ERN eUROGEN registry
Report on Expertise Area 1.7

INTRODUCTION

This report entails the 2025 ERN eUROGEN registry analysis of the Expertise Area 1.7 Anorectal malformations. This report aims to give insight in the current clinical practices using the Clinical Practice Snapshot data about the patients entered. Only HCPs that entered more than 5 patients for 1.7 Anorectal malformations were included individually in these analyses. HCPs that entered less than 5 patients, were grouped in an 'Other' HCP. This 'Other' HCP consists of BE10 (N=4) and IT11 (N=4). As patient numbers are not similar across HCPs, the results cannot be equally compared between HCPs, but the analyses give an indication of trends.

The Clinical Practice Snapshots should only contain data about the first year of treatment which starts from the date the patient first visits the ERN eUROGEN HCP for the anorectal malformation. However, sometimes information outside the 1-year window was added, and at other times, the dates are unknown. If this occurs, we interpreted this variable for this patient as 'Not performed'. An example: A patient had the first visit to the hospital (start treatment) at 23-01-2021, and the reconstructive surgery took place at 08-02-2022 (more than a year after the start of treatment). This surgery should not have been entered in the Clinical Practice Snapshot of the ERN eUROGEN registry. If this information was there, we interpreted it as 'No reconstructive surgery within the first year'.

Please keep in mind these reports are meant to inform you about some general treatment characteristics using the Clinical Practice Snapshot data, not to perform in-depth statistical analysis. If you have any suggestions about information to add to these reports, or to delete because the information is not relevant, please let us know and it will be considered for the next report.

EA 1.7; ANORECTAL MALFORMATIONS

Descriptive statistics

The table below provides an overview of the descriptive statistics for patients from Expertise Area 1.7 Anorectal malformations. Corresponding figures were made of the variables, and they are displayed on the next pages.

	Total N=207	Germany DE09 (N=51)	Italy		Netherlands			Sweden SE03 (N=35)	Others OTH (N=8)
			IT05 (N=8)	IT58 (N=18)	NL01 (N=28)	NL03 (N=21)	NL09 (N=38)		
Type of malformation									
Non-syndromic anorectal malformation; N (%)	153 (73.9%)	45 (88.2%)	3 (37.5%)	9 (50.0%)	24 (85.7%)	17 (81.0%)	27 (71.1%)	24 (68.6%)	4 (50.0%)
Syndromic anorectal malformation; N (%)*	54 (16.1%)	6 (11.8%)	5 (62.5%)	9 (50.0%)	4 (14.3%)*	4 (19.0%)	11 (28.9%)	11 (31.4%)	4 (50.0%)
Diagnostics, timing after first visit & abnormalities									
Ultrasound of kidney and bladder									
Performed; N (%)	163 (78.7%)	39 (76.5%)	7 (87.5%)	15 (83.3%)	26 (92.9%)	20 (95.2%)	21 (55.3%)	31 (88.6%)	4 (50.0%)
Days first visit to USS; Median (range)	2 days (0;203)	0 days (0;112)	17 days (0;203)	0 days (0;1)	2 days (0;29)	2 days (0;57)	2 days (0;70)	5 days (0;154)	1 day (0;1)
Abnormalities found on USS : N (%)	37 (22.7%)	12 (30.8%)	2 (28.6%)	5 (33.3%)	-	2 (10.0%)	5 (23.8%)	1 (3.2%)	2 (50.0%)
Voiding cystourethrogram (VCUG)									
Performed; N (%)	67 (32.4%)	11 (21.6%)	3 (37.5%)	4 (22.2%)	3 (10.7%)	-	25 (65.8%)	16 (45.7%)	5 (62.5%)
Days first visit to VCUG; Median (range)	43 days (0;245)	29 days (12;245)	53 days (20;133)	32 days (14;175)	41 days (7;65)	-	70 days (2;158)	26 days (3;68)	35 (0;122)
VUR diagnosed; N (%)	19 (28.4%)	6 (54.5%)	1 (33.3%)	-	-	-	5 (20.0%)	6 (37.5%)	1 (20.0%)
X-ray of spine/sacrum									
Performed; N (%)	109 (52.7%)	4 (7.8%)	3 (37.5%)	16 (88.9%)	25 (89.3%)	19 (90.5%)	18 (47.4%)	18 (51.4%)	6 (75.0%)
Days first visit to X-ray; Median (range)	3 days (0;207)	1 day (0;1)	1 day (0;10)	5 days (0;115)	3 days (0;29)	3 days (0;142)	2 days (0;70)	4 days (0;15)	46 days (0;207)
Abnormalities found on X-ray: N (%)	28 (25.7%)	4 (100%)	1 (33.3%)	4 (25.0%)	6 (24.0%)	1 (5.3%)	4 (22.2%)	5 (27.8%)	3 (50.0%)
Ultrasound of spine/sacrum									
Performed; N (%)	147 (71.0%)	34 (66.7%)	3 (37.5%)	14 (77.8%)	25 (89.3%)	20 (95.2%)	19 (50.0%)	29 (82.9%)	3 (37.5%)
Days first visit to USS&S; Median (range)	3 days (0;217)	4 days (0;217)	3 days (1;18)	9 days (0;142)	2 days (0;29)	3 days (0;57)	2 days (0;70)	5 days (2;49)	4 days (1;5)
Abnormalities found on USS&S; N (%)	31 (21.1%)	10 (29.4%)	-	3 (21.4%)	7 (28.0%)	1 (5.0%)	4 (21.1%)	6 (20.7%)	-
MRI of spine/sacrum									
Performed; N (%)	38 (18.4%)	7 (13.7%)	2 (25.0%)	3 (16.7%)	8 (28.6%)	3 (14.3%)	2 (5.3%)	10 (28.6%)	3 (37.5%)
Days first visit to MRI; Median (range)	56 days (1;313)	74 days (3;249)	168 days (148;187)	30 days (30;212)	22 days (7;46)	67 days (65;71)	2 days (1;2)	104 days (9;292)	66 days (12;313)
Abnormalities found on MRI; N (%)	25 (65.8%)	7 (100%)		2 (66.7%)	6 (75.0%)	1 (33.3%)	1 (50%)	5 (50.0%)	1 (33.3%)
Echocardiogram									
Performed; N (%)	146 (70.5%)	22 (43.1%)	7 (87.5%)	18 (100%)	25 (89.3%)	20 (95.2%)	18 (47.4%)	31 (88.6%)	5 (62.5%)
Days first visit to ECG; Median (range)	1 day (0;147)	5 days (0;102)	5 days (0;147)	1 day (0;116)	3 days (0;65)	10 days (0;102)	1 day (0;70)	0 days (0;57)	1 day (1;14)
Abnormalities found on ECG; N (%)	65 (44.5%)	10 (45.5%)	3 (42.9%)	13 (72.2%)	16 (64.0%)	1 (5.0%)	14 (77.8%)	7 (22.6%)	1 (20.0%)

