragangliomas extraadrenales son un dilema diagnóstico para el patólogo. La evaluación citológica de los hallazgos neuroendocrinos de las células tumorales ayuda a evitar los diagnósticos erróneos de carcinoma o sarcoma. Su patrón microscópico es trabecular, con cordones de células tumorales definiendo nidos de células cuboidales con patrón Zellballen, separados por septos fibrovascularizados. Los indicadores fiables de malignidad son invasión extensa de órganos adyacentes (ganglios linfáticos) y diseminación metastásica a distancia (pulmón e hígado)[6].

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Recurrent cecocolic intussusception in a young woman

Intususcepción cecocólica recurrente en una mujer adulta joven

A 21-year-old female, white race, native resident in western Europe and with no relevant personal history, was admitted with a severe, progressively worsening, abdominal pain lasting for 7 days. She denied vomits, weight loss or alterations in bowel movements. No other extraintestinal symptoms were present. Laboratory tests only revealed mild leukocytosis. Abdominal ultrasonography suggested an abdominal mass in the right flank. Abdominal CT showed a colonic intussusception with cecum, ascending colon, fat and mesenteric vessels (intussusceptum) invaginated into the transverse (intussuscipiens) (Fig. 1); within this area a nodular mass with 4 cm was identifiable. After a multidisciplinary discussion a colonoscopy was proposed. It revealed a significant narrowing in the transverse and ascending colon and a congestive and extensively ulcerated pseudopolypoid mass with 4 cm in the cecum (Fig. 2). Forceps biopsies were performed. After clinical improvement and before a histological evaluation was available, the patient was referred to our outpatient department. Nevertheless, 8 days after the discharge, she recurred to the emergency department with a similar abdominal pain. An intussusception relapse was again suggested by abdominal ultrasonography. A surgical approach was then decided and a right hemicolectomy performed. Histological evaluation revealed a cecal lymphoproliferative disorder constituted by medium-size cells with high proliferative and apoptotic activities (Fig. 3). Immunostaining revealed CD3 and CD20 positivity (Fig. 4) and CD23, CD30, B cell leukemia/lymphoma 2 (bcl-2), ciclin D1 and Pax 5 negativity. These features were compatible with a Burkitt lymphoma. The patient was then referred to the hemato-oncology department.

Burkitt lymphoma is an aggressive non-Hodgkin B-cell lymphoma related to the translocation and deregulation of the c-myc gene.[1]

Burkitt lymphoma is rare in Western Europe and North America and it is usually diagnosed in children and young adults. Currently there are three types of Burkitt lymphoma: sporadic, endemic and immunodeficiency-related.

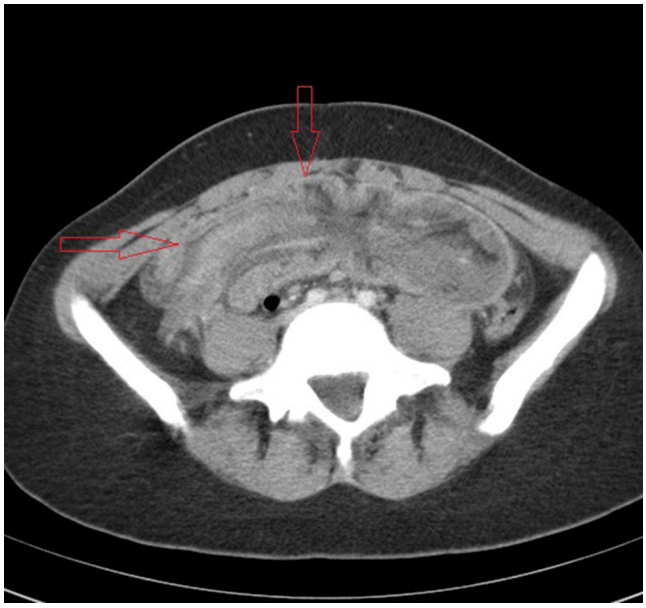

Figure 1 CT scan revealing a cecocolic intussusception (*arrows*).

