Hirschsprung's disease (HD) is a birth defect characterized by aganglionosis of the distal colon. Because of obstruction proximal to the affected segment of the colon HD often presents with a failure to pass meconium during the first 24–48 hours after birth. Incidence numbers are estimated to be 1 in 5000 live births, with a pronounced predominance of males over females [1,2].

Nowadays, after surgery for HD, the majority of patients have satisfactory clinical outcomes with mild constipation, especially after reaching adulthood [3–5]. Some reports, however, show a substantial number of patients with complaints including severe persistent constipation and fecal soiling [6–9]. Constipation in postoperative HD patients may be caused by the absence of the internal anal sphincter reflex or by a residual part of aganglionic colon remaining after surgical reconstruction. Neither of these causes, however, explains why some patients suffer from severe constipation while others have no defecation problems whatsoever.

Apart from the two causes mentioned there are other causes that could be responsible for the severe constipation seen in some HD patients. One is dyssynergic defecation, a disorder characterized by a paradoxical involuntary contraction of the external anal sphincter which leads to a functional neuromuscular obstruction (see Fig. 1) [10]. Recently, another study showed that dyssynergic defecation is responsible for severe constipation in patients with inflammatory bowel disease [11]. While the exact cause of dyssynergic defecation is unknown for the majority of the affected patients, development during childhood, pregnancy, and trauma have been identified as possible causes that might set off the defecation problems [12]. Most constipation complaints in HD patients are attributed to the disease itself. Nevertheless, it is possible that some HD patients have never mastered the act of defecation properly owing to preoperative obstipation and postoperative pain in the anal canal causing them to suffer from dyssynergic defecation as a result [13]. Thus besides being unable to relax their internal anal sphincter, HD patients might have the additional problem that their external anal sphincter contracts involuntarily during defecation.

With all this in mind, we gave HD patients with severe persistent constipation a full anorectal examination, which consisted of an anal pressure profile, balloon retention test, and defecometry. We hypothesized that not all postoperative defecation complaints were attributable to HD and that dyssynergic defecation – for which viable treatment options are available – may increase the severity of constipation in these patients.
subject was at rest while we recorded the maximum voluntary procedure. We obtained basal anal pressure measurements while the patient was at rest near the anal canal with adhesive tape to prevent slippage during the procedure. We used Unisensor K12981 solid-state (Boston type) circumferential catheters with an outer diameter of 12 F with two microtip sensors to connect the rectal balloon completely. This technique has been described previously [14–16]. It provides information about the extent to which the patient experiences rectal filling, rectal capacity, rectal compliance, and whether the anal canal responds to rectal filling by squeezing [14–16].

According to our unpublished data and previous studies the normal value of the rectal volume at maximum tolerable sensation for a child of six years is 135 mL and for an adult it is 240 mL [17,18].

1.3. Defecometry

For this test the same types of catheter and the same patient position, i.e. sitting upright on a commode, were used as in the case of the balloon retention test. First, we filled the balloon with 50 mL of water of 37 °C. We then asked the patient to evacuate the balloon. If the patient was unable to expel the balloon, we increased the volume into the balloon with 50 mL of water until the earlier measured urge sensation volume was reached. While the patient tried to evacuate the balloon, we measured the maximum rectal pressure, maximum anal sphincter pressure, and the time needed for evacuation. These variables provide insight into the parameters involved in the defecation process [19]. This test assesses whether the patient voluntarily contracts the external anal sphincter during defecation. If this is not the case, the patient suffers from dyssynergic defecation. Earlier studies showed that normal subjects can expel a balloon filled with 100 mL of barium sulfate paste in a median of 7 seconds [20]. In our
laboratory setting healthy subjects were able to expel the rectal balloon with 50 mL of water at body temperature. Failure to evacuate the balloon in one minute indicates an outlet obstruction which might possibly be caused by involuntary contraction of the external anal sphincter, otherwise known as dyssynergic defecation [21].

1.4. Statistical analysis

The data were analyzed with SPSS 20.0 for Windows (IBM SPSS Statistics, IBM Corporation, Armonk, NY). We used Q-Q plots to determine whether the values were distributed normally. Subsequently, normally distributed values were reported as means with standard deviations and abnormally distributed values were reported as medians with minimum and maximum values, as appropriate.

