

Do adult patients with congenital colorectal conditions know their diagnosis?

M. C. Vargas¹ · L. A. Wehrli¹ · A. Louiselle¹ · J. Ketzner¹ · M. L. Reppucci¹ · L. Juddy-Glossy¹ · V. I. Alaniz¹ · D. T. Wilcox¹ · D. N. Wood¹ · A. Peña¹ · L. De La Torre¹ · A. Bischoff¹

Abstract

Purpose Limited research exists about the knowledge that adult patients have about their congenital colorectal diagnosis.

Methods This was an IRB approved, prospective study of patients in the Adult Colorectal Research Registry who completed surveys between October 2019 and March 2022. Surveys were administered through REDCap after patients consented to being contacted for research purposes. Patients provided demographic data, which was linked to surgical records, and the diagnoses provided by patients were compared with diagnoses recorded by the original surgeons.

Results One hundred and thirty-one questionnaires were collected, 115 patients had anorectal malformations (ARM) and 16 had Hirschsprung disease (HD). Seven patients who had ARM were unaware that they had an ARM or HD. The type of ARM recorded by the surgeon was unavailable for comparison with the patient's reported diagnosis in four cases. Of the 111 remaining patients with ARM, only 32 of them (29%) knew what their own type of anomaly was. Female patients recalled their diagnosis more often than male patients (42.4% vs 13.5%). All 16 participants with HD correctly identified their diagnosis severity as HD with or without total colonic aganglionosis.

Conclusion The results of this study demonstrate patient's limited understanding of their type of ARM and highlight the urgent need to enhance communication and education strategies, such as issuing patients with medical diagnosis identification cards. It is critical for clinicians to better communicate with patients to ensure that they and their relatives truly understand their precise diagnosis. Adequately informed patients can better advocate for themselves, adhere to treatments and precautionary recommendations and navigate the complexities of transitional care. Consequently they can more effectively manage their lifelong complications.

Keywords Anorectal malformations · Imperforate anus · Hirschsprung disease · Total colonic aganglionosis