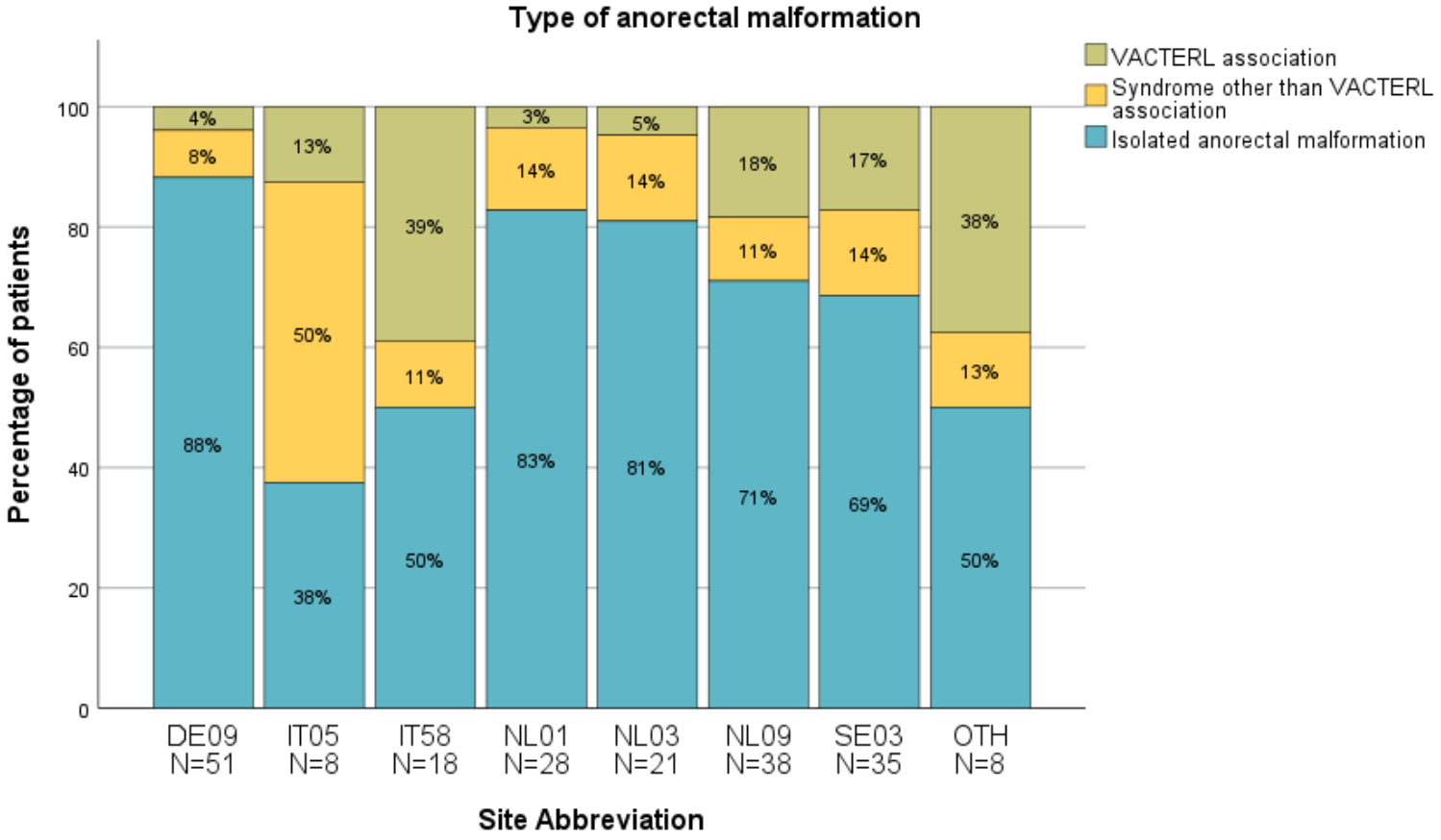
	Total	Germany	Italy		Netherlands			Sweden	Others
	N=207	DE09 (N=51)	IT05 (N=8)	IT58 (N=18)	NL01 (N=28)	NL03 (N=21)	NL09 (N=38)	SE03 (N=35)	OTH (N=8)
Surgery and treatment									
Reconstructive surgery									
Performed within 1 year after 1st visit; N (%)	180 (87.0%)	40 (76.5%)	8 (100%)	15 (83.3%)	21 (75.0%)	21 (100%)	35 (92.1%)	32 (91.4%)	8 (100%)
Performed in own HCP; N (%)	167 (92.8%)	34 ((85.0%)	7 (87.5%)	15 (100%)	21 (100%)	17 (81.0%)	34 (97.1%)	31 (96.9%)	8 (100%)
Age at surgery; Median (range)	95 days (1;6042)	109 days (1;6042)	192 days (140;314)	71 days (3;154)	112 days (45;545)	32 days (1;216)	110 days (1;253)	42 days (1;920)	144 days (1;414)
Stoma/enterostomy									
Patients with stoma/enterostomy; N (%)	66 (31.9%)	11 (21.6%)	2 (25.0%)	7 (38.9%)	5 (17.9%)	8 (38.1%)	7 (18.4%)	21 (60.0%)	5 (62.5%)
Patient with closed stoma; N (%)	60 (90.9%)	9 (81.8%)	2 (100%)	6 (85.7%)%)	5 (100%)	8 (100%)	7 (100%)	19 (90.5%)	4 (80.0%)
Days stoma was in place; Median (range)	164 days (35;343)	218 days (69;303)	300 days (256;343)	159 days (111;224)	150 days (35;188)	213 days (167;299)	118 days (69;337)	120 days (78;225)	147 days (111;317)
Stool regulators									
Treated with stool regulators; N (%)	131 (63.9%)	34 (66.7%)	7 (87.5%)	10 (55.6%)	23 (82.1%)	11 (52.4%)	17 (44.7%)	23 (69.7%)	6 (75.0%)

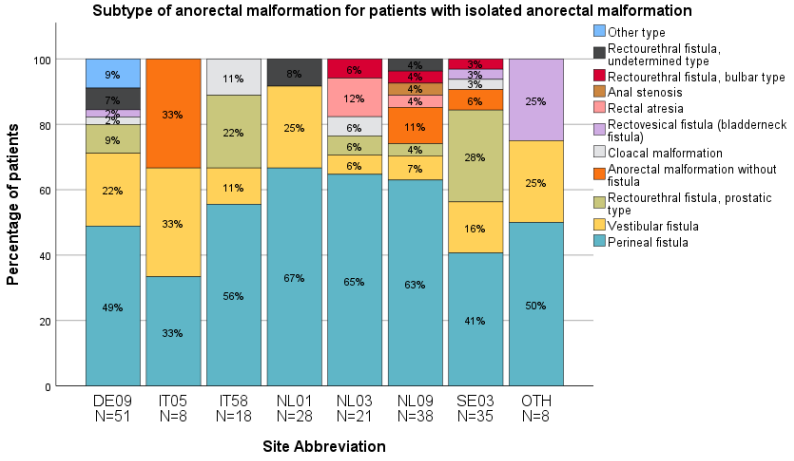
* in the corresponding figure (Type of anorectal malformation), one patients is represented twice, as this patient has been diagnosed with both a VACTERL association and another syndrome.

