

Constipation and dilated bowel: Hirschsprung's disease is not always the case

Savas P. Deftereos,¹ Soultana Foutzitz,² Georgios Karagiannakis,³ Maria Aggelidou,⁴ Dimitrios C. Cassimos,⁵ Katerina Kambouri⁶

¹Department of Radiology, Democritus University of Thrace (D.U.Th.)

University Hospital of Alexandroupolis;

²Consultant Radiologist, University Hospital of Alexandroupolis;

³Consultant Radiologist, General Hospital of Lixouri "Mantzavinatio"

Cephalonia; ⁴Consultant Paediatric Surgeon, University Hospital of Alexandroupolis; ⁵Department of Paediatrics, Democritus University of Thrace (D.U.Th.), University Hospital of Alexandroupolis; ⁶Department of Paediatric Surgery, Democritus University of Thrace (D.U.Th.), University Hospital of Alexandroupolis, Greece

Abstract

A case of a 2-month-old boy with constipation and a localized abdominal distension in the right abdomen and hypogastrium is presented. Plain radiograph, ultrasound exam as well as a barium enema were suggestive of Hirschsprung's disease (HD) (ultrashort segment disease). Nevertheless, rectal suction biopsy was negative for neuronal abnormalities and unexpectedly on plain radiograph vertebral abnormalities were noticed. Subsequently magnetic resonance imaging of the spine and abdomen was performed, to evaluate possible spinal cord lesions and potential abnormalities of the perineal region musculature. A syringe throughout medullary cone was noted. The well-known issue, that the diagnosis of HD does not depend on the imaging (radiological) findings, was confirmed. The step-by-step diagnostic approach from the initial thought of ultrashort segment HD to the later imaging-based diagnosis of syringomyelia is discussed in this present paper.

Introduction

Chronic constipation with abdominal distension in children of any age is not a

rare condition. Hirschsprung's disease (HD) is one of the most common and obvious causes of the above symptoms especially in neonates and mainly boys. Abdominal x-ray and/or the barium enema study findings can strengthen the diagnostic thought of HD. The imaging evaluation of a suspected HD is preferred to initiate with a plain abdominal radiography followed by a contrast enema study of the colon. An Ultrasound exam can offer more information. Although classical radiological findings of HD (narrowed distal colon with proximal dilation) are useful in approaching the diagnosis of HD, manometry, rectal mucosal biopsy or both are fundamental for the diagnosis of HD. Nevertheless, there are many different diseases and conditions which sometimes require a thorough view (as well as review) of the medical history, diagnostic tests and radiological studies in order to exclude diseases with inflammatory, metabolic and neurogenic origin. More specifically ultrashort segment HD is a controversial entity regarding diagnostic criteria and acceptable therapy.

Herein, we present a case of an infant with constipation in which the initial diagnostic thought of HD was rejected after the completion of thorough radiological investigation.

Case Report

A 2-months-old boy presented to the pediatrician for routine vaccination. Seizing the opportunity of this appointment, the parents mentioned their worries about constipation and failure to thrive. Poor height and weight gain compared to the child's age was observed falling from the 50th centile for height and weight to the 10th centile. Physical examination revealed abdominal distension in the right flank and hypogastrium and diminished bowel sounds. On rectal examination the ampulla was empty, while the anal sphincter had normal tone.

Initially a plain radiograph and an ultrasound exam of the abdomen were performed. A dilated bowel segment was imaged, in the right flank, full of feces with a peripheral intraluminal slit of air displacing other bowel loops to the left. Absence of air in the rectum was also observed (Figure 1A). The ultrasound exam revealed a hyper-echoic «line» with posterior acoustic shadow, consistent with presence of air (Figure 2). Subsequently, 48 h after rectal examination a fluoroscopic barium enema was obtained. The barium sulfate (BaSO₄) relative high-density suspension was preferred over water soluble contrast because of its ability for fine-detailed delineation of a pos-

Correspondence: Savas P. Deftereos, Department of Radiology, Democritus University of Thrace (D.U.Th.) University Hospital of Alexandroupolis, Dragana, Alexandroupolis 68100, Greece. E-mail: sdefter@med.duth.gr

Key words: Hirschsprung's disease; syringomyelia; children.

Contributions: SPD wrote the first draft of the manuscript. All authors participated equal in the preparation of the manuscript as well as in the final version of the manuscript and this submission.

Conflict of interests: the authors declare no potential conflict of interests.

Availability of data and materials: The data used to support the findings of this study are available from the corresponding author upon request.

Ethics approval and consent: Informed consent was obtained from patient's parents.

Received for publication: 24 May 2020.

Revision received: 18 September 2020.

Accepted for publication: 25 September 2020.

This work is licensed under a Creative Commons Attribution NonCommercial 4.0 License (CC BY-NC 4.0).