Their epidemiology, clinical presentation and genetic features are different. Endemic variant, which is strongly related to Epstein–Barr virus infection, is mainly present in Africa and affects children. Sporadic variant is more common in Europe and United States and it is also more common in children. It rarely affects adult patients. Immunodeficiency-related Burkitt lymphoma occurs mainly in HIV-positive patients. A male predominance occurs in all variants.[2]

Clinical presentation differs between variants. Endemic variant usually involves facial bones, namely the maxilla, jaw and orbit with a rapid spread to extranodal sites including kidneys, breasts, bone marrow and meninges. Sporadic variant often has an abdominal presentation with bowel obstruction or gastrointestinal bleeding. Distal ileum, cecum, stomach and mesentery are frequently involved. Immunodeficiency-related Burkitt lymphoma often presents with lymph node involvement.[3]

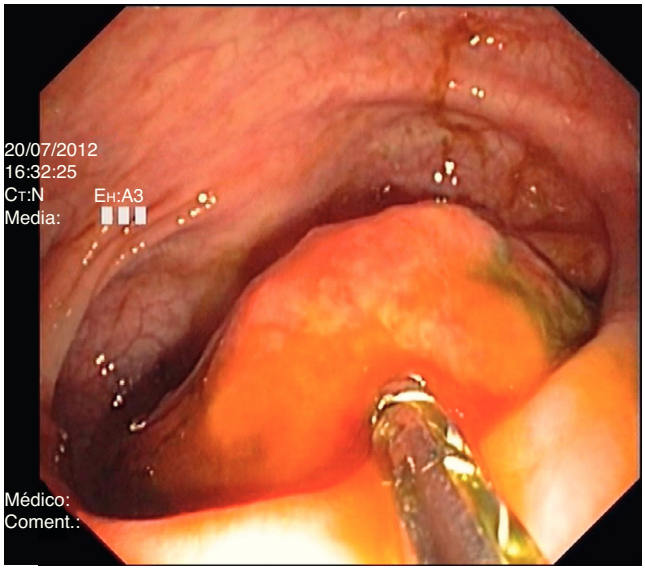

Figure 2 Colonoscopy revealing an ulcerated pseudopolypoid mass in the cecum.

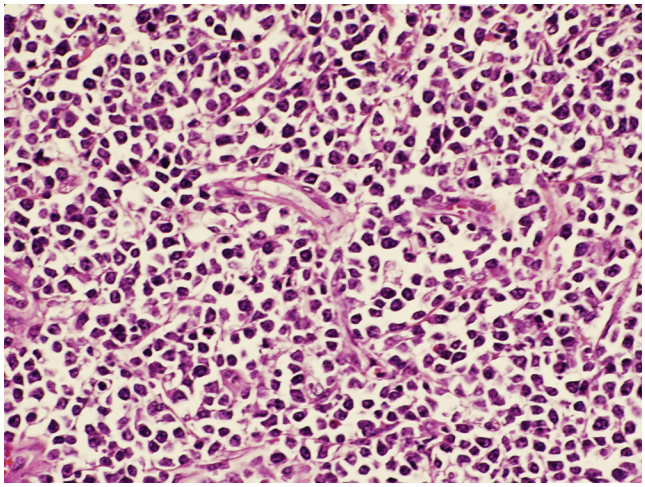

Figure 3 Histological analysis revealing medium-size cells with high proliferative and apoptotic activities (*hematoxylin–eosin staining*).

Although Burkitt lymphoma variants comprise some differences, histological features are similar. Tumor cells are usually monotonous, medium-sized, nuclei with multiple nucleoli and a basophilic cytoplasm. High rates of proliferation and apoptotic activities are typical. A ''*starry-sky*'' pattern is characteristic and refers to numerous macrophages ingesting apoptotic tumor cells. Immunostaining also contributes for the diagnosis. Burkitt lymphoma cells lack CD5, bcl-2 and CD23. In contrast they present positivity for CD19, CD20, CD22, CD10 and CD43.[4]

A definitive diagnosis of intestinal Burkitt lymphoma is usually achieved after histological evaluation of the surgical specimen. Nevertheless, endoscopy may achieve the diagnosis in some cases. Chemotherapy is the mainstay treatment for all variants of Burkitt lymphoma.[5] Surgical approach may be recommended in selected cases, namely for symptoms relief or to avoid complications during chemotherapy.[6]