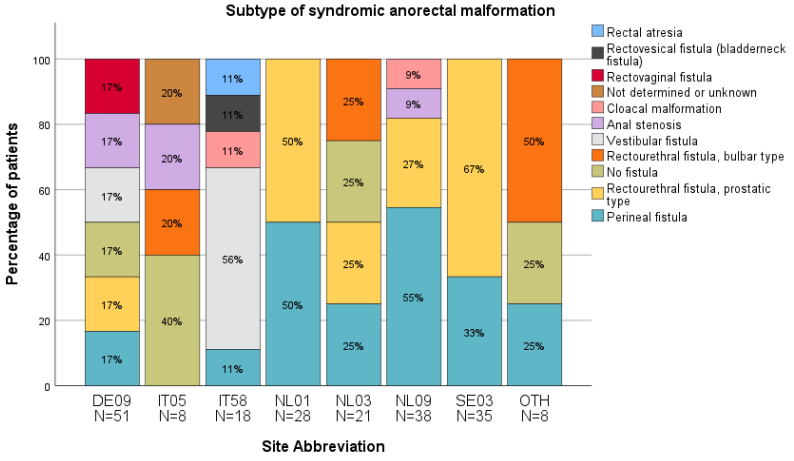
Type of malformation

Type of anorectal malformation

Most patients were diagnosed with an isolated form of anorectal malformation. Overall, there was a large variability in the types of isolated anorectal malformations, but most patients were diagnosed with a perineal fistula. For patients with a syndromic form of anorectal malformation, there was a wide variety of types.



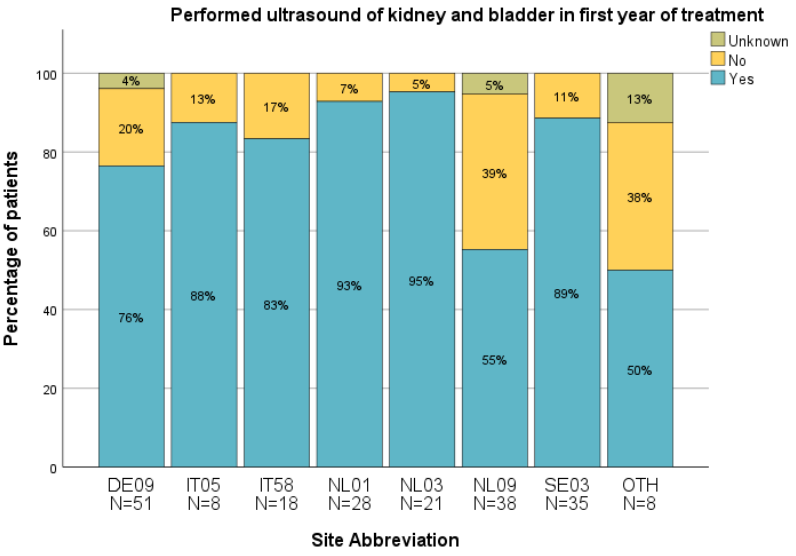


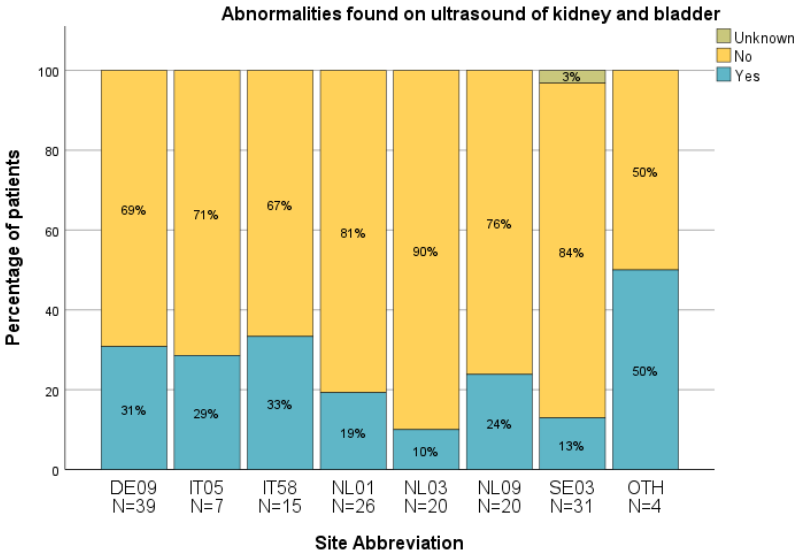


Diagnostics, timing after first visit & abnormalities

Ultrasounds of kidney and bladder and abnormalities found in the first year of treatment

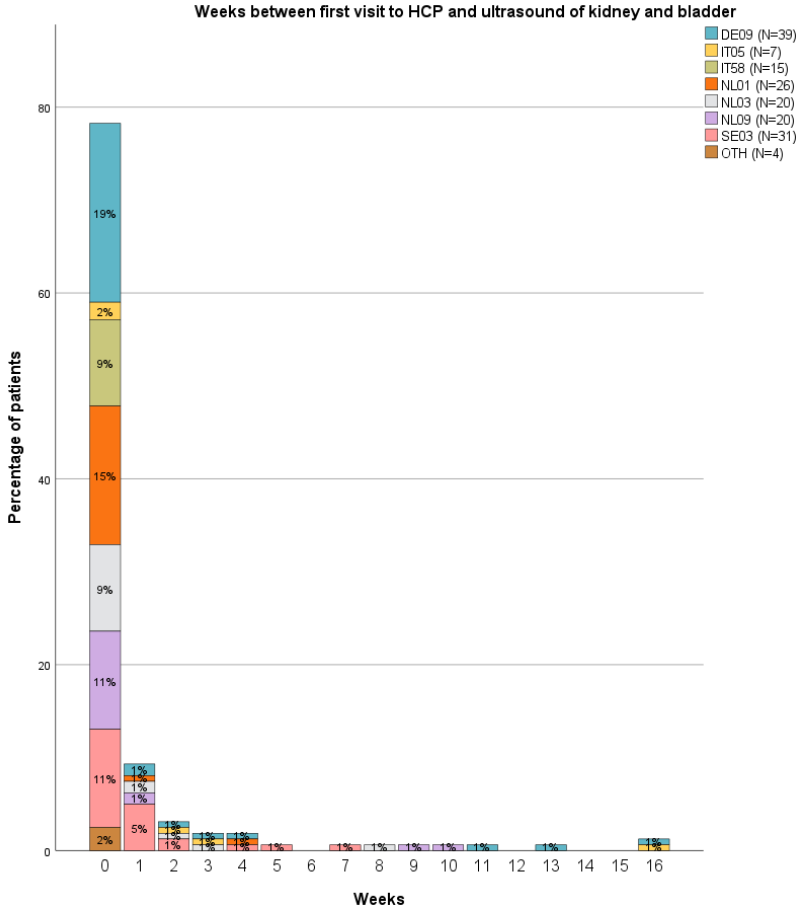
An ultrasound of the kidney and bladder was performed in most patients. When an ultrasound was performed, only in a few patients abnormalities were found.