2. Results

During the three-year inclusion period we examined nine male and one female HD patient for severe constipation. The median age at the time of testing was 12 years and the range was 7 to 19 years. The mean duration of constipation was 4.0 ± 2.9 years; the shortest duration was six months and the longest nine years. All ten patients had histopathologically proven HD based on an absence of ganglion cells in both submucosal and myenteric plexuses as determined by rectal suction biopsies. One patient had an ultra short variant of HD based on the absence of ganglion cells at 3 cm on rectal suction biopsy and the absence of the rectoanal inhibitory reflex. This patient received conservative treatment in the form of laxatives. The nine other patients all had a rectosigmoid variant of HD and had received Duhamel pull-through procedures. We examined the pathology reports of the nine patients treated surgically and found that each report mentioned distal aganglionosis with sufficient ganglion cells in the proximal anastomosed surface. Postoperative complications were limited to three patients: one case of stenosis of the proximal anastomosis and two cases of anastomotic leakage. All three complications occurred directly following the surgical procedure and were treated subsequently. The time between the surgical corrections and subsequent complications and the anorectal function tests was 7, 9, and 17 years. The stenosis was treated with dilatation after which the symptoms decreased over time. The two cases of
anastomotic leakage were treated with laparotomies combined with abdominal flushing and a temporary ileostomy in order to treat the persistent peritonitis. These two patients recovered fully during the course of several weeks. We found no statistically significant differences in the values between patients who had a postoperative complication and patients who experienced no postoperative complaints. Two patients experienced an episode of Hirschsprung’s associated enterocolitis, one preoperative and the other postoperative. All ten patients used laxatives and/or had enemas to cope with their constipation at the time the diagnoses were made. None of the patients had severe comorbidities troubling their defecation, apart from one patient who had Down’s syndrome. This patient had a mild form of mental retardation and was capable of understanding the tasks involved in the anorectal function tests.

The anal pressure profile results showed an increased anal sphincter resting pressure with a median value of 70 mm Hg and a maximum squeezing pressure median of 220 mm Hg (see Table 1). Furthermore, we found that in all ten patients the rectoanal inhibition reflex was absent, confirming their initial diagnosis of HD.

The defecometry test results showed that four out of ten patients completely failed to expel the balloon and, in the event of expulsion, patients either required more volume or more time in order to succeed (see Table 1 and Fig. 2). Vastly increased pressures were seen in both the rectum and the anal sphincter (see Table 1). This combination of sufficient propulsive force and increased anal pressure while squeezing led to the diagnosis of dyssynergic defecation. Proper coordination of defecation was absent in all ten patients we tested by means of defecometry.

The balloon retention test results showed that overall increased volumes were required to sense the balloon. We measured an increased first sensation volume of 295 mL in one patient and 150 mL in another, while normal values range from 30 mL to 50 mL depending on the patient’s age. We found serious enlargement of the rectum in two patients with maximum tolerable volume values of 600 mL and 845 mL (see Table 1). We found overall enlargement in eight out of ten patients (see Fig. 3). One patient failed to notice constant sensation and urge sensation but he did notice the maximum tolerable volume of 600 mL.

