CASE REPORT

Currarino's syndrome in twins presenting as neonatal intestinal obstruction—identical presentation in non-identical twins

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SUMMARY

We report a case of non-identical twins who presented with identical neonatal intestinal obstruction with features of anorectal stenosis, presacral mass and sacral anomaly consistent with Currarino's syndrome or triad. Plain sacral radiograph, contrast enema and MRI were diagnostic. Initial management involved a defunctioning colostomy followed by a posterior sagittal anorectoplasty with excision of the teratoma±anterior sacral meningocele and finally closure of colostomy in a staged multidisciplinary approach. The twins' father is also affected with features of Currarino's syndrome but was diagnosed during family screening. Currarino's syndrome presenting with identical neonatal low intestinal obstruction in a non-identical set of twins is rare and interesting. Antenatal diagnosis of Currarino's syndrome is difficult and may prove to be a challenge even in the postnatal period. Sacral spine radiograph, contrast enema and MRI are diagnostic. Management requires high index of suspicion, low threshold for MRI and multidisciplinary staged approach.

BACKGROUND

The Currarino's triad or syndrome (CS) is an autosomal dominant hereditary condition which is a variant of caudal regression syndrome characterised by the triad of sacral agenesis abnormalities, anorectal malformation most commonly in the form of anorectal stenosis and presacral mass consisting of a teratoma, anterior sacral meningocele or both. Neonatal presentation with identical intestinal obstruction is rare and may mimic or be associated with Hirschsprung's disease.^{1–3} We present a set of non-identical twins with complete Currarino triad one of whom had a teratoma and an anterior sacral meningocele⁴ and their father with incomplete triad. Currarino's Syndrome (CS) is characterised by intrafamilial and interfamilial phenotypic heterogeneity, incomplete penetrance of some mutations and onset in different periods of life. The presentation of this case adds to a better delineation of CS.

Figure 1 Ultrasound scan (USS) of the lumbosacral spine of the twin 2 (A). Mixed echogenic lesion. (B) Predominantly cystic lesion.

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Figure 2 Plain and contrast radiographs. (A) Abdominal and pelvic radiograph showing scimitar sacral deformity. (B) Lower gastrointestinal contrast film with mass effect of the presacral lesion and dilated rectosigmoid.

CASE PRESENTATION

Twin 2

A female preterm infant twin 2 was born to a primigravida at 36+2/40 weeks weighing 2400 g by artificial rupture of membranes and ventouse delivery in good condition following an uneventful pregnancy with normal antenatal scans. She started vomiting after feeds, became lethargic with abdominal distention and failure to pass meconium for 48 h and was referred to us. She developed gross abdominal distention at 36 h of age. Examination showed a normally sited, normal-sized anus and a non-tender but distended abdomen. Rectal examination showed a narrow rectum with inability to pass a finger through the stenosis. A feeding tube size 6 could be passed and saline rectal washout had a good result and decompressed the loaded dilated upper rectum and reduced abdominal distention.

Twin 1

The twin brother was referred with similar symptoms and physical findings but feculent vomiting and passed small amounts of stool through the normally sited normal-sized anus but had a posterior shouldering at the rectal level. Nasogastric tube

insertion brought out feculent material. He had significant abdominal distension at day 3 of life.

INVESTIGATIONS

Twin 2

The patient responded to rectal washouts and rectal suction biopsies showed normal ganglion cells; acetyl cholinesterase histochemistry showed normal staining pattern. Abdominal ultrasound scan (USS) was normal and pelvic USS showed low conus medullaris at L2/3, tethered dorsal roots related to a cystic lesion, possibly an anterior sacral myelomeningocele and the coccyx could not be seen (figure 1A, B). An abdominal X-ray showed dilated loops of bowel, a soft tissue mass in the pelvis, no air in the rectum and a scimitar-shaped sacrum with absence of the fourth and fifth sacral elements on its right side (figure 2A, B). A lower gastrointestinal contrast study showed anorectal stenosis and a dilated proximal rectum (figure 3A, B). Spinal MRI confirmed a presacral mass (figure 4A, B). The sacrum was hypoplastic with only four segments visible, the 4th segment being small. It was defective anteriorly with a cystic structure arising from the defect measuring up to 9 mm. While this might represent an anterior meningocele, there were further smaller

Figure 3 Twin 2—lower gastrointestinal contrast radiographs. (A) Anorectal stenosis and (B) presacral space widening.

