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Scientific Letters

Bilateral Breast Cancer and Peutz-Jeghers Syndrome

Cáncer de mama bilateral y síndrome de Peutz-Jeghers

Breast cancer (BC) is the most common malignant tumor in women as a result of a combination of physical, chemical, biological and genetic factors. Most BC is sporadic, and less than 10% of cases present a family relationship, fundamentally due to alterations in the BRCA1 and BRCA2 genes. Only a small number of hereditary cases can be attributed to mutations in other genes, such as p53 in Li-Fraumeni syndrome, PTEN in Cowden syndrome, MSH2 and MLH1 in Muir-Torre syndrome, ATM in ataxia telangiectasia and STK11 in Peutz-Jeghers syndrome (PJS).[1]

PJS is a rare disease characterized by the association of hamartomatous polyps in the digestive tract, hyperpigmented macules on the lips and oral mucosa and an increased risk for developing digestive and extradigestive neoplasms. Its hereditary pattern is autosomal dominant (AD) with a variable penetrance, and in a high percentage of cases (80%–94%) it is associated with mutations in the specific 19p13.3 locus of the LKB1/STK11 gene.[2,3]

PJS was first defined in 1954 by Bruwer et al.[4] and it is classified in the hamartomatous polyposis group together with juvenile polyposis, hereditary mixed polyposis syndrome, Cowden syndrome and the Ruvalcava-Myrhe-Smith syndrome, and it shares a high risk of colorectal cancer.[5] In PJS, there is also a high probability of neoplasia in the rest of the digestive system, and other locations at early age, most frequently breast (54%), ovarian (21%), cervical (19%) and lung (15%) cancer.[5,6]

This rare syndrome (1/200 000 newborns), which equally affects both sexes, is usually diagnosed during childhood based on family history, presence of typical melanotic macules or problems caused by the growth of intestinal polyps that cause symptoms of invagination, obstruction or hemorrhage.

Although pigmentation can appear at any age, it is most frequent during infancy and early childhood, with melanin deposits on the lips (95%) or in the mouth (83%), and less frequently on hands, feet and periorificial locations. The majority of the clinical manifestations are gastrointestinal, although the hamartoma–adenoma–carcinoma sequence is controversial for some authors[7] and it is known as the "landscape effect". The alterations, especially in the lamina propia, can lead to epithelial cancers.[8] On occasion, the initial manifestation is the appearance of a neoplasm at an early age due to the risk accumulation.

We present the case of a patient affected by BC in whom the hereditary cause was discovered incidentally. The patient is a 51-year-old Caucasian woman with a history of chronic anemia (treated with oral iron supplements) who had also undergone cholecystectomy. She reported no family history of breast cancer or any genetic alterations. At the age of 50, the patient had been diagnosed with bilateral breast carcinoma after a mammography had detected suspicious microcalcifications, which was later confirmed by magnetic resonance imaging (MRI) (Fig. 1). Bilateral mastectomy was performed along with selective biopsy of sentinel axillary lymph nodes, which was negative; immediate reconstruction was therefore performed with a Becker expander/implant. The pathology study reported the carcinoma to be a high-grade intraductal type (Van Nuys index >8) in both breasts.

The patient came to the emergency room complaining of rapid onset discomfort in the right hypochondrium and nausea, with no associated fever. Blood tests showed mild hypertransaminasemia, and the study was completed with ultrasound and cholangio-MRI, providing a diagnosis of choledocholithiasis. During ERCP, numerous duodenal polyps were found as well as a choledochal calculus, which was extracted. Several gastric and duodenal polyps were removed by upper gastrointestinal (UGI) endoscopy (Fig. 1). The presence of perioral melanotic macules and the histologic confirmation of hamartomatous polyps established the diagnosis of PJS.[9]

All patients diagnosed with PJS should be followed-up for life and sent for genetic counseling in accordance with the recommendations proposed by a committee of European experts that met in Mallorca in 2007, published in a consensus document (clinical guidelines) with a level of evidence of B or C.[9] The objective is twofold: (1) to detect the presence of polyps with an appropriate size for endoscopic extirpation in order to

☆ Please cite this article as: Lorenzo Liñán MÁ, Lorenzo Campos M, Motos Micó J, Martínez Pérez C, Gumbau Puchol V. Cáncer de mama bilateral y síndrome de Peutz-Jeghers. Cir Esp. 2013. http://dx.doi.org/10.1016/j.ciresp.2012.05.025

