An evaluation of colorectal diseases: surgical aspects and new insights into the mechanisms of fecal continence
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Summary and conclusions
Colorectal diseases can be inborn, like congenital anorectal malformations (CARMs) or they appear later in life like ulcerative colitis. Depending on the severity of the disease they can both be treated either conservatively or surgically. Although the main aim is to obtain optimal clinical outcomes, it is known that after treatment these patients may still suffer from symptoms relating to their disease like fecal incontinence. Fecal incontinence is a debilitating disorder with considerable impact on quality of life. To achieve a better understanding of the pathophysiology of fecal incontinence in these patients we must begin by improving our understanding of the physiology of fecal continence. In the first part of this thesis we investigated the fecal continence mechanisms in both healthy subjects and patients. In the second part we apply the fundamental knowledge presented in the first part to clinical research by evaluating patients with different colorectal diseases and investigating their long-term clinical and fecal continence outcomes.

PART I — FECAL CONTINENCE MECHANISMS

A functional sphincter is believed to be the foundation of fecal continence. Nevertheless, in practice this notion appears to be incomplete. It does not correspond with the fact that despite severe sphincter defects some patients are continent, while others with only minor defects are incontinent. Besides, this knowledge does not clarify why people suffer from liquid stool incontinence more often than from solid stool incontinence. The findings we present in the first part of this thesis allow us to take the next step toward understanding the issues involved in fecal continence.

In Chapter 2 we show that apart from controlling fecal continence by voluntary contractions, the puborectal muscle can also control it by involuntary contractions. To demonstrate this we performed a study in healthy subjects using the balloon retention test. We observed prolonged contractions at the level of the puborectal muscle that increased gradually during progressive filling of the rectal balloon. We also observed rotation of the anal canal in the direction of the pubic bone. The resultant change in anorectal angle caused by puborectal muscle contraction corresponded to the increasing pressure at the level of the puborectal muscle. We conclude that this gradual increase in pressure is the result of involuntary contractions of the puborectal muscle. The involuntary contractions are evoked by gradual filling of the rectal balloon, which indicates that the puborectal continence reflex is activated upon dilatation of the proximal anal canal at the level of the puborectal muscle. We postulate therefore that it might be activated through a stretch receptor.

In Chapter 3 we present the finding that the puborectal continence reflex is only activated in case of solid stool, while the anal-external sphincter continence reflex is activated in case of both solid and liquid stool. The test that mimics solid stool, i.e. the gradual filling of the rectal balloon, revealed that the pressure at the level of the external anal sphincter and the puborectal muscle increased simultaneously. Interestingly, water injected into the rectum immediately
activated the anal-external sphincter continence reflex, while the puborectal continence reflex did not respond, not even after 1000 mL of water was injected. This observation alerted us to the fact that the anal-external sphincter continence reflex controls fecal continence in case of both solid and liquid stool, while the puborectal continence reflex contributes solely to solid stool continence. One might object that the injected water probably flowed freely into the colon. We agree with this objection. Nevertheless, it is indirect proof of our hypothesis that the anal-external sphincter continence reflex is activated through a contact receptor, because even water is sufficient to activate the reflex. Conversely, the puborectal continence reflex is not activated in such an analogical situation. No response of the puborectal muscle to the water injected into the rectum indicates that the puborectal continence reflex is less readily evoked by liquid stool. A likely explanation is that during the flow of water insufficient pressure is built up, while this is the case during solid stool. This observation supports our hypothesis that the puborectal continence reflex might be initiated through a stretch receptor.

Our conclusion is that both fecal continence reflexes control solid stool, while only the anal-external sphincter continence reflex controls liquid stool. Consequently, damage to the anal-external sphincter continence reflex will lead to liquid stool incontinence, while the puborectal continence reflex will still prevent solid stool incontinence. This finding might explain the higher prevalence in the general population of liquid stool incontinence in comparison to incontinence for solid stool.

In Chapter 4, we present the finding that voluntary contractions of the puborectal muscle are regulated by a different neural pathway than involuntary contractions. We observed that voluntary contractions of the puborectal muscle were significantly decreased in patients with pudendal nerve damage, while involuntary contractions were not associated with the condition of the pudendal nerve. We conclude that the puborectal continence reflex, which evokes involuntary contractions of the puborectal muscle, is not regulated by the pudendal nerve. In previous research we found that the anal-external sphincter continence reflex is also not regulated by the pudendal nerve. Consequently, patients who suffer from pudendal neuropathy do not necessarily need to deal with complete fecal incontinence because the two fecal continence reflexes are not regulated by the pudendal nerve. Further research is required to identify the exact neural pathway in order to develop a strategy to prevent accidental surgical damage to the nerves, thus preventing patients from developing fecal incontinence.

PART II — EVALUATION OF COLORECTAL DISEASES

Congenital abnormality of the anatomy can sometimes be the underlying cause of fecal problems, as is the case in patients with CARMs. Fecal problems can also arise following surgery in patients suffering from a colorectal disease such as ulcerative colitis. The sex distribution in patients with CARMs remains unknown and the prevalence of concomitant congenital heart defects is under debate. Patients with CARMs often suffer from fecal incontinence and it is unknown whether these patients possess the fecal continence reflexes. For patients with colorectal diseases who had undergone surgery further research is required to improve surgical procedures to obtain better fecal continence outcomes. In the last part of
this thesis patients with CARMs and patients who underwent ileal pouch-anal anastomosis (IPAA) were investigated regarding their clinical and fecal continence outcomes.

