A VERY RARE CASE OF CAUDAL REGRESSION SYNDROME WITH LEFT SIDED ISOMERISM

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PJR January - March 2019; 29(1): 67-69

ABSTRACT

We present the case of a 16 year old girl, known case of diabetes mellitus and neurogenic bladder who presented in the emergency department with right flank pain and fever and was referred for CT scan with suspicion of renal infection/abscess on the basis of her lab reports. CT scan confirmed the diagnosis of renal abscess with incidental findings of caudal regression syndrome and left sided isomerism.

Introduction

Caudal regression syndrome (CRS) is a rare anomaly with incidence up to 1:100,000[1] without any gender predilection and is a spectrum of morphological absence of caudal vertebral bones ranging from isolated partial coccygeal agenesis to the lumbosacral agenesis. The patients with CRS may present with anorectal malformations with neurogenic bladder, fecal incontinence and sensorimotor deficits and gait abnormalities. Whereas, left sided isomerism is a type of heterotaxy syndrome (also called situs ambiguous) in which there is abnormal arrangement of body organs and is characterized by a broad spectrum of abnormalities with the presence of multiple splenules on the right side instead of single left sided spleen, azygous or hemiazygous continuation of IVC, midline liver, malrotation, bilateral hyparterial bronchi and bilateral bilobed lungs.[2] A few cases have been reported in literature which suggests possible association between these two anomalies.

Case Presentation

A 16 year old young girl; known case of diabetes mellitus, neurogenic bladder and fecal incontinence presented in the accident and emergency department with the complaints of right flank pain for 1 week and undocumented fever. Her CBC showed high TLC (17.3) and urine DR showed increase amount of urinary glucose (1000 mg/dl) with numerous pus cells. CT scan was requested to further investigate the cause of fever, high TLC with abnormal urine DR with suspicion of pyelonephritis/renal abscess. CT scan revealed enlarged swollen right kidney with a small hypodense area in interpolar region with subcapsular extension and multiloculated collection in ipsilateral perinephric region consistent with right acute pyelonephritis with perforated renal abscess (Fig. 1), urinary bladder showed thick walls with multiple diverticula consistent with already known neurogenic bladder (Fig. 2a). Apart from this, there was an incidental note of absence of lower sacral bony elements and coccyx likely due to caudal regression syndrome (Fig. 2b). There was also note of midline liver, multiple right sided splenules, hemiazygous continuation of IVC posterior which was posterior to the aorta, absent hepatic segment of IVC with hepatic veins draining directly into the right atrium, right sided stomach and intestinal malrotation; all these findings were consistent with left sided isomerism. (Fig. 3a, 3b)

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Submitted 31 December 2018, Accepted 27 January 2019

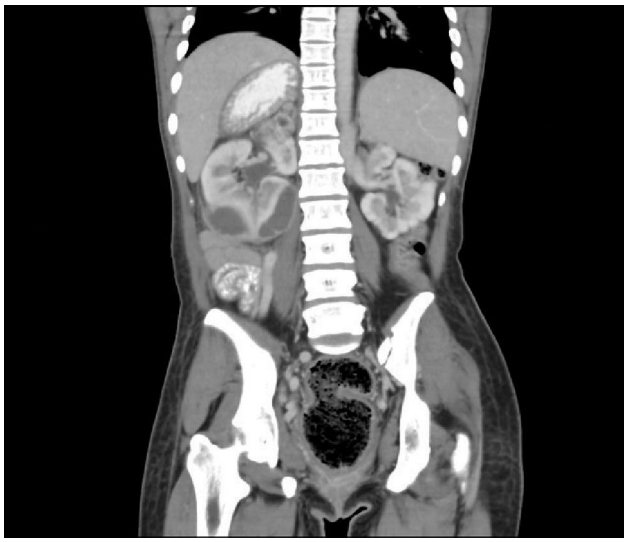

Figure 1: Coronal (CECT) showing enlarged and swollen right kidney with subcapsular multiloculated collection. Additionally right sided contrast filled stomach can be seen.

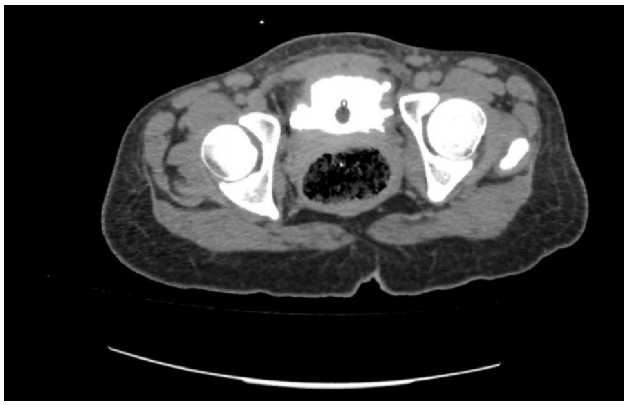

Figure 2a: Axial CECT showing thick walled urinary bladder with multiple urinary bladder diverticuli consistent with neurogenic bladder.