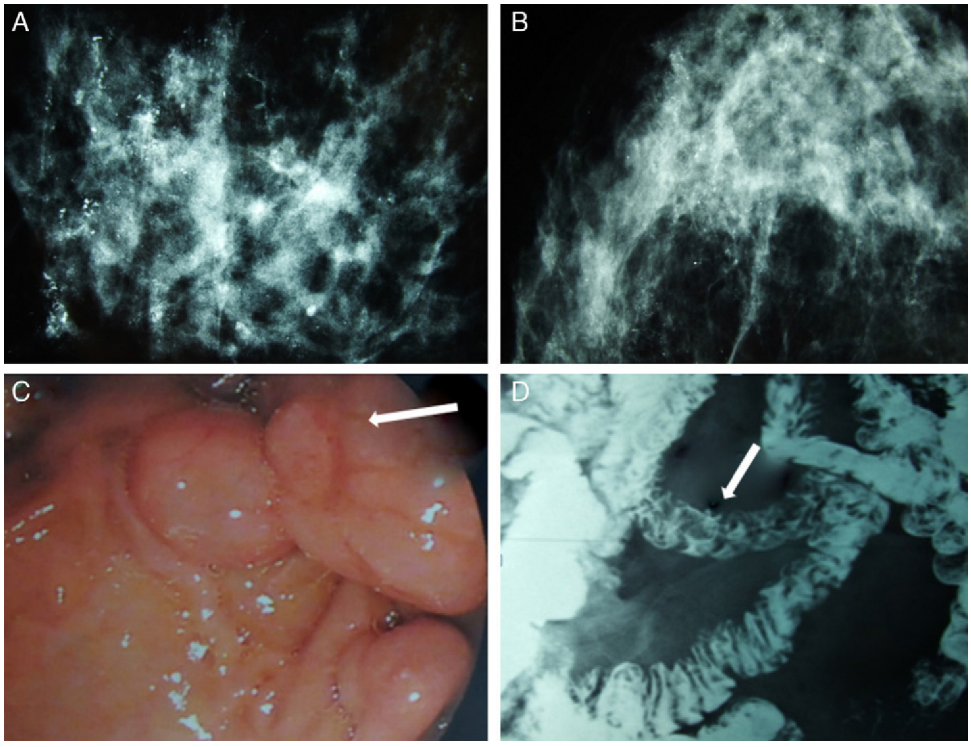

Fig. 1 – Mammography, barium transit study and digestive ultrasound images: (A) diffuse microcalcifications in the right breast; (B) diffuse microcalcifications in the left breast; (C) upper GI ultrasound reveals hamartomatous polyps in the third duodenal portion (white arrow); (D) gastrointestinal tract where filling defects are observed in the proximal jejunum that represent polypoid formations (white arrow).

Table 1 – Recommendations for Testing and Follow-up in Patients Diagnosed With Peutz-Jeghers Syndrome, With Their Grades of Recommendation.

General	
Annual hemogram and liver function enzymes	
Annual clinical examination	
No routine drug prophylaxis is recommended	Grade B
Urogenital system	
Annual testicular examination from birth until the age of 12	Grade C
Testicular ultrasound if alterations are detected during examination	Grade C
Cervical cytology every 2–3 years starting at age 25	Grade C
Gastrointestinal system	
Upper endoscopy and colonoscopy at the age of 8	Grade C
If polyps are detected, repeat every 3 years until the age of 50	
If polyps are not detected, repeat at the age of 18 and every 3 years until the age of 50	
Colonoscopy every 1–2 years after the age of 50	Grade C
Endoscopic video capsule every 3 years starting at the age of 8	Grade B
Follow-up with pancreatic studies is not recommended	Grade B
Intraoperative enteroscopy in PJS undergoing laparotomy	Grade B
Mammary glands	
Monthly self-examination starting at the age of 18	
Annual magnetic resonance (or ultrasound) from ages 25–50	Grade C
Annual mammography starting at age 50	Grade C

Taken from AD Beggs et al.[9]
PJS, Peutz-Jeghers syndrome.

avoid possible complications; and (2) to provide early cancer diagnosis.

These recommendations, shown in Table 1, require a close multidisciplinary follow-up with analyses as well as examinations of the digestive and urogenital systems, in addition to programmed breast examinations at a younger than usual age. Our patient currently continues to undergo these follow-up tests and her genetic study is pending, which could define the recommended interval for screening and checkups within her family, in order to avoid future neoplasia.

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2173-5077/$ – see front matter

http://dx.doi.org/10.1016/j.cireng.2013.07.017

Retroperitoneal Haemorrhage After Traumatic Rupture of a Phaechromocytoma

Hemorragia retroperitoneal tras rotura traumática de un feocromocitoma

Pheochromocytomas are neuroendocrine tumors of the adrenal medulla (originating in the chromaffin cells) or extra-adrenal chromaffin tissue that occur during involution after birth and secrete excessive quantities of catecholamines. Traumatic rupture of a pheochromocytoma and the resulting retroperitoneal hemorrhage are rare complications,and management can be difficult.

We present the case of a 45-year-old male who, in a suicide attempt, fell from the fifth floor of a building and suffered severe trauma. His medical history included arterial hypertension, insulin-dependent diabetes and the presence of a right adrenal mass that was being studied after having been incidentally detected during an abdominal ultrasound performed some weeks earlier. Meanwhile, the patient was receiving psychiatric drug treatment for anxiety and depression.

Upon arriving at our Emergency Department, the patient presented blood pressure of 180/110 mmHg, heart rate 135 bpm and hemoglobin 9.9 g/dL. Emergency abdominal computed tomography (CT) revealed a right adrenal mass measuring 10.5 cm×9 cm×8.8 cm with signs of active bleeding and a retroperitoneal hematoma (Fig. 1). The patient also presented facial trauma, bilateral pneumothorax and fractures of the right transverse processes from L1 to L5, sacrum, coccyx and right tibia and open fracture of the right astragalus.

Given the suspected active bleeding of the adrenal mass, the patient was considered a candidate for radiological embolization prior to the possible need for emergency surgery. Due to the hemodynamic stability and to the fact that hemoglobin levels were maintained above 9 g/dL for 24 h after hospitalization, we opted for a conservative approach and initiated treatment with urapidil and labetalol, which provided adequate blood pressure control 36 h after hospitalization.

Catecholamine determination in urine showed levels of metanephrine of 20 659 mcg/dL (normal, 64–302 mcg/dL) and norepinephrine 5644 mcg/dL (normal, 12–85 mcg/dL). These

☆ Please cite this article as: Gómez Hernández MT, et al. Hemorragia retroperitoneal tras rotura traumática de un feocromocitoma. Cir Esp. 2013;91:197–9.