©Copyright: the Author(s), 2020

Licensee PAGEPress, Italy

Clinics and Practice 2020; 10:1270

doi:10.4081/cp.2020.1270

sible transition zone. The preparation with laxatives was omitted. The barium was diluted with saline instead of water, to avoid increased water absorption by the bowel surface and subsequently was heated up to body temperature. A markedly dilated rectum and a somewhat normal caliber anorectal ring with presence of a not so discernible transition zone was demonstrated (Figure 1B and C). Reviewing the abdominal X-ray, a senior radiologist, raised the suspicion of vertebral abnormalities at the level of lumbar spine (Figure 1A). These findings previously have been understated due to infant's wrong position and the full of fecal content rectum. Specifically, a hemivertebra was noticed respectively to the first lumbar vertebra (L1) and a synostosis of L3 with L4 vertebrae. Further evaluation was completed with a Magnetic Resonance Imaging (MRI) (Figure 1C and 3) revealing a syrinx (expanding cyst like lesion) in the medullary cone and multiple vertebral abnormalities. The brain MRI did not demonstrate Chiari malformation. The diag-

nosis of Hirschsprung's Disease *passed away* and depression of autonomic nervous system activity due to syrinx was suggested.

Discussion

Constipation is an extremely common problem in the pediatric population with similar prevalence rates for boys and girls. However, constipation is a symptom and not a disease.¹ The vast majority of infants and children with constipation suffer from functional constipation, secondary to a wide variety of underlying mechanisms. From all the known and referred reasons, the most common clinical entities of organic constipation are HD, congenital anorectal malformations, neurologic disorders, encephalopathy and spinal cord abnormalities (e.g. spina bifida).² Patients with organic causes for intestinal motility disorders can present soon after birth or even in early infancy, as our case did.

The most common congenital disorder of intestinal motility is HD, also known as congenital megacolon, which, despite its rarity, is the most serious entity among the pediatric intestinal motility disorders.^{3,4} Congenital anorectal malformations include a series of congenital lesions ranging from a not so obvious malposition of the anus to complex anomalies of the hindgut and urogenital organs. In most cases, anorectal malformation present with the absence of anus in its normal position. It can also present with bowel outlet opening in the perineal region, outside the usually well-developed sphincter complex coinciding with severe anomalies of the urogenital tract.⁵ The diagnosis and classification of anorectal malformations remain on the basis of clinical assessment.⁶

Finally, physicians after a thorough clinical examination must take in account that imaging may contribute significantly to the investigation of constipation.⁷ However, scrupulous reading of radiographs is necessary in order to recognize even subtle imaging details especially spinal deformities which can be related with spinal cord pathology.

In this present case a hemivertebra and synostosis of lumbar vertebrae, were recognized on the plain radiograph, leading to further investigation. An MRI study revealed a syrinx (expanding *cyst like* lesion) at the level of conus medullaris and the thought that syringomyelia is the cause of megacolon (actually megarectum) was raised.

Syringomyelia is a disorder in which a cyst forms within the spinal cord.⁸ Generally, syringomyelia is related to

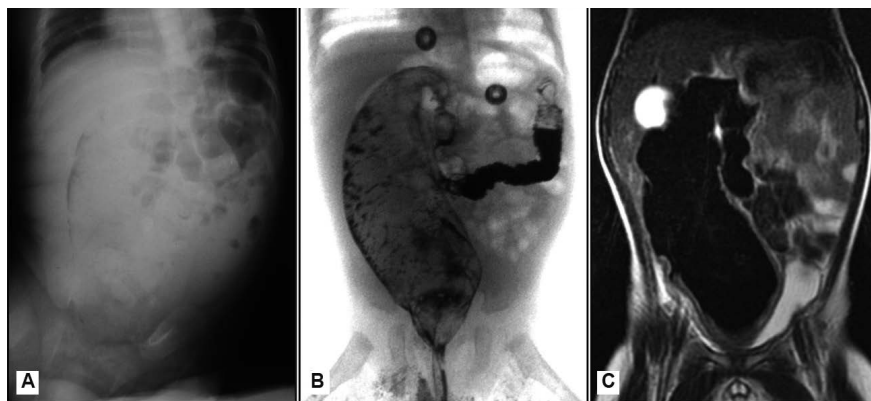


Figure 1. A) The rectum is massively distended, slightly right positioned and is occupied by large volume of impacted feces. Note a peripheral line of intraluminal air in the same portion of bowel; B) Fluoroscopic barium enema revealed a markedly dilated rectum and a somewhat normal caliber anorectal ring with presence of a not so discernible transition zone. C) MRI (T2WI) in coronal plane. Similar imaging findings such as those of barium enema.



Figure 2. Ultrasound scan, transverse image. A hyperechoic line with posterior acoustic shadow, consistent with presence of air.