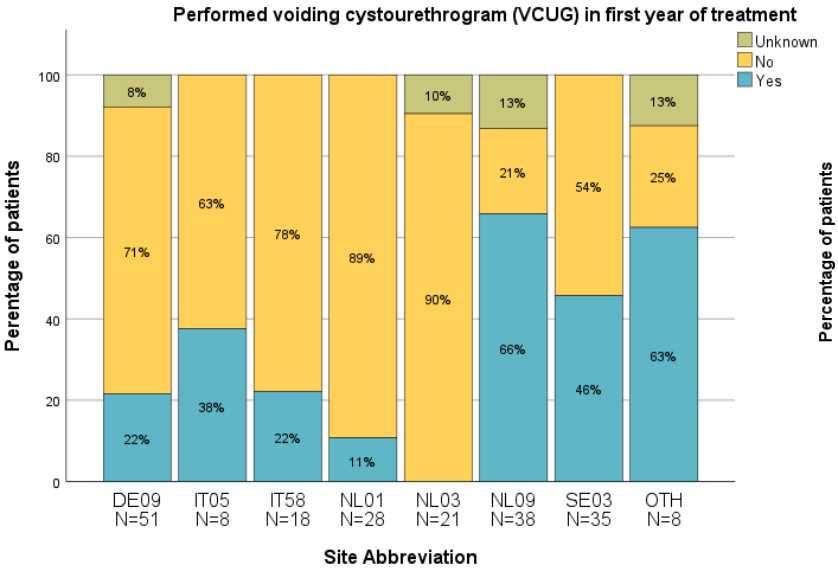
Weeks between first visit to HCP and ultrasound of kidney and bladder

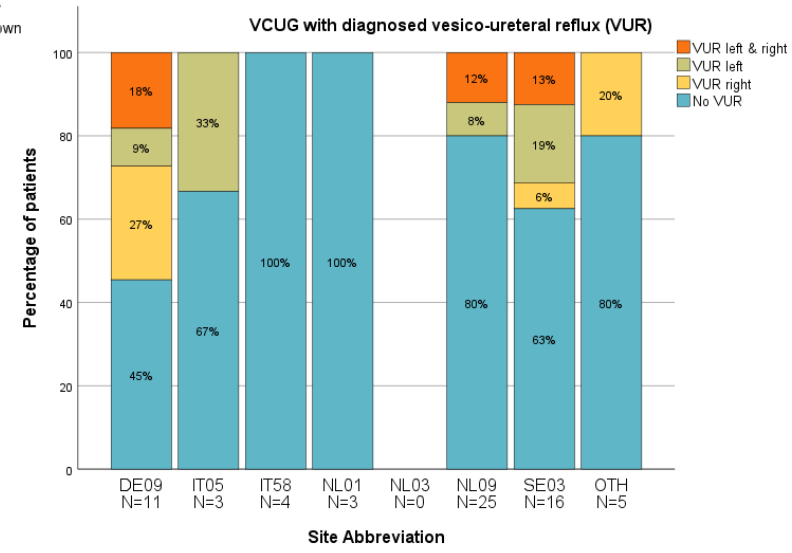
Most ultrasounds were performed in the same week as the first visit to the HCP, with ultrasounds being performed until 16 weeks after the first visit to the HCP.



Voiding cystourethrogram (VCUG) and vesicoureteral reflux (VUR) in the first year of treatment

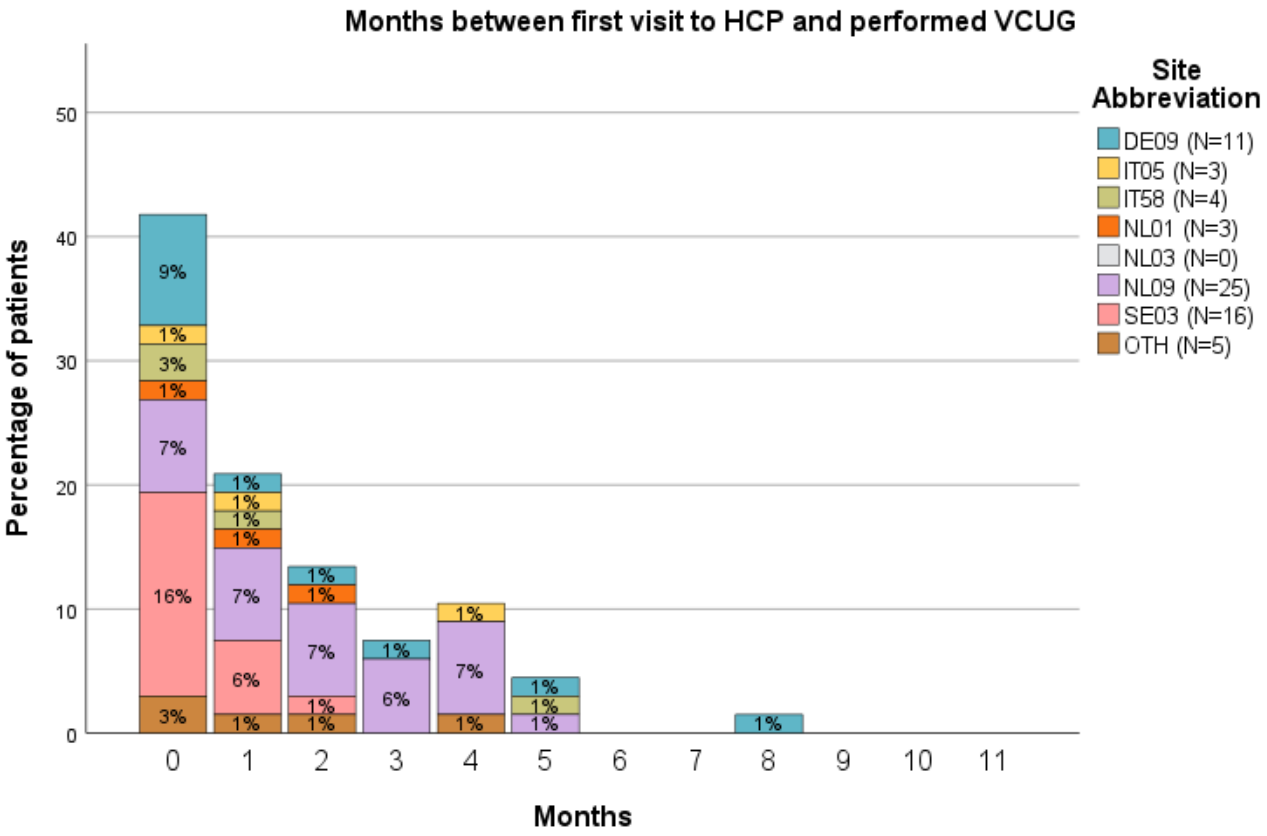
VCUG was not performed in most patients. If a VCUG was performed, no VUR was diagnosed in most cases.





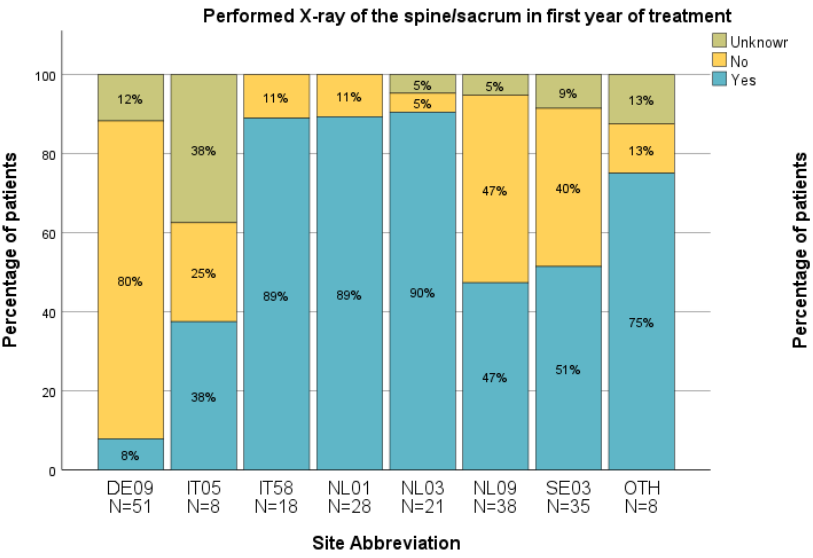
Months between first visit to HCP and VCUG