Figure 4 MRI scan of pelvis (A). Lateral view confirming partial sacral agenesis (B). Cross section with dual pathology in the presacral mass.

cystic structures extending towards and probably involving the anal canal, suggesting this may be a teratoma rather than a simple meningocele suggesting dual presacral pathology. There was some minor wall enhancement of these structures.

Twin 1

The patient responded to rectal washouts and rectal suction biopsies showed normal ganglion cells and acetyl cholinesterase histochemistry showed normal staining pattern. Abdominal USS showed left hydronephrosis measuring anteroposterior diameter of 6 mm with calyceal dilation. Pelvic USS revealed a large rectum impacted by faeces displacing the bladder to the right by a presacral mass (figure 5A, B). Abdominal radiograph showed

multiple loops of dilated bowel throughout the abdomen with no gas seen within the pelvis (figure 6A, B). Lower gastrointestinal contrast enema showed calibre change or transition zone within the rectum itself and filling defects of plugs, dilated upper recto-sigmoid and reflux into the small bowel via ileocaecal valve (figure 7A, B). MRI confirmed hemisacral agenesis with anterior sacral defect and complex cystic mass in keeping with a teratoma: the features suggested Currarino triad (figure 8A, B).

DIFFERENTIAL DIAGNOSIS

Differential diagnosis included Hirschsprung's disease, meconium ileus, anorectal stenosis, VACTERL association in the initial phase but then it was clear that it was Currarino triad or syndrome.

TREATMENT

Initial management involved rectal saline washouts and bowel decompression followed by a defunctioning left descending colostomy. Both of them then underwent presacral mass excision performed by the neurosurgical team (teratoma and presacral anterior meningocele in twin 2 and presacral teratoma in twin 1) via posterior sagittal approach and had anorectoplasty carried out simultaneously. Both of them had the closure of colostomy after 8 weeks from the second operation uneventfully.

OUTCOME AND FOLLOW-UP

Both parents had genetic counselling, initial pedigree screening and physical examination. The father had rectal stenosis with sacral abnormality and was referred to the adult surgical team for further management. Histopathological examination of excised tissues confirmed mature presacral teratoma in both twins and anterior sacral meningocele in twin 2. Both twins are thriving well with excellent cosmetic and functional results at follow-up apart from constipation in twin 1.

Figure 5 Ultrasound scan (USS) of twin 1 showing presacral mass (A). Longitudinal view (B). Transverse scan- mass of 2.74 cm X 2.49 cm

Figure 6 Twin 1—Plain and contrast radiographs. (A) abdominal radiograph—note scimitar sacrum. (B) Lower gastrointestinal contrast showing mass effect of presacral lesion, stenotic anorectum and dilated rectosigmoid.

DISCUSSION

Currarino's syndrome or classic triad is a rare autosomal dominant disorder with variable expression. It could be complete, mild or minimal depending on expressivity.^{5–13} Embryologically it is a variant of caudal regression syndrome. It comprises a sacral congenital anomaly, usually partial sacral agenesis or more commonly a sacral dysgenesis in the form of scimitar sacrum associated with a presacral mass—either a teratoma or an anterior sacral meningocele or both—and an anorectal malformation usually in the form of an anorectal stenosis.