3. Discussion

While most postoperative HD patients have good clinical outcomes and do not suffer from severe defecation complaints, a substantial number of patients seem to struggle with ongoing severe constipation [6–9]. One could argue that this is possibly caused by residual aganglionosis following surgical correction in patients treated for HD. Nevertheless, the appearance and number of ganglion cells in the proximal anastomosed surface were normal in all nine patients in our study who had been treated surgically. This meant that resection had been adequate. On the basis of rectal suction biopsies, the tenth patient was treated conservatively with laxatives for his ultra short variant of HD. While this diagnosis, along with its treatment possibilities, remains undetermined [22], pelvic muscle control was still completely absent in this patient. This pointed to dyssynergic defecation. In all ten patients the rectoanal inhibition reflex was absent as is seen in HD [23]. Consequently, all patients were, by definition, slightly constipated. Nevertheless, absence of this reflex does not explain why a substantial number of the postoperative HD patients suffer from more severe complaints than others, as the rectoanal inhibitory reflex is absent in all HD patients. We demonstrated that a piece of this complex puzzle could be dyssynergic defecation aggravating the already troublesome defecation. Following defecometry we diagnosed all ten patients in our study with dyssynergic defecation. This meant that in addition to a nonrelaxing internal anal sphincter the external anal sphincter of these patients also contracted paradoxically during defecation. While most constipation complaints in HD patients are attributed to the disease itself, it is possible that a significant number of HD patients have never mastered the act of defecation properly and, consequently, these patients have dealt with dyssynergic defecation since early childhood. While hard evidence for this theory is still lacking, the extremely enlarged rectums we found in two patients seem to indicate longstanding constipation caused by dyssynergic defecation.

Several reasons for the onset of dyssynergic defecation have been described and one of these factors could be responsible for its onset in postoperative HD patients. Firstly, Rao et al. suggested that the onset of dyssynergic defecation symptoms during childhood, in otherwise healthy patients, may be owing to faulty learning of proper defecation [12]. Secondly, Hyman suggested that pain in the rectum after surgery and washouts may lead to patients avoiding bowel movement as these are often associated with increased pain [13].
possibility might be that postoperative complications or Hirschsprung’s associated enterocolitis could make patients more prone to developing dyssynergic defecation, especially since these are associated with additional treatment and sometimes even with a redo of the surgical procedure. Our results showed that three out of ten patients (30%) had a postoperative complication. Even though this is a relatively high number, the small number of patients in our study makes it difficult to statistically define postoperative complications as the predetermining factor for dyssynergic defecation. It still remains unclear whether dyssynergic defecation in HD patients with severe complaints is caused by congenital disease, surgical correction for HD, or that it develops at a much later age regardless of the patients’ medical history. Moreover, further research on a nonselected cohort of both adequately and poorly functioning HD patients is necessary to determine the exact incidence of dyssynergic defecation in the entire HD disease population.

Persistent constipation owing to dyssynergic defecation was shown to have a negative influence on overall quality of life as it significantly impairs social life, sex life, work life, and family relationships [12]. Our data demonstrated the added risk of serious enlargement of the rectum with decreased elasticity of the rectal wall owing to long-standing constipation caused by dyssynergic defecation. We found one patient who measured a maximum tolerable volume of 845 ml at the age of ten years. Absence of the rectoanal inhibitory reflex, as is seen in HD, will probably lead to a slight increase in rectal volume in all patients. Some of these values are so vastly increased, however, that they are too high even for HD. Moreover, dyssynergic defecation in HD patients may initiate a vicious circle of fear of pain during defecation and defecation avoidance behavior that may eventually cause overflow incontinence.

Nowadays, dyssynergic defecation is treated with a high fiber diet, laxatives, and biofeedback training [24]. Several reports already recognize the positive effects of biofeedback therapy on constipation in non-HD patients [25–29]. Besides improving the defecation process, biofeedback training reportedly also improves overall quality of life [30]. One case report showed that biofeedback training has great potential for patients with constipation and fecal seepage in the presence of HD [31]. In other rectal diseases, such as inflammatory bowel diseases, dyssynergic defecation was shown to be treatable by biofeedback therapy [11]. This also points out the great potential of this therapy in HD patients with severe constipation. Further research on the effects of biofeedback training in this relatively young group of patients, who have dyssynergic defecation in addition to HD, is in progress. It will have to show to what extent it is possible to treat the complaints characteristic of this particular group of patients.

4. Final conclusion

It is important to consider the diagnosis of dyssynergic defecation when dealing with severe constipation in postoperative HD patients. Our results show that a number of HD patients have problems with wrongly contracting their external anal sphincters, in addition to a nonrelaxing internal anal sphincter characteristic for HD. If dyssynergic defecation is diagnosed at an early age, viable treatment options are available that may prevent irreversible long-term complications, such as significant enlarge-ment of the rectum and, eventually, overflow incontinence.

Author contributions

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References