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Case Report

A Clinical Case of Recurring Abdominal Pain: when Should Doctors Suspectcongenital Intestinal Malrotation?

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Abstract

The author highlights the incredible story of a Recurring-Abdominal-Pain female patient who lived her entire life with an undiagnosed congenital Intestinal Malrotation (IM). The intrinsic difficulty of identifying rare abdominal diseases is known. Therefore, this paper reports the clinical history of an IM patient in order to help Medical Specialists and, particularly, Family Physicians to better know and suppose this congenital disease. This to prevent this condition from remaining unnoticed and unsolved in other patients in the future. In addition, the author presents a simple table with six diagnostic clues to suggest when Medical Specialists and, particularly, Family Physicians should recommend a gastroenterological examination with, eventually, a concomitant CT scan of the abdomen to their recurring-abdominal-pain patients.

Keywords: Recurring abdominal pain; Intestinal malrotation; CT scan; Congenital disease

Introduction

Rare congenital diseases require a deep medical knowledge and, most of the time, the support of different diagnostic investigations to be first detected and then successfully treated. High-impact communication, trust and self-criticism are key principles which help both Medical Specialists and Family Physicians (FPs) to build a strong and empathic doctor-patient relationship and identify rare pathologies [1,2]. Especially regarding rare abdominal diseases, the knowledge of related signs and symptoms and a concomitant high-quality doctor-patient communication are crucial to prevent them from being undiagnosed or unnoticed.

At present, one of the most challenging symptoms with a very high frequency within the young and old population is represented by Recurring Abdominal Pain (RAP). Reust et al. [3] defined RAP as at least three acute episodes of pain that occur over at least three months, affecting significantly patient's quality of life. RAP differential diagnoses are divided into organic gastrointestinal-, organic non gastrointestinal- and functional etiology [4,5]. McFerron et al. [6] summarized the most frequent differential diagnoses of RAP in the following table (Table 1). Despite completeness and clarity of the table, we didn't find any mention of Intestinal Malrotation (IM) as disorder in the referenced table. RAP can be explained by three main disorders: Organic gastroenterological, organic non-gastroenterological and functional disorders.

Congenital IM is defined in the literature as rare disease. IM manifests itself after complete or incomplete intestinal rotation around the superior mesenteric artery axis during fetal development [7,8]. With an incidence of approximately 0.2% of all births, 40% of the diagnoses happen in the first week after birth and 75%-85% of them within the first year of life. These estimates range from 0.0001% to 0.19% in adults [9-11]. IM is therefore a daily diagnosis in the pediatric field but can often remain undiagnosed or overlooked in adults. In this manuscript, the author reports the incredible story of a 58-years old female patient who lived her entire life with a congenital IM. In addition, a brief table with six IM diagnostic clues is provided. This to help doctors and particularly family physicians to better know and recognize this rare disease. The author hopes that the insights of this manuscript will allow doctors to suspect a congenital abdominal disease and suggest a gastroenterological examination with, eventually, a concomitant CT-Scan to their outpatients with similar symptoms in the future.

Case Presentation

A written informed consent was obtained from the patient after describing the nature and the purpose of the case report.

A 58-years-old female patient, farmer in her native country presented herself in our Emergency Room reporting recurring episodes (10-12 times per year) of abdominal pain since she was a child. The patient did not report any triggering abdominal traumas in the past. Symptoms were, most of the time, associated with bilious vomiting and diarrhea. The very first clinical examinations were conducted 40 years ago from her FP who recommended Paracetamol 1 g when needed to reduce the pain. However, being unable to reduce symptoms, the young woman went back to the FP in the following months to try to solve the underlying pathology. Even after repeated clinical, sonographic and endoscopic investigations, the pathology remained unclear. By not finding the adequate treatment and because of her workload as farmer and housewife, the patient started to slowly present mental depression. Her daily activities and her RAP could be one of the major risk factors of the following depression. The psychiatric symptoms were treated with Clomipramine 25 mg and Maprotiline 75 mg. Following the current guidelines, these drugs would not be currently indicated for the treatment of the psychiatric disorder. By not solving the underlying disease, the patient decided to move away from her FP up to the present year.

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Table 1: Differential diagnosis of recurrent abdominal pain.

Differential Diagnosis of Recurrent Abdominal Pain		
Organic GI disorders	**Organic non-GI disorders**	**Functional disorders**
Acid peptic disease Parasitic infections	Respiratory and urinary inflammations/infections	Idiopatic abdominal pain
Gallbladder/Pancreatic disease	Gynecologic disorders	Abdominal migrain
Eosinophilic disease Polyps, foreign body Surgical disorders	Other diseases (DM Prophyria, Trauma)	Idiopatic dyspepsia
Tumors	Collagen vascular disease	Bowel Syndrome

Organic gastrointestinal, organic non-gastrointestinal and functional etiologies are the main etiologies of abdominal pain.