CS was originally described by Currarino as ASP association, consisting of an anorectal malformation, a sacral bony defect

and a presacral mass with suggestion of autosomal dominant transmission. It could be complete with all three elements of the classic triad allowing early diagnosis in the first decade in about four-fifth of the cases or incomplete with one or two features only which is mainly diagnosed in adults.^{5–7}

Being an autosomal dominant hereditary condition, whenever possible molecular genetic diagnosis should be sought in addition to genetic counselling and family pedigree and it should include radiological screening by sacral radiograph of every member the affected family. Sacral anomaly is caused by mutations of the HOX gene, HLXB9 linked to the 7q36 region. Over 32 mutations have been identified in HLXB9

Figure 7 Twin 1—lower gastrointestinal contrast films (A). filling defect at anorectal junction. (B) Shouldering and rectosigmoid dilatation. (C) Increased lower presacral space and sacral anomaly on lateral view.

Figure 8 Twin 1—MRI scans (A) sagittal and (B) cross sectional views confirming presacral teratoma.

including missense, non-sense, frameshift, slice site mutations or heterogeneous whole gene deletions. In over 50% of these patients, Hirschsprung's disease or its variants in the form of congenital colorectal motility disorder are present and a high index of suspicion and rectal suction biopsy should be sought for actively. A search for other associated anomalies may be fruitful.^{7–10}

CS is a spectrum disorder with a wide variation in severity from very mild to very severe disease. A significant interfamilial and intrafamilial variability in expression has been found without any definite correlation genetically or phenotypically.

CS presents with the classic triad in majority of cases but clinical presentations may be more dramatic or subtle in the form of neonatal intestinal obstruction, chronic constipation, recurrent urinary tract infection, bladder dysfunction including urinary incontinence, congenital renal, urinary or female internal genital tract anomalies, recurrent perianal sinus, tethered cord or cauda equina syndrome or as an anterior sacral meningocele, difficulty in riding a bicycle combined with headaches and meningitis to a variety of clinical specialities. Up to one-third of the patients are asymptomatic.^{1–13}

A multidisciplinary diagnostic and therapeutic protocol has been reported to have early diagnosis and appropriate treatment. The presence of a crescent-shaped defect of the sacral bone should prompt to DNA screening to fully exclude the triad. Anorectal malformations in general and anorectal stenosis in particular, with or without Currarino's syndrome, should have lumbosacral MRI performed.^{5 7}

Delayed or misdiagnosis is common with high treatment-related morbidity and mortality especially associated with undiagnosed presacral mass at centres having no or limited experience in detecting and treating these cases if a structured

approach to this rare problem has not been used. A protocol with rational timing of multidisciplinary surgical procedures may help avoid undesirable outcome.^{5 6}

Anorectal examination in all patients with a history of chronic constipation since early childhood may help in detecting presacral mass which may lead to further imaging and investigations including family screening thus allowing early diagnosis. Multidisciplinary approach with teams consisting of paediatric surgeons, neurosurgeons, neonatologist or paediatrician, anaesthesiologist, radiologist and pathologist is required to ensure adequate and timely treatment, considering the risk of malignancy in a teratoma and of nerve injury during operation.

Learning points

- ▶ Currarino's syndrome presenting with identical neonatal low intestinal obstruction in neonatal period in a non-identical set of twins is rare and interesting.
- ▶ The father had the same genetic condition and escaped detection until the age of 40 following the diagnosis and treatment of twins.
- ▶ The antenatal diagnosis of Currarino's syndrome is difficult and it can prove to be a challenge in the postnatal period.
- ▶ Sacral spine radiograph showing sacral bony deformity is the key finding which leads to suspicion of the diagnosis of Currarino's triad and induces search for the other two soft tissue lesions, namely, anorectal stenosis and presacral mass lesion.
- ▶ Management requires multidisciplinary staged approach with colonic faecal diversion as the first step and then presacral teratoma and/or anterior meningocele excision and repair combined with posterior sagittal anorectoplasty for anorectal stenosis in the second stage followed by closure of colostomy as the final step which is the best way forward.

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