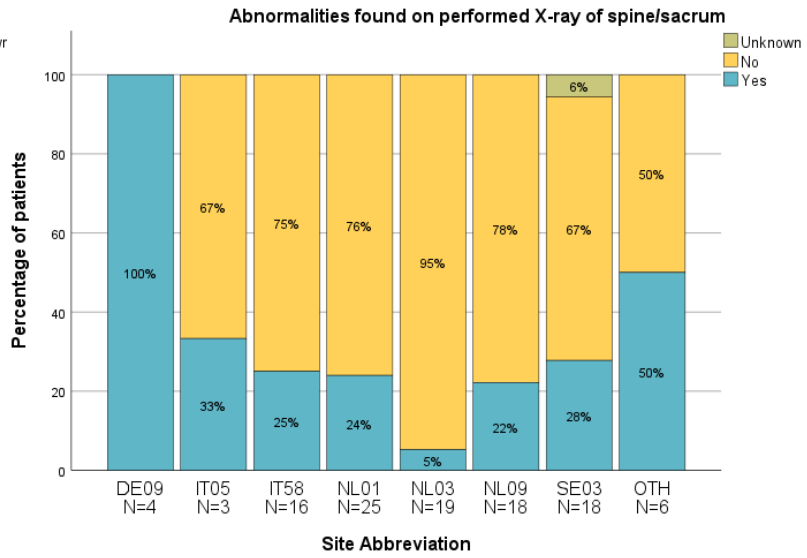
Almost all VCUGs were performed within the first half year after the first visit to the HCP.



X-ray of spine/sacrum and abnormalities found in the first year of treatment

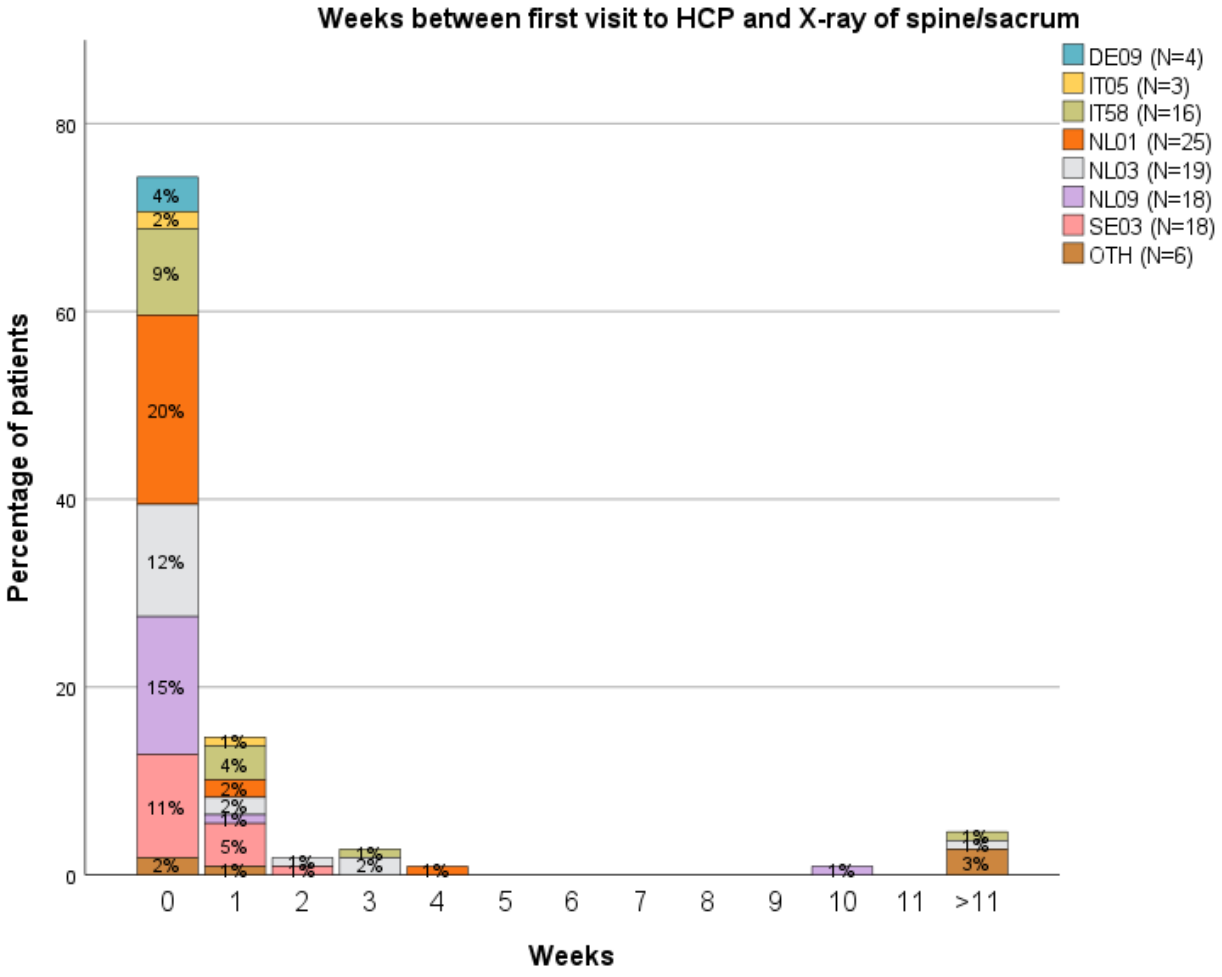
An X-ray was performed in part of the patients. Most X-rays did not reveal any abnormalities.





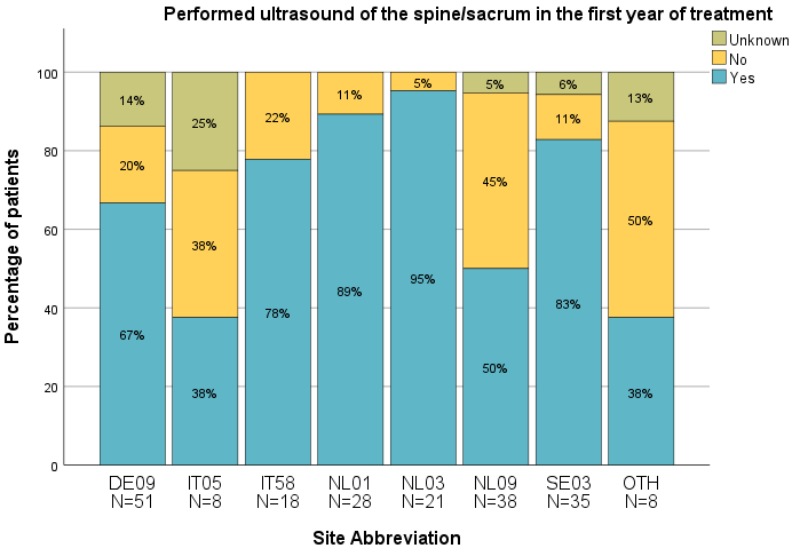
Months between first visit to HCP and X-ray of spine/sacrum