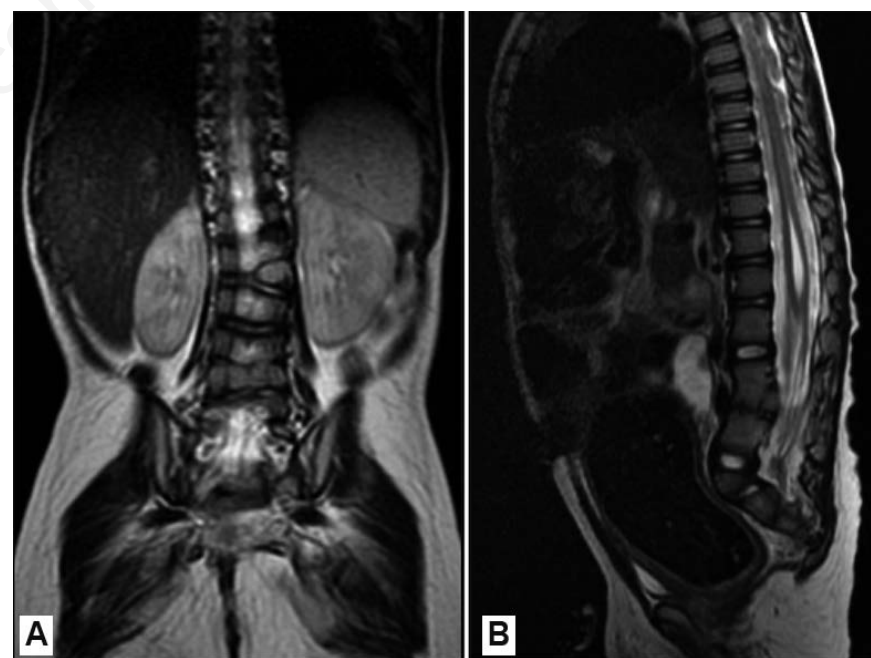


Figure 3. Magnetic resonance imaging: A) Coronal T2 weighted image. Hemivertebra is present at the level of the middle of left kidney; B) Vertebral abnormalities, tethered cone and a syrinx in medullary cone are present.

Arnold-Chiari malformation, meningocele and diastatomyelia but also can occur as a complication of trauma, meningitis, hemorrhage, tumor, or arachnoiditis.⁹

Syringomyelia usually progresses slowly; the course may extend over many years.¹⁰ Symptomatic presentation depends primarily on the location of the lesion within the neuraxis. Lumbar syringomyelia is characterized by atrophy of the proximal and distal leg muscles with dissociated sensory loss in the lumbar and sacral dermatomes. Lower limb reflexes are reduced or absent. Impairment of sphincter function and bladder control is common.^{9,10}

MRI is the main, safe and painless imaging modality to diagnose syringomyelia even before symptoms appear. Additional tests may be helpful as electromyography (EMG), and lumbar puncture.⁹⁻¹¹

Conclusions

The step-by-step diagnostic approach is

needed in controversial and not obvious cases of constipation to confirm or exclude the diagnosis of HD or other causes of megarectum (*e.g.* functional megarectum). The role of radiology is fundamental in diagnosing this cases and subsequently helpful for the clinicians in formulating an appropriate treatment strategy.

References

1. van den Berg MM, Benninga MA, Di Lorenzo C. Epidemiology of childhood constipation: a systematic review. *Am J Gastroenterol* 2006;101:2401-9.
2. Nowicki MJ, Bishop PR. Organic causes of constipation in infants and children. *Pediatric Ann* 1999;28:293-300.
3. Wetherill C, Sutcliffe J. Hirschsprung disease and anorectal malformation. *Early Hum Dev* 2014;90:927-32.
4. Gfroerer S, Rolle U. Pediatric intestinal motility disorders. *World J Gastroenterol* 2015;7;21:9683-7.
5. Holschneider A, Hutson J, Peña A, et al. Preliminary report on the International Conference for the development of standards for the treatment of anorectal malformations. *J Pediatr Surg* 2005;40:1521-6.
6. Rintala RJ. Congenital anorectal malformations: anything new? *J Pediatr Gastroenterol Nutr* 2009;40:Suppl 2:79-82.
7. Das K, Mohanty S. Hirschsprung disease - current diagnosis and management. *Indian J Pediatr* 2017;84:618-623.
8. Sternberg ML, Gunter ML. Syringomyelia. *J Emerg Med* 2017; 53:e31-2.
9. Vandertop WP. Syringomyelia. *Neuropediatrics* 2014;45:003-9.
10. Veilleux M, Stevens JC. Syringomyelia: electrophysiologic aspects. *Muscle Nerve* 1987;10:449-58.
11. Bruzek AK, Starr J, Garton HJL, et al. Syringomyelia in children with closed spinal dysraphism: long-term outcomes after surgical intervention. *J Neurosurg Pediatr* 2019;13:1-7.