At the age of 58, wife and mother of three, the patient presented herself in the emergency room of our hospital with an exacerbation of her RAP. The patient presented herself in the Emergency Room with an acute pre-renal failure caused by several episodes of biliary vomiting. Coherently with the other episodes, the pain started from the umbilical area until involving the entire abdominal area. The laboratory tests showed leukocytes 10.5 × 10^9/L, CRP 0.88 mg/dl, Creatinine 1.92 mg/dl and Sodium 124 mmol/l. By not finding the right underlying diagnosis even after a careful clinical history, physical examination and radiology investigations (e.g. sonography, abdominal radiography and endoscopy) the patient was subjected to a CT scan with contrast which allowed doctors to diagnose an Intestinal Malrotation (Figure 1).

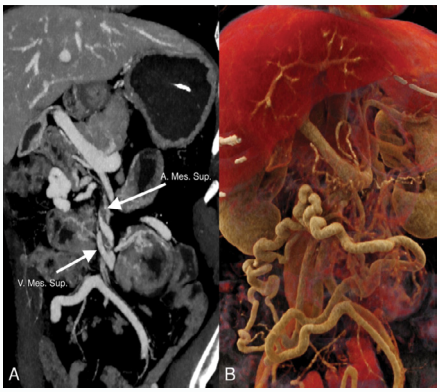

Figure 1: CT Image and 3D Reconstruction.
Intestinal malrotation has modified the course of the vessels: the superior mesenteric vein rotates around the superior mesenteric artery (A). In addition, the decrease in the thickness of the mesenteric vessels led to a dilation of the enteric collateral venous vessels (B).

The CT-Scan shows the anatomical site of the mesenteric artery and vein (A). The 3D-reconstruction of the CT-Scan allows a better comprehension of the anatomy (B).

Normally, the superior mesenteric vein follows parallel the course of the superior mesenteric artery. However, in our patient the superior mesenteric vein was rotating around the superior mesenteric artery. Surgeons performed a diagnostic laparoscopy and a following open LADD procedure with an intestinal derotation in order to definitively solve the clinical condition. After one month from surgery, the patient could halve the anti depressive therapy. After 3 months, she completely eliminated Clomipramine and Maprotiline from its therapy. RAP signs and symptoms have disappeared, and patient's quality of life has significantly improved.

Discussion

Between the different gastrointestinal congenital diseases (e.g. diaphragmatic hernia, esophageal and jejunoileal atresia, meconium ileus, omphalocele and others), IM represents a very "hard-to-diagnose" surgical condition. While analyzing the history of the woman, her difficult workload as farmer and housewife could be a significant factor for the development of her psychiatric disease. The difficulty in identifying this rare disease and the concomitant loss of confidence with her FP played also an important role. From that moment, the patient did no longer present herself to other medical doctors or hospitals to search for a solution of her disease. The creation of a solid doctor-patient confidence has therefore even the ability to influence patients' attitude towards other medical specialists or other hospitals. Moreover, the intrinsic difficulty of diagnosing this congenital anomaly is recognized. The physical, sonography and endoscopy investigations are, most of the time, not sufficient to diagnose IM. The CT Scania, consequently, mandatory for the diagnose.

The current literature and other similar case reports [6,11-13] allowed the author to list the most relevant IM signs and symptoms (Table 2). This table aims to help both doctors to better know the clinical presentation of this type of rare abdominal disorder. The most significant diagnostic clues are represented by: recurring abdominal pain, normal sonography and endoscopic findings, bilious vomiting, diarrhea, absence of previous abdominal surgeries.

Table 2: IM-Diagnostic clues of congenital Intestinal Malrotation.

Intestinal Malrotation
Chronic abdominal Pain
Diarrhea
Bilious vomiting
Normal sonography findings
Normal lower endoscopic finding
Absence of abdominal surgeries

The most significant diagnostic clues for IM supposition are represented by: recurring abdominal pain, normal sonography and endoscopic finding, bilious vomiting, diarrhea, positive familiar history for abdominal symptoms, absence of CT scan investigations, and absence of previous abdominal surgeries.

The author reports the most important diagnostic clues that help suspecting IM and suggest, eventually, a CT-Scan.

All these diagnostic elements together suggest the presence of IM and are of primary importance. By considering this paper and the above-mentioned IM diagnostic clues, a deep gastroenterological examination with, eventually, a concomitant CT scan could be key for RAP patients. This could allow identifying and acting in the early disease phases, preventing this of remaining unnoticed.

Conclusions

IM is a very difficult-to-diagnose congenital disease. This case report shows the importance of medical knowledge and of doctor-patient communication for the detection of rare pathologies. With this manuscript authors aim to help doctors and particularly FPs to better know and suspect congenital abdominal diseases. Patients with the above-mentioned symptoms and this kind of diagnostic work-up should be evaluated from a gastroenterology specialist. An additional CT scan examination would be, when recurrence is particularly high, of primary importance.

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