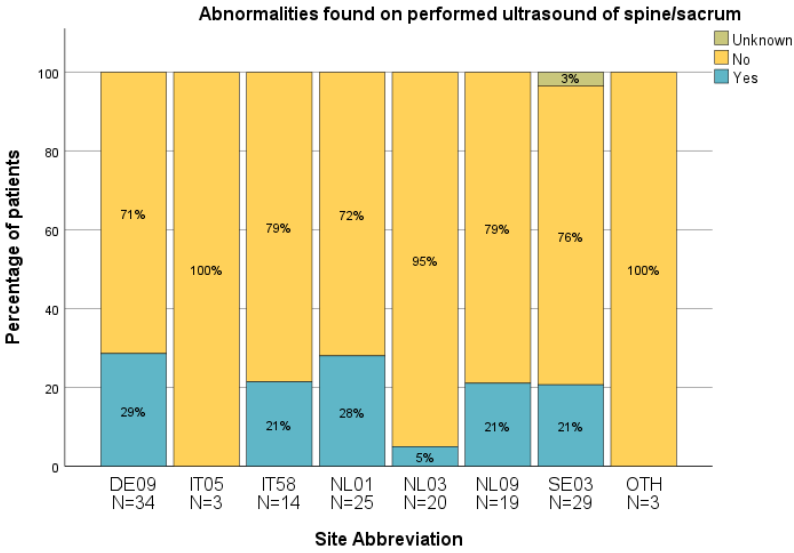
The X-ray was mostly performed in the same week as the first visit to the HCP.



Ultrasound of spine/sacrum and abnormalities found in the first year of treatment

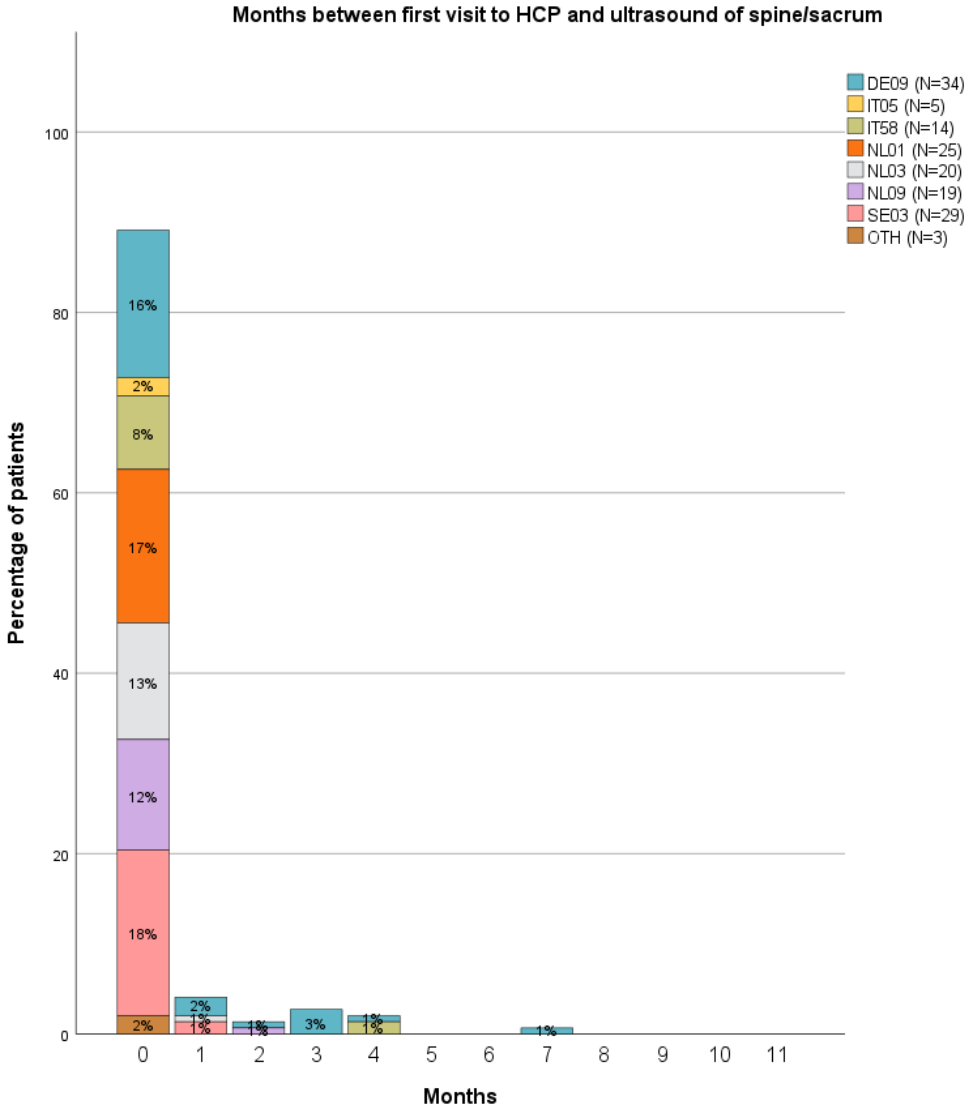
In most patients, an ultrasound of the spine/sacrum was performed. Most of the time, no abnormalities were found.





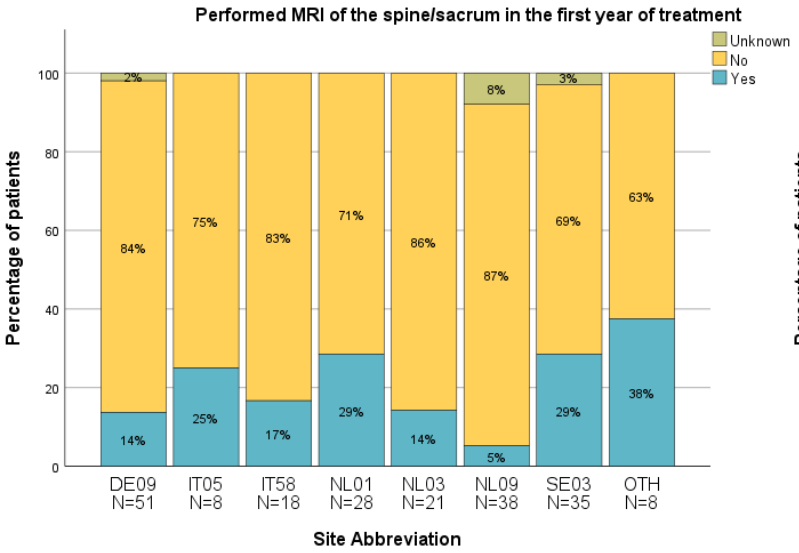
Months between first visit to HCP and ultrasound of spine/sacrum

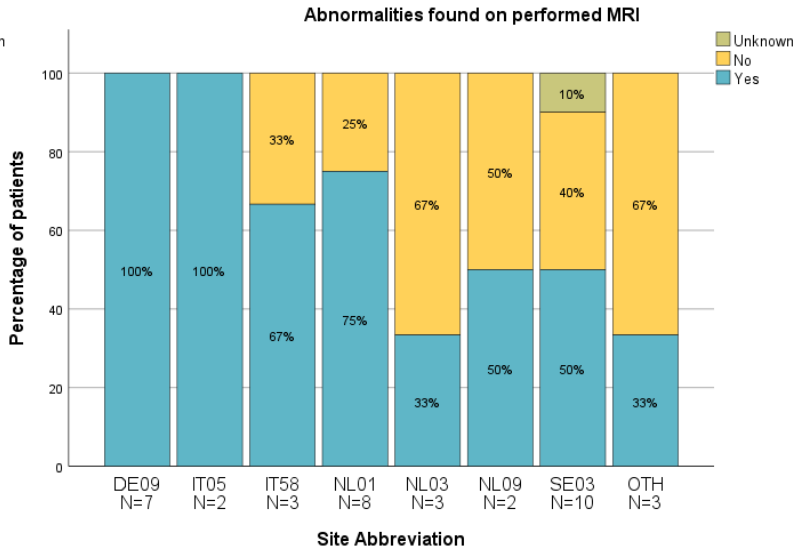
The ultrasound of the spine/sacrum was mostly performed in the same month as the first visit to the HCP.



