Duodenal Hematoma and Pancreatitis Complicating Endoscopic Intestinal Biopsy in a Boy with Noonan Syndrome

E. Leva*, F. Macchini1, A. Di Cesare1, R. Arnoldi1, V. Gentilino1, G. Brighelli1, G. Farris1, A. Morandi1, M. Torricelli1, Massimo Agosti2 and Eugenio Cocozza2

1Department of Pediatric Surgery, Fondazione IRCCS “Ca’ Granda” – Ospedale Maggiore Policlinico, Milano, Italy
2Azienda Ospedaliera di Circolo, Varerse, Italy

Keywords: Duodenal hematoma; Pancreatitis; Noonan syndrome; Children; Growth retardation

Introduction

Duodenal intramural hematoma is a rare condition, mostly described in children and young adults [1,2]. It can be a complication of duodenal biopsy, especially in patients with predisposing hemorrhagic diathesis. It can determine secondary pancreatitis because of ampullary hematoma. Noonan Syndrome (NS) is an autosomal dominant disorder characterized by short stature, typical facial dysmorphisms, congenital heart defects and other anomalies [3,4] and can determine secondary pancreatitis because of ampullary hematoma [1,2,5-7].

Noonan Syndrome (NS) is an autosomal dominant disorder characterized by short stature, typical facial dysmorphisms, congenital heart defects and other anomalies [8-10] such as bleeding problems which have been reported in up to 55% of patients [8-14]. NS is a clinical diagnosis.

Case Report

A 14 years old boy was transferred to our department for acute pancreatitis from a first level hospital. Three days before, multiple endoscopic duodenal biopsies were performed at the Department of General Gastroenterology of the hospital of origin, to rule out celiac disease because of unexplainable growth retardation and short stature. Previously, antigliadin and antiendomysium antibodies were recorded low and IGF-1, treated with GH for the last 8 years and, finally, a paediatric neuropsychiatric consultation for attention deficit. On examination he presented apirexial and stable vital signs but tender abdomen, painful over the epigastrum and right hypocondrium and absent bowel sounds. An ultrasound (US) scan showed an enlarged oedematous pancreas with heterogeneous echogenicity, a circumscribed heterogeneous duodenal parietal collection (10 x 5 x 8 cm) involving the second and third portions and some corpusculated free fluid in peritoneal cavity. CT (computed tomography) scan confirmed and better defined those findings consisting with duodenal parietal hematoma conditioning an acute pancreatitis.

Vital signs were strictly monitored. Laboratory test showed increased serum amylase (1403 U/L) and lipase (1656 U/L) but normal full blood count, electrolytes, liver and kidney functions. Full coagulation study was done, resulting in slightly increased PT and reduced levels of factors VII, X and factor V, while platelet aggregation and secretion resulted normal.

For this reason the patient received vitamin K and factor VII supplementation.

Conservative treatment was undertaken namely nasogastric drainage, absolute starving, total parenteral nutrition, somatostatin, analgesics, ranitidine and antibiotics. Follow-up with serial US scans showed a progressive regression of both the hematoma and the peritoneal fluid collection within a couple of weeks, confirmed by CT scan as well. Laboratory test showed regression of amylase and lipase values. Vital signs were stable throughout the recovery.

The patient was started on oral feeds at day 14 since initial diagnosis, stopped parenteral nutrition at day 20 and was discharged the day after on low fat diet. During the recovery, a more accurate clinical evaluation of the patient have been collected: neonatal and infancy feeding difficulty with delayed weaning at 12 months, mild motor delay (sitting at 10 months, walking at 20 months, talking at 18 months), bilateral orchidopexy at 1 year of age, growth retardation with normal GH and low IGF-1, treated with GH for the last 8 years and, finally, a paediatric neuropsychiatric consultation for attention deficit. 

*Corresponding author: Ernesto Leva, Department of Pediatric Surgery, Fondazione IRCCS “Ca’ Granda” – Ospedale Maggiore Policlinico, Milano, Italy. E-mail: ernesto.leva@unimi.it

Received March 08, 2012; Accepted March 22, 2012; Published March 25, 2012


Copyright: © 2012 Leva E, et al. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.
neuropsychiatric consultation for attention deficit and behavioural problems (hyperactivity, fidgety, irritability).

Furthermore, a focused objective examination by the genetics team revealed characteristic features of NS, namely height of 140.5 cm (below 3rd centile), wide internipple space (teletelia), and chest slightly excavatum, hypertelorism, triangular shaped face, down-slanting palpebral fissures.