Although the literature describes a male preponderance in patients with CARMs we observed an equal sex distribution in our clinic and were able to confirm this observation with the study we present in Chapter 5. By means of this study we addressed the dogma of a male preponderance of CARMs in these patients and offer a probable explanation. We found that girls tend to suffer from mild forms of CARM that are difficult to detect and thus require more time to diagnose than the severe forms. In contrast, boys suffer more from severe forms of CARM that are easy to detect. Hence, the prevalence of CARMs in girls is currently underestimated on account of late or lack of diagnosis. If it goes untreated, CARM can result in severe chronic constipation, and mild forms of CARM can be difficult to diagnose. We remedy this by encouraging physicians to be alert to the fact that patients, but especially girls, with chronic constipation might suffer from a mild form of CARM.

CARMs can be concomitant with congenital heart defects. Heart defects can cause increased perioperative morbidity and mortality, and knowledge of their presence is essential. In the study presented in Chapter 6, we found that 17% of the patients diagnosed with CARMs also suffered from a congenital heart defect. Besides, we observed that congenital heart defects were distributed equally between mild and severe forms of CARM. It is striking that more than half of the congenital heart defects in patients with CARMs had been missed during their earlier general pediatric examination. No newly diagnosed congenital heart defects were found after three months of age at the time CARM was diagnosed. We therefore recommend pediatric cardiac screening of all patients diagnosed with CARM within three months after birth.

Patients with CARMs are known to suffer from fecal incontinence, the prevalence of which can reach up to 60%. We addressed the issue of how to predict which patients will have favorable fecal continence outcomes after treatment in the study presented in Chapter 7. We found that patients with CARMs possess the fecal continence reflexes, i.e. the anal-external sphincter continence reflex and the puborectal continence reflex, regardless of the form of CARM. Although these patients possess the fecal continence reflexes, they still suffer from fecal incontinence, which indicates that the cause of their incontinence lies elsewhere. We found that these patients frequently struggle with dyssynergic defecation. This is the paradoxical contraction of the external anal sphincter and puborectal muscle while trying to defecate, which can lead to overflow incontinence. We observed that the presence of the fecal continence reflexes predicted good clinical outcomes when treated for dyssynergic defecation. We therefore recommend determining the presence of the fecal continence reflexes and dyssynergic defecation to predict whether pelvic physical therapy could help to develop fecal continence in these patients with CARMs.

Finally, fecal incontinence can be a long-term consequence in patients who underwent colorectal surgery like proctocolectomy with IPAA. On the one hand, the surgery saves the
patients’ life, while at the same time fecal incontinence may have a drastic negative impact on these patients’ quality of life. This became clear in the study presented in Chapter 8, in which we observed that more than one-third of the patients who had undergone IPAA, suffer from different forms of incontinence or soiling that severely impaired their quality of life. Besides, to prevent fecal incontinence in these patients, we observed that the more proximal the anastomosis, the better the fecal continence outcomes, and that a stapled anastomosis resulted in better fecal continence outcomes. Finally, we found that IPAA performed in three stages appears to give the best fecal continence results without increasing complications. By making good use of these surgical procedures one might be able to prevent incontinence in many patients following proctocolectomy with IPAA.

CONCLUSIONS

The fundamental research presented in the first part of this thesis demonstrates that the puborectal continence reflex, which regulates the involuntary contractions of the puborectal muscle, maintains fecal continence. This reflex is only initiated by solid stool. Conversely, the anal-external sphincter continence reflex controls fecal continence in case of both solid and liquid stool. We found that the voluntary contractions of the puborectal muscle are regulated by the pudendal nerve, while the puborectal continence reflex is not. Unfortunately, the exact neural pathway remains unknown.

The important message conveyed by the studies presented in the second part of this thesis is that the clinical outcomes of patients with different colorectal diseases can still be improved, especially outcomes regarding fecal continence. Because diagnosing mild forms of CARM can be difficult, patients may suffer from severe chronic constipation in the absence of an accurate diagnosis. This finding is especially important for girls because they suffer from mild forms of CARM more often than boys. Patients with CARMs may have other congenital anomalies; about 17% of the patients with CARMs suffer from concomitant congenital heart defects. Patients with CARMs should therefore be screened for heart defects prior to surgery, at least if they are younger than three months of age. Additionally, patients with CARMs may suffer from fecal incontinence due to dyssynergic defecation. Determining the presence of the fecal continence reflexes allows us to predict favorable fecal continence outcomes after pelvic physical therapy. Patients who underwent proctocolectomy with IPAA may also suffer from fecal incontinence, the prevention of which seems an option. Stapled proximal anastomoses during IPAA surgery are frequently associated with better fecal continence outcomes, whereby the three-stage procedure appears to give the best fecal continence results without increasing complications. Further research on patients with IPAA is required to investigate the presence of the fecal continence reflexes.

In summary, the current thesis brings us a bit closer toward understanding the mechanisms of fecal continence. This knowledge is essential for predicting which patients will suffer from fecal incontinence, for preventing fecal incontinence and for treating the patients who suffer from this physically and psychosocially debilitating disorder.