MRI of spine/sacrum and abnormalities found in the first year of treatment

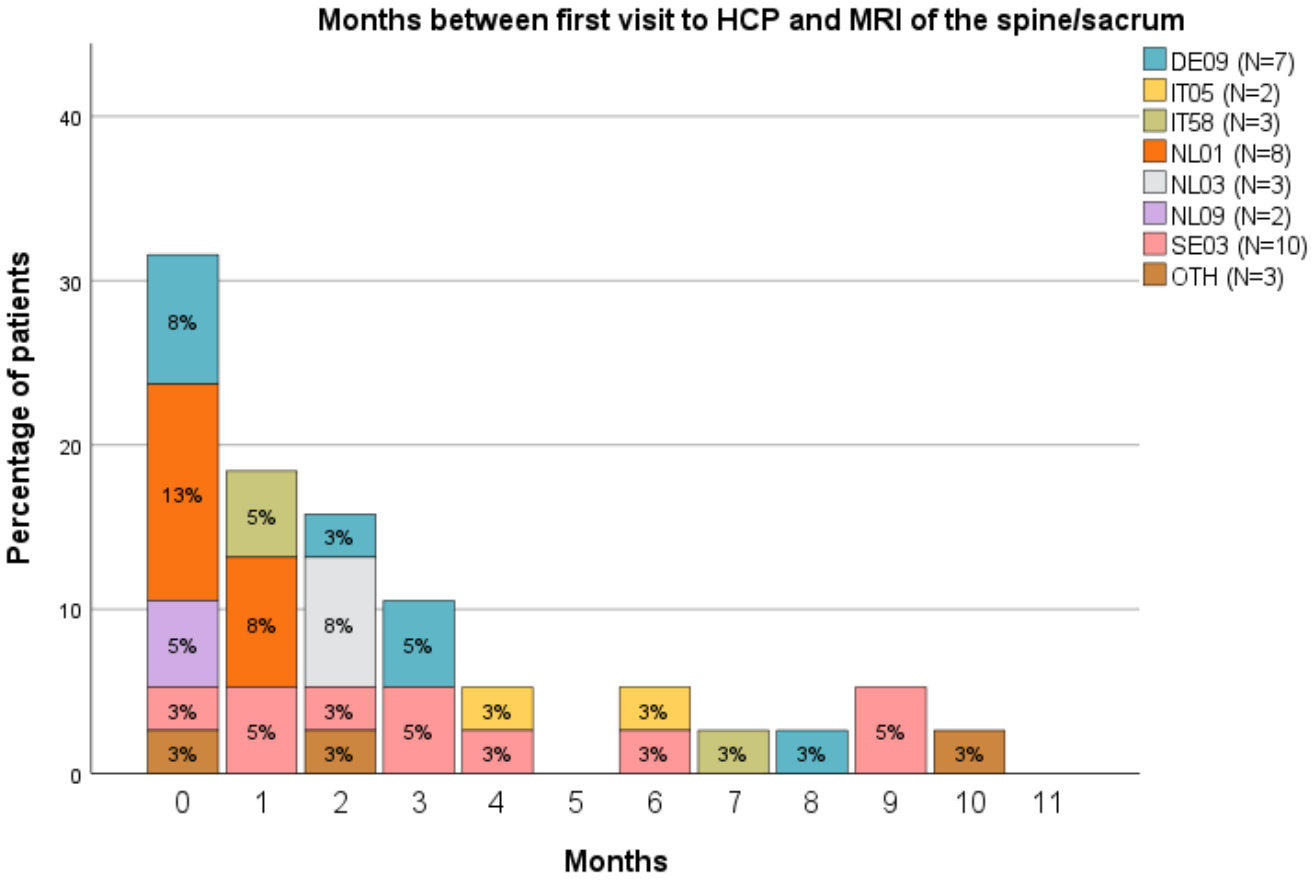
For most patients, no MRI was performed. If an MRI was done, it did reveal an abnormality in most cases.





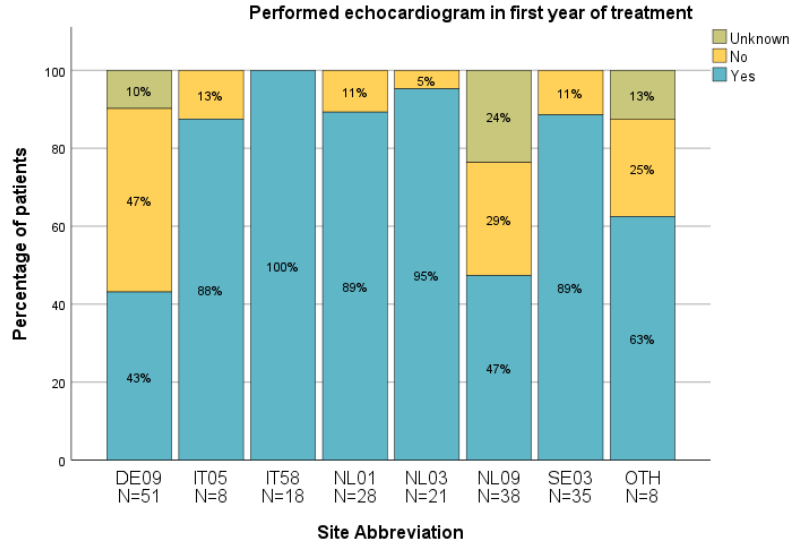
Months between first visit to HCP and MRI of spine/sacrum

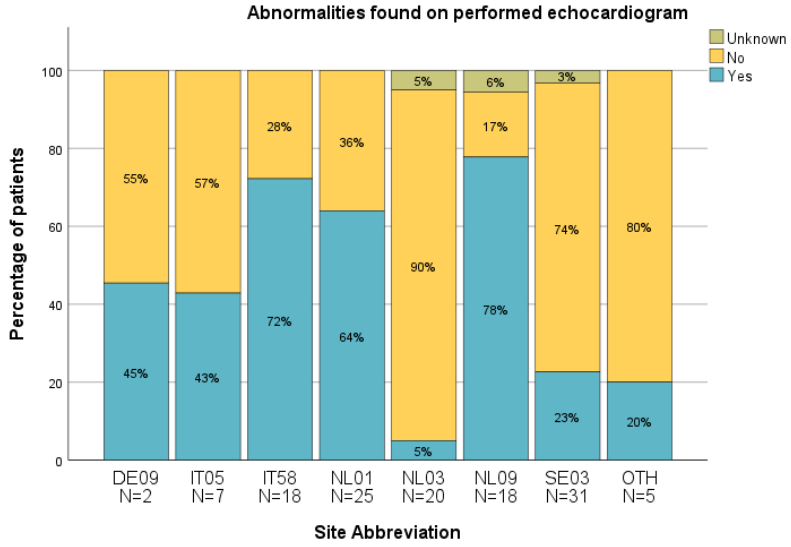
The few MRIs that were performed, were mostly performed within the first half year after the first visit to the HCP.



