Diagnosis and treatment of Currarino syndrome: a single-center case series

Department of Pediatric Surgery, Kobe Children's Hospital, Kobe, Japan

Chieko Hisamatsu, Wataru Sasaki, Ayano Uematsu, Shohei Yoshimura, Yuichiro Tomioka, Keisuke Fukui, Taichi Nakatani, Shizu Murakami, Akiko Yokoi, Tadashi Hatakeyama

Purpose Currarino syndrome is a rare genetic disorder characterized by the triad of anorectal malformations, presacral masses, and sacral malformations. We reviewed the diagnosis and management of this syndrome.

Table：10 children with Currarino syndrome treated at our hospital between 1993 and 2024

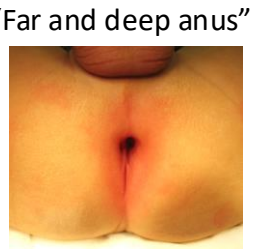
No	Sex	Family history	Age at diagnosis	Initial symptoms	Sacral anomaly	Anorectal malformation	Treatment of anus	Presacral mass	Treatment of presacral mass	Perioperative complications
1	M	father & son	newborn	**constipation**	defects below S3	**anal stenosis**	GE, laxative	**s/o lipomeningocele**	removal(N) + spinal cord untethering	transient neurogenic bladder
2	M		newborn	**anal defect**	partial defect of S4	**imperforate anus** (rectoperineal fistula)	colostomy→ PSARP	**lipomeningocele ＋dermoid cyst**	removal(N&P)	urethral injury, wound dehiscence of anoplasty
3	M	dizygotic twins	newborn	**constipation**	partial defect	**anal stenosis**	GE, laxative	**mature teratoma**	removal(N&P) + untethering	(—)
4	M		newborn	**constipation**	partial defect	**anal stenosis**	GE, laxative	**mature teratoma**	removal(P)	rectal injury →colostomy →closure
5	M	brothers	infant	**constipation**	partial defect below S3	**anal stenosis**	GE, laxative, bougie	**s/o meningocele**	(observation)	(—)
6	M		newborn	**constipation**	hemivertebra (left defect)	s/o imperforate anus→**anal stenosis**	colostomy →closure, GE, laxative, bougie, disimpaction	**s/o meningocele**	(observation)	ileus after colostomy closure
7	F	sisters	6 years old	**constipation**	defects below S3	**anal duplication**	resection	**lipomeningocele**	removal(N) + untethering	(—)
8	F		infant	**constipation**	hemivertebra (right defect)	**anal stenosis**	GE, laxative	**lipomeningocele**	2y:untethering 3y:removal(N&P)	(—)
9	M	(—)	newborn	**anal defect**	partial defect	**imperforate anus** (low type)	cut back	**mature teratoma**	removal(N&P) + untethering	(—)
10	F	(—)	newborn	**constipation**	hemivertebra (right defect)	s/o Hirschsprung disease→**anal stenosis**	GE, laxative, disimpaction 6y:inflammation →colostomy →PSARP	**mature teratoma**	1m:removal(N) + untethering 2m:removal(P)	2m:rectal injury, transient neurogenic bladder 6y:wound dehiscence of anoplasty

(GE: glycerin enema, N: neurosurgery, P: pediatric surgery)

Diagnosis

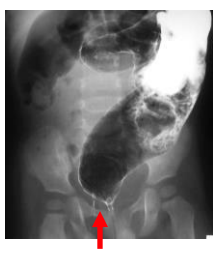
[No.1] Appearance of the anus:

"Far and deep anus"

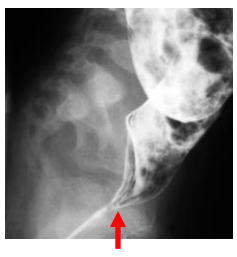


The anus did not expand.

Barium enema:

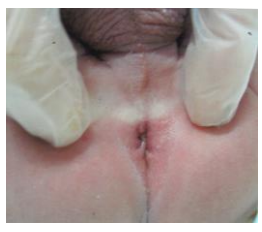


Dilation to the lower end of the rectum

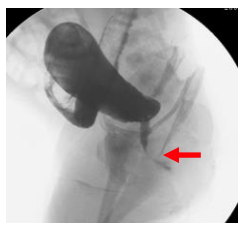


Displacement of the rectum dorsally by the sacral mass

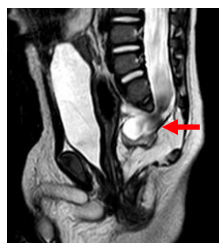
[No.6] A case requiring differential diagnosis from imperforate anus



Feces were expelled, but 6Fr. catheter did not pass.

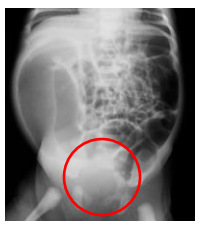


The anal fossa detected by the contrast agent from the stoma

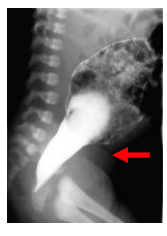


Presacral mass

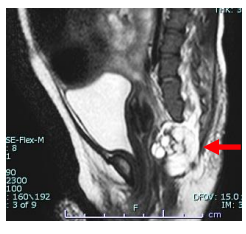
[No.10] A case requiring differential diagnosis from Hirschsprung disease



Intestinal dilatation ＋Right sacral defect



Caliber change-like



Presacral mass

Treatment

1) Anorectal malformation
- Anal stenosis (7 cases): conservative treatment (6), anoplasty (1)
- Imperforate anus (2 cases): anoplasty (2)
- Anal duplication (1 case): fistula excision

2) Presacral mass

Tumor removal (7 cases): mature teratoma (4), lipomeningocele (3)

Tumor communicating with the spinal canal (5 cases)

Spinal cord untethering (6 cases)

Simultaneous surgery for anoplasty and mass resection (1 case)

3) Perioperative complications

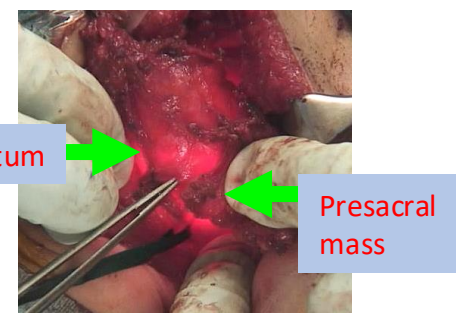
Intra-operation: rectal injury (2 cases. 1 case involved colostomy), urethral injury (1 case)

Post-operation: transient neurogenic bladder (2 cases), wound dehiscence of anoplasty (2 cases)

Discussion

- Do not overlook sacral anomaly on X-rays of constipation patients.
- Evaluation the necessity of surgery for anorectal malformations and presacral masses
- Strategies to reduce complications

  Using a biologically transparent illumination device in the rectum to prevent rectal injury during mass resection [No.2]



Postoperative complications occur at a high rate. The degree of sacral dysplasia may influence postoperative outcomes.

(Sakurai T, et al. Pediatr Surg Int. 2021)

Conclusion

1. The diagnosis of constipation requires exclusion of Currarino syndrome.
2. The treatment plan for this syndrome is determined by the abdominal and neurological symptoms and the location of the presacral mass.
3. Caution is required to avoid rectal injury during presacral mass resection.