“This bicycle gives me a headache”, a congenital anomaly

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Abstract

Background: The combination of a presacral mass, a sacral bone deformity, and an anorectal malformation are also known as the Currarino triad or Currarino syndrome. The syndrome is associated with a very high rate of severe and intractable constipation and urinary incontinence. However, it can also result in less common complaints and symptoms. Although the syndrome is known since 1981 and the involved genes are clarified to a great extent, the diagnosis may be delayed or missed if unrecognized.

Case presentation: A 24-year old female presented with periodical headaches. She was born with an imperforate anus, absent rectum and colon, double bladder, and sacral defect. Soon after birth she underwent several surgical procedures for anorectal and bladder reconstructions. The patient now came to her pediatric urologist for urinary incontinence and mentioned severe headaches on the side, particularly when riding a bike. Finally, she solved her headache problem by stopping to ride her bicycle. On physical examination no abnormalities were found except the ileostomy that was present ever since soon after birth and her urinary incontinence. Blood tests showed no abnormalities. Additional MRI showed a large and previously not known anterior meningocele at the level of the sacrum. Surgical treatment consisted of closure of the dura by posterior approach.

Conclusion: In this case report we describe the late discovery with an atypical presentation of an anterior meningocele in a young adult with urinary incontinence, a sacral defect, an anorectal malformation and headaches during bicycle riding. After surgical treatment of our patient the meningocele regressed. Three months after successful surgery she had no complaints and was able to ride a bike again.

Background

The Currarino syndrome is a congenital syndrome with a classical triad consisting of an anorectal malformation, a sacral defect and a presacral mass or tumor [1]. It was first classically described in three cases by the radiologist Currarino. Since defects on the HLX9 gene were found to be the cause, more than 350 cases have been described, all with variable expression [2]. The variable expression makes recognition often difficult, particularly in adolescents and adults with little or atypical symptoms [3,4]. With this case report we aim to demonstrate such an atypical presentation and discuss the etiology, management and outcome of patients with the Currarino syndrome.

Case presentation

Case

A 24-year old female presented with urinary incontinence and periodical headaches. She was born with an imperforate anus, absent rectum and colon, double bladder (Figure 1), and a dysgenetic sacrum defect (Figure 2). Soon after birth she was given an ileostomy, and later underwent an anorectal and bladder reconstructions. She now came to her pediatric urologist for urinary incontinence and mentioned severe headaches on the side, particularly when riding a bike. She solved her headache problem by stopping to ride her bicycle. Her parents then sold the bicycle since it became useless. Recently she expressed the wish to become dry and additional investigations were scheduled. On physical examination a normal looking woman presented herself with an ileostomy and urinary incontinence without further palpable abnormalities in the abdomen or back.

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Blood tests showed no abnormalities; particularly alpha foeto-protein and B-HCG were within normal ranges. MRI showed a large and previously not known anterior presacral meningocele (Figure 3) as well as a tethered cord due to a tight filum terminale (Figure 4). She refused testing for defects on the HLXB9 gene on chromosome 7q36. She was referred to a neurosurgeon and treatment of the meningocele consisted of a laminectomy S1-S2.
Anterior meningoceles can be conservative if no symptoms are present. However, most anterior meningoceles give symptoms and need surgical treatment because there will be no spontaneous regression. Conservative management is furthermore associated with a 30% mortality rate mostly due to meningitis. Therefore, surgical closure is generally recommended [9,10]. A posterior approach, transsacral or sagittal, is the preferred one because of the lowest complication rate [9,12]. The anterior laparotomy approach still has a high morbidity and mortality (22%) due to infections and fistula formation. Transvaginal and transrectal punctures have also previously been suggested but have a similar high morbidity and mortality.

Conclusion
This case report illustrates the presentation of an anterior meningocele as part of the Currarino syndrome. The Currarino syndrome can present itself partially and often remains unknown for several years. Severe constipation or mild ano-rectal malformations combined with any sacral anomaly should make clinicians aware of the possibility of the presence of a Currarino syndrome. MRI is then warranted to rule out any sacral mass or spinal abnormalities and a clinical geneticist should be consulted for further analysis of the syndrome. Our case further demonstrates that atypical symptoms such as periodical headache while riding a bicycle can lead to an unexpected diagnosis.

Consent
Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor of this journal.

Competing interests
All authors declare not to have any conflicts of interest or source of funding, fees or salaries related to the publication of this manuscript.

Authors’ contributions
WFJF and IdB contributed to conception and design of the study. Data acquisition and analysis was conducted by HPV and IdB. HPV, WFJF, EjvL, CM and IdB were responsible for data interpretation. All authors contributed to drafting of the article or revising of content. All authors gave approval for the final version of the manuscript.

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Received: 4 December 2012 Accepted: 1 October 2013
Published: 14 October 2013
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doi:10.1186/1756-0500-6-412

Cite this article as: Versteegh et al. “This bicycle gives me a headache”, a congenital anomaly. BMC Research Notes 2013 6:412.