Despite the genetic tests for NS resulted negative, the phenotypic features of the child confirmed the diagnosis.

Cardiac and ophthalmological evaluation, made to rule out some important anomalies associated to the syndrome, resulted normal (Figure 1).

A 1 month follow-up was unremarkable and the patient could start on free diet. Thereafter, no other problems were registered during the routine gastroenterologic follow-up.

Discussion

Noonan Syndrome is an autosomal dominant disorder characterized by short stature, typical facial dysmorphisms, congenital heart defects and other anomalies [8-10]. Its incidence is 1:1000-2500 live births [13-22]. Approximately 50% of all NS patients have a mutation in the PTPN11 gene mapping on the long arm of chromosome 12 [23]. NS is a clinical diagnosis. There is great variability in expression and the phenotype changes significantly with time and becomes less pronounced with increasing age [9,10,24]. Facial features include hypertelorism, down-slanting palpebral fissures, eyelid ptosis, a high arched palate; low set posteriorly rotated ears and short neck. During childhood the contour of the face becomes more triangular and coarse and chest deformities (pectus carinatum or excavatum, broad chest with wide internipple space) become significant [9,13,22]. In adolescent and young adult the increasing age [9,10,24].

In childhood, intramural hematoma of the duodenum usually results from trauma [15] but may also occur spontaneously in patients with bleeding disorders [3] or who are receiving anticoagulation therapy [4] and can be a rare complication of duodenal endoscopic biopsy [2] or therapeutic injection to bleeding ulcers [16]. Anatomically, the relatively fixed retroperitoneal position of the third portion of duodenum and its adjacency to the lumbar spine make it more prone to shear injury when force is applied [17]. In addition, the rich submucosal vascular plexus and the lack of a serosal layer in the retroperitoneum may all increase the likelihood of bleeding [5].

Celiac disease is a common cause of growth retardation in children and its diagnosis is still based on multiple endoscopic duodenal biopsies [34-39].

In the reported clinical case, the duodenal hematoma and the secondary pancreatitis were consequence of a medical invasive procedure which seemed to be necessary to rule out celiac disease in a patient with short stature, growth retardation and levels of antigliadin and antiendomysium antibodies slowly higher than normal.

Postbiopsy duodenal hematoma has been reported mainly in children or young adults. The clinical symptoms of presentation are severe abdominal pain and emesis. Acute pancreatitis is frequently associated as consequence of parietal hematoma conditioning ampullary obstruction [1,2,5,6,18]. For this reason duodenal biopsy should be obtained as far from the papilla as possible [2]. The diagnosis of duodenal hematoma can be suspected on the base of upper gastro intestinal series [19] but is usually made on the base of ultrasound imaging and confirmed by CT scan [17,20]. The latter investigation allows demonstrating duodenal perforation, which would impose surgical treatment in order to avoid a significant increase in mortality [21]. In absence of perforation, in hemodynamically stable patients, a conservative management with nasogastric suction and parenteral hyperalimentation, is now recommended and can lead to resolution of all symptoms within 2-3 weeks [1,2,7], confirmed by US imaging and laboratory blood investigations.

As reported by Sgouros et al. [7], post-biopsy duodenal hematoma is a possible complication of duodenal endoscopic biopsies in Noonan syndrome. In the present experience, a hemorrhagic complication occurred in a young child without a previous diagnosis of coagulative disorders. A more detailed evaluation of his clinical features and of his past pathological history arose the suspicion of Noonan syndrome, lately confirmed.

A haematological study was conducted, that revealed a deficiency of factors V, VII and X, never previously reported in literature. These findings determined the necessity to provide a vitamin K and factor VII supplementation. Further molecular studies are at now in process.
to determine the exact etiology of this correlation. A conservative treatment was sufficient to treat this child successfully.

The genetic tests for NS resulted negative in our patient, as expected since more than 50% of NS patients have no mutations of PTPN11 and other associated genes. Furthermore, the phenotypic features of the child were typical of NS confirming the diagnosis.

A focused objective examination on the patient revealed characteristic features of NS, namely height of 140.5 cm (below 3rd centile), wide internipple space (teletelia), and chest slightly excavatum, hypertelorism, triangular shaped face, down-slaning palpebral fissures.

The present experience stresses the necessity of a correct clinical evaluation in case of hemorrhagic complications after diagnostic procedures in children not previously studied. In presence of specific malformations, NS has to be ruled out because it may represent a common cause of bleeding. An appropriate anamnesis and a general evaluation of the child can avoid significant complications.

References