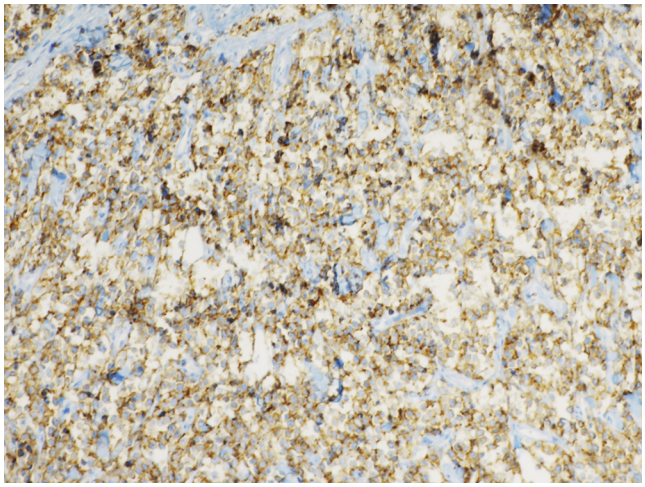

Figure 4 Medium-size cells with positive immunostaining for CD 20.

Intussusception consists in a prolapse of a proximal bowel segment into a distal segment. It is much more common in children than in adults (20:1). Among children intussusception is idiopathic in 90% of the cases. In contrast, a causal lesion is identified in 90% of the cases occurring in adults.[7] Tumors, colonic diverticulum or Meckel's diverticulum may cause intussusception. Bowel obstruction with abdominal pain, vomiting and often with a palpable mass is the most common clinical presentation. Currently, diagnosis is mainly based on ultrasonography and/or computed tomography.[8] Although a temporary relief of intussusception may be achieved by endoscopy, a surgical approach is recommended for a definitive resolution. Moreover, considering the existence of a *leading point*, the surgical approach is advisable.[9]

Authors describe a case of a white adult female with recurrent cecocolic intussusception caused by an intestinal Burkitt lymphoma. Colonoscopy achieved a temporary relief of the intussusception. Nevertheless, an early intussusception relapse occurred and a definitive surgical approach was performed.

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Is H63D a 'minor' HFE polymorphism?

¿Es la H63D un polimorfismo ''menor''?

Iron overload is associated with a variety of genetic and acquired conditions. Of these, HFE-hemochromatosis is by far the most frequent, well-defined inherited cause, and the majority of patients are homozygotes for the C282Y polymorphism. H63D is considered a 'minor' HFE polymorphism[1] but some clinical studies suggest an association between H63D homozygosity and iron overload. However, its role in the development of iron overload remains unclear.

We aimed to study the prevalence and phenotypic expression of H63D homozygotes in a cohort of patients referred to our hepatology clinic.

Method

Patients with increased serum ferritin (1.5× upper limit of normal, >300 ng/mL in females, >450 ng/mL in males – values adopted from the *Hemochromatosis and Iron Overload Screening Study*[2]) and increased transferrin saturation (>45% in females, >50% in males) were enrolled in our study, for three consecutive years. Genetic testing for C282Y/H63D/S65C mutations of the HFE gene was performed by polymerase chain reaction. Standard biochemical markers of iron status, including serum ferritin and transferrin saturation, were obtained. Other laboratorial data included complete blood count, fasting glucose, liver function tests, lipid profile and serology for hepatitis B and C. Also, baseline demographic and clinical characteristics (with quantification of alcohol consumption) were recorded. Chronic renal disease, shunts, chronic haemolytic anemia, thalassemia major, sideroblastic or spur cell anemia, parenteral iron overload and *porphyria cutanea tarda* were excluded.

Results

230 consecutive patients fulfilled the inclusion criteria, and were enrolled in our study. After HFE genetic analysis, the H63D homozygous mutation was identified in 6.96% ($n = 16$) of the individuals. The mean age was 53.8 years (28–76), and ten (62.5%) were male. In this group, we found median value of serum ferritin of 550.3 ng/mL and transferrin saturation of 57.6%.

Other causes of liver disease were found in 15: Non-Alcoholic Fatty Liver (NAFLD) in 8 patients, chronic alcohol consumption (>60 g/day) in 3, chronic Hepatitis B in 3 and C in 1 patient. One patient with H63D homozygosity was negative for other hepatic diseases, viral infections, alcohol abuse and NAFLD.

Discussion

It has been suggested that the H63D mutation contributes to iron overload, increasing serum iron and