Do Adult Patients with Congenital Colorectal Conditions Know Their Diagnosis?

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BACKGROUND:

Congenital Colorectal Conditions (2/5000 live births/year) consist of Anorectal Malformations (ARM) and Hirschsprung's Disease (HD).

- 7 Types of ARM and 3 Sub-Classifications of HD

Limited previous research shows a lack of adult patient understanding of their congenial condition:

- A recent survey found 15% of patients with ARMs were unaware of their type of ARM.[1]
- Previous studies on adult patients with Congenital Heart Diseases (CHD) reported 32–49% were unaware of their diagnosis or incorrectly identified it and only 50–61% could describe their anomaly. [2-4]
- Focus groups of adult patients with ARM and CHD highlight patients' desire to have better information about their condition, quality of life, and genetic heritability. [2, 5]

METHODS:

Patients recruited from the Adult Colorectal Research Registry from October 2019 to March 2022 were asked to complete a REDCap Survey. Responses were crossed referenced with primary post-operative reports to check for congruency.

Do you know the type of malformation that you were born with?	Yes No
Were you born with:	Anorectal Malformation Hirschsprung's Disease Other
Which type of anorectal malformation were you born with?	Recto-perineal Fistula Recto-urethral Bulbar Fistula Imperforated Anus Without Fistula Recto-urethral Prostatic Fistula Cloaca Other I Don't Know
Which type of Hirschsprung's disease were you born with?	Short Segment (Recto-sigmoid Colon) Long Segment (Descending/ Transverse Colon) Total Colonic Agangliosis Other I Don't Know

RESULTS:

37.8% patients (n=48) correctly identified their type of colorectal anomaly.

5.5% patients (n=7) were unaware that they had a congenital colorectal diagnosis.

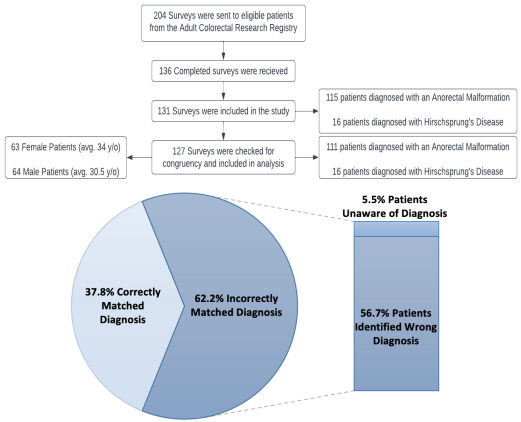
All patients with HD correctly identified their diagnosis.

29% of patients with ARM (n=32) knew their type of anomaly.

- Female patients recalled their diagnosis more often than males (42.4% vs 13.5%, p value 0.0007).

The most common female ARM patient-reported diagnosis was "Cloaca" (37.3%); however, 61% of patients were reported to have this anomaly.

The most common male ARM patient-reported diagnosis was "Imperforated Anus without Fistula" (63.5%); however, only 7.7% of male patients were reported to not have a fistula.



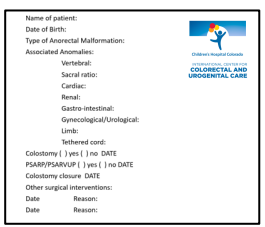
DISCUSSION:

Patients with colorectal disorders suffer lifelong medical and psychological complications that vary in severity depending on the type of anomaly. Some malformations are known to have a worse prognosis for bowel control and may have associations with renal failure, obstetric complications, and genetic predilection.[5, 6, 7]

Our results show most adults with congenital colorectal disorders are not aware of their precise diagnosis. We hypothesis this could be attributed to the complexity in terminology and anatomy as supported by prior literature.

CONCLUSION:

We recommend the development of structured educational programs starting at a young age for patients as well as medical diagnosis cards to ensure patients are adequately informed and empowered to make health decisions.


Name of patient:
Date of Birth:
Type of Anorectal Malformation:
Associated Anomalies:
Vertebral:
Sacral ratio:
Cardiac:
Renal:
Gastro-intestinal:
Gynecological/Urological:
Limb:
Tethered cord:
Colostomy ( ) yes ( ) no DATE
PSARP/PSARVUP ( ) yes ( ) no DATE
Colostomy closure DATE
Other surgical interventions:
Date Reason:
Date Reason:
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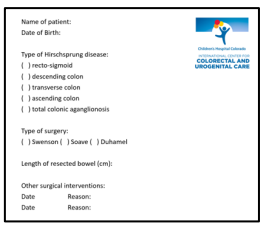
Name of patient:
Date of Birth:
Type of Hirschsprung disease:
( ) recto-sigmoid
( ) descending colon
( ) transverse colon
( ) ascending colon
( ) total colonic aganglionosis
Type of surgery:
( ) Swenson ( ) Soave ( ) Duhamel
Length of resected bowel (cm):
Other surgical interventions:
Date Reason:
Date Reason:
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