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.

References

1. Ferry JA. Burkitt's lymphoma: clinicopathologic features and differential diagnosis. Oncologist. 2006;11:375–83.
2. Armitage JO, Weisenburger DD. New approach to classifying non-Hodgkin's lymphomas: clinical features of the major histologic subtypes. Non-Hodgkin's Lymphoma Classification Project. J Clin Oncol. 1998;16:2780–95.
3. Blum KA, Lozanski G, Byrd JC. Adult Burkitt leukemia and lymphoma. Blood. 2004;104:3009–20.
4. Yano T, van Krieken JH, Magrath IT, Longo DL, Jaffe ES, Raffeld M. Histogenetic correlations between subcategories of small noncleaved cell lymphomas. Blood. 1992;79:1282–90.
5. Levine AM. Challenges in the management of Burkitt's lymphoma. Clin Lymphoma. 2002;3 Suppl. 1:S19–25.
6. Fallon SC, Redell MS, El-Bietar J, Lopez ME, Vasudevan SA, Brandt ML. Intestinal perforation after treatment of Burkitt's lymphoma: case report and review of the literature. J Pediatr Surg. 2013;48:436–40.
7. Weilbaecher D 2nd, Bolin JA, Hearn D, Ogden W 2nd. Intussusception in adults. Review of 160 cases. Am J Surg. 1971;121:531–5.
8. Takeuchi K, Tsuzuki Y, Ando T, Sekihara M, Hara T, Kori T, et al. The diagnosis and treatment of adult intussusception. J Clin Gastroenterol. 2003;36:18–21.
9. Haas EM, Etter EL, Ellis S, Taylor TV. Adult intussusception. Am J Surg. 2003;186:75–6.

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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

transferrin, and that its relationship with hemochromatosis is independent.[3,4]

In our study, H63D+/+ mutation in the HFE gene was detected in 6.96% of our cohort of patients with iron overload. Underlying causes were found in 93.7% such as NAFLD, chronic alcohol consumption and chronic hepatitis B and C. These data confirm the association between hyperferritinemia and these conditions, especially fatty liver. However it does not clarify whether siderosis was related to the underlying disease, namely steatosis, rather than homozygosity for the H63D mutation. Therefore, co-morbid factors may complicate the interpretation of data in population studies of the expression of H63D homozygosity.

In one patient, however, the investigation for other causes was negative, and H63D homozygosity may have played a role. The causative role of H63D+/+ mutation in HH or iron overload has been demonstrated,[3] but with less penetrance and a considerable variation of phenotypic expression.[5] Environmental factors, variable gene penetrance or other gene mutations may explain the variable phenotypic expression of H63D homozygosity.

Conflicts of interests

None declared.

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References

1. European Association for the Study of the Liver. EASL clinical practice guidelines for HFE hemochromatosis. J Hepatol. 2010;53:3–22.
2. Adams PC, Reboussin DM, Barton JC, McLaren CE, Eckfeldt JH, McLaren GD, et al. Hemochromatosis and iron-overload screening in a racially diverse population. N Engl J Med. 2005;352:1769–78.
3. De Diego C, Opazo S, Murga MJ, Martinez-Castro P. H63D homozygotes with hyperferritinaemia: is this genotype the primary cause of iron overload. Eur J Haematol. 2006;78:66–71.
4. Aguilar-Martinez P, Bismuth M, Picot MC, Thelcide C, Pageaux GP, Blanc F, et al. Variable phenotypic presentation of iron overload in H63D homozygotes: are genetic modifiers the cause. Gut. 2001;48:836–42.
5. Kelley M, Joshi N, Xie Y, Borgaonkar M. Iron overload is rare in patients homozygous for the H63D mutation. Can J Gastroenterol Hepatol. 2014;28:198–202.

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