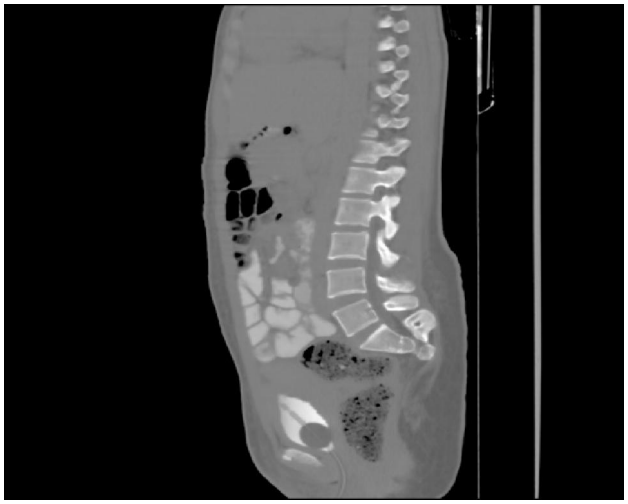

Figure 2b: Sagittal (CECT, bone window) showing absence of lower sacral bony elements and coccyx. Note elongated trabeculated urinary bladder consistent with neurogenic bladder.

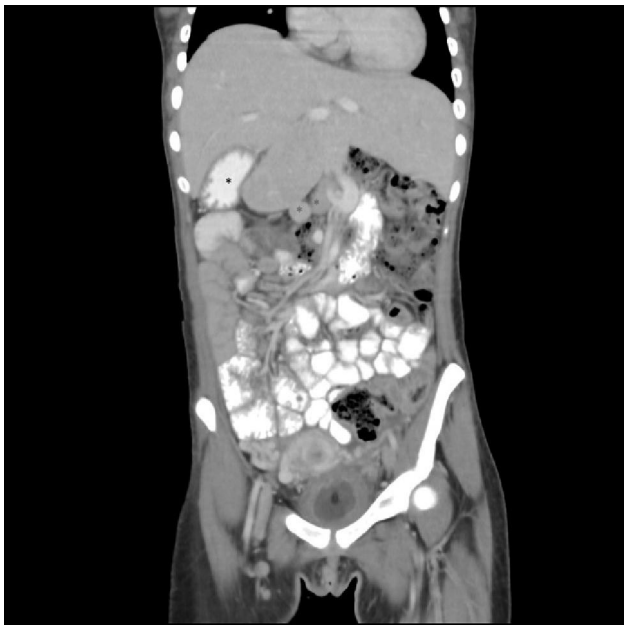

Figure 3a: Coronal (CECT) showing midline liver, multiple right sided splenules (*), contrast filled right sided stomach (*), absent hepatic segment of IVC. and intestinal malrotation.

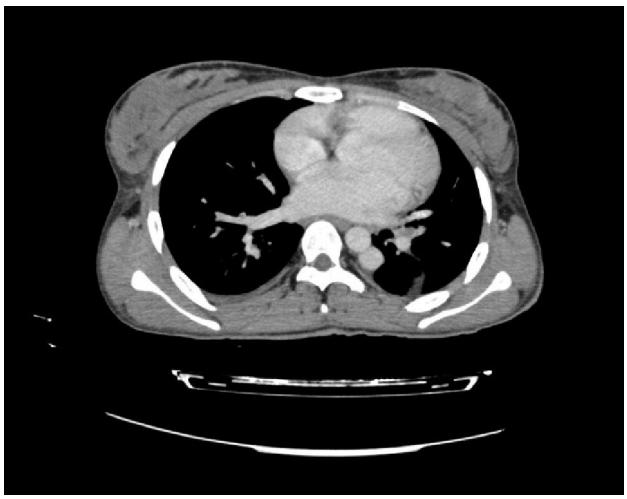

Figure 3b: Axial CECT showing hemiazygous continuation of IVC posterior which was posterior to aorta.

Discussion

Caudal regression syndrome is a rare neural tube anomaly which occurs due to insult in early pregnancy either due to disturbance in process of primary neurulation or due to derailment in process of differentiation and degradation of normally developed neural tube.[1] Many factors have been described in literature associated with CRS like infection, toxins, hyperglycemia, ischemia and maternal diabetes.[3] The patients

with CRS usually present with neurogenic bladder, anorectal malformations, sensorimotor deficits, gait problems, narrow hips, intergluteal cleft etc. Radiographic appearance of CRS depends upon severity of disorder and ranges from dysgenesis of lumbosacral vertebra to complete sacral agenesis. Furthermore, on imaging CRS can be categorized into 2 types.[4] Type 1 is when the conus medullaris terminates above the normal level and is blunted; this is associated with CSF cyst at the lower end of conus medullaris. These patients suffer more serious sacral deformities. Type 2 is when the elongated conus medullaris terminates below the normal level with tethering of filum terminale or intraspinal lipoma. These patients suffer more serious neurological deficits.

Left sided isomerism is more common in females and is characterized by atypical arrangement of viscera with presence of multiple right sided splenules, azygous or hemiazygous continuation of IVC,[2] midline liver, bowel malrotation,[5] bilateral hyparterial bronchi and bilateral bilobed lungs and altered vascular supply to organs.

Few cases have been reported in literature which suggests that heterotaxy syndrome can be associated with CRS.[6] A study conducted by Ticho et al. showed that 1.8% patients with heterotaxy syndrome had CRS, and just 1 patient (0.6%) with left sided isomerism had CRS.[7]

Conclusion

It is important to recognize both these anomalies i-e CRS and left sided isomerism and their rare association because these are associated with significant morbidity depending on the extent of severity of disease; so that appropriate care should be taken at early stages.

Conflict of Interest: Authors declared none.

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