Echocardiogram (ECG) of spine/sacrum and abnormalities found in the first year of treatment

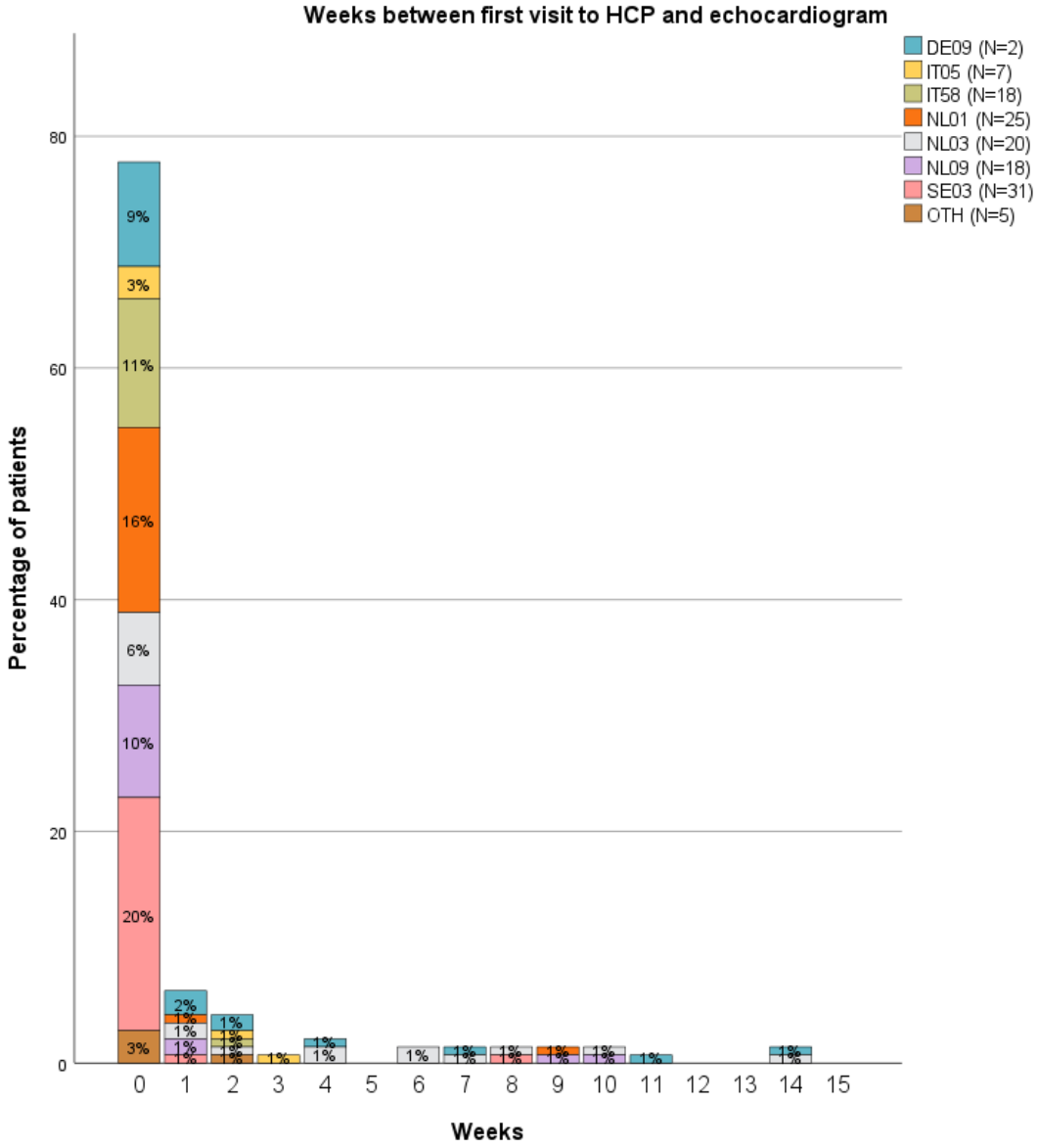
For most patients, an ECG was performed. In part of the patients, abnormalities were found.





Weeks between first visit to HCP and ECG

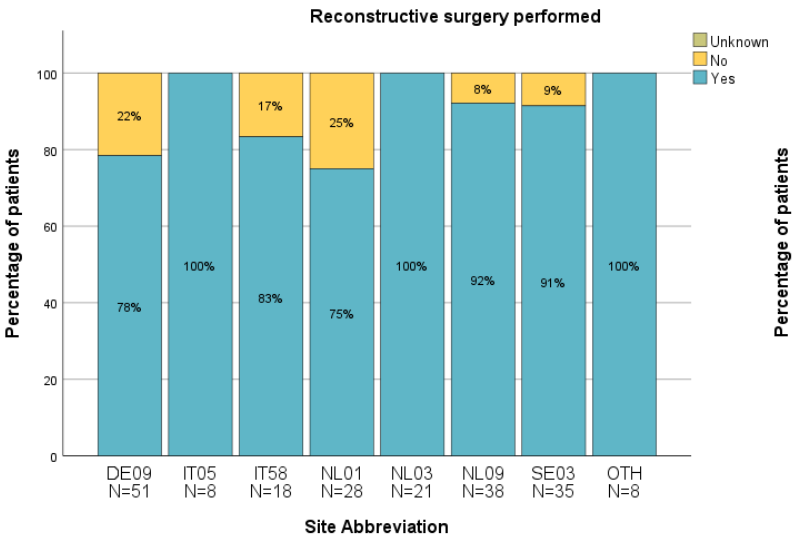
Most ECGs were performed in the same week as the first visit to the HCP.

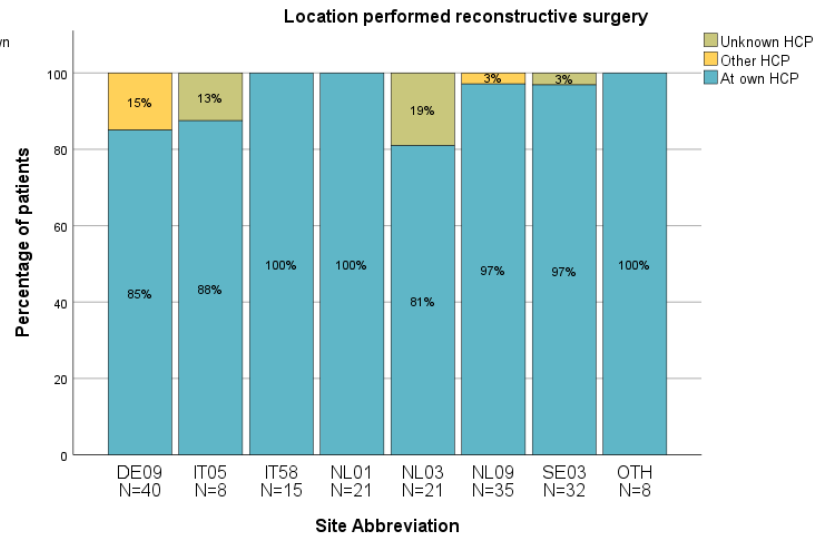


Surgery and treatment

Reconstructive surgery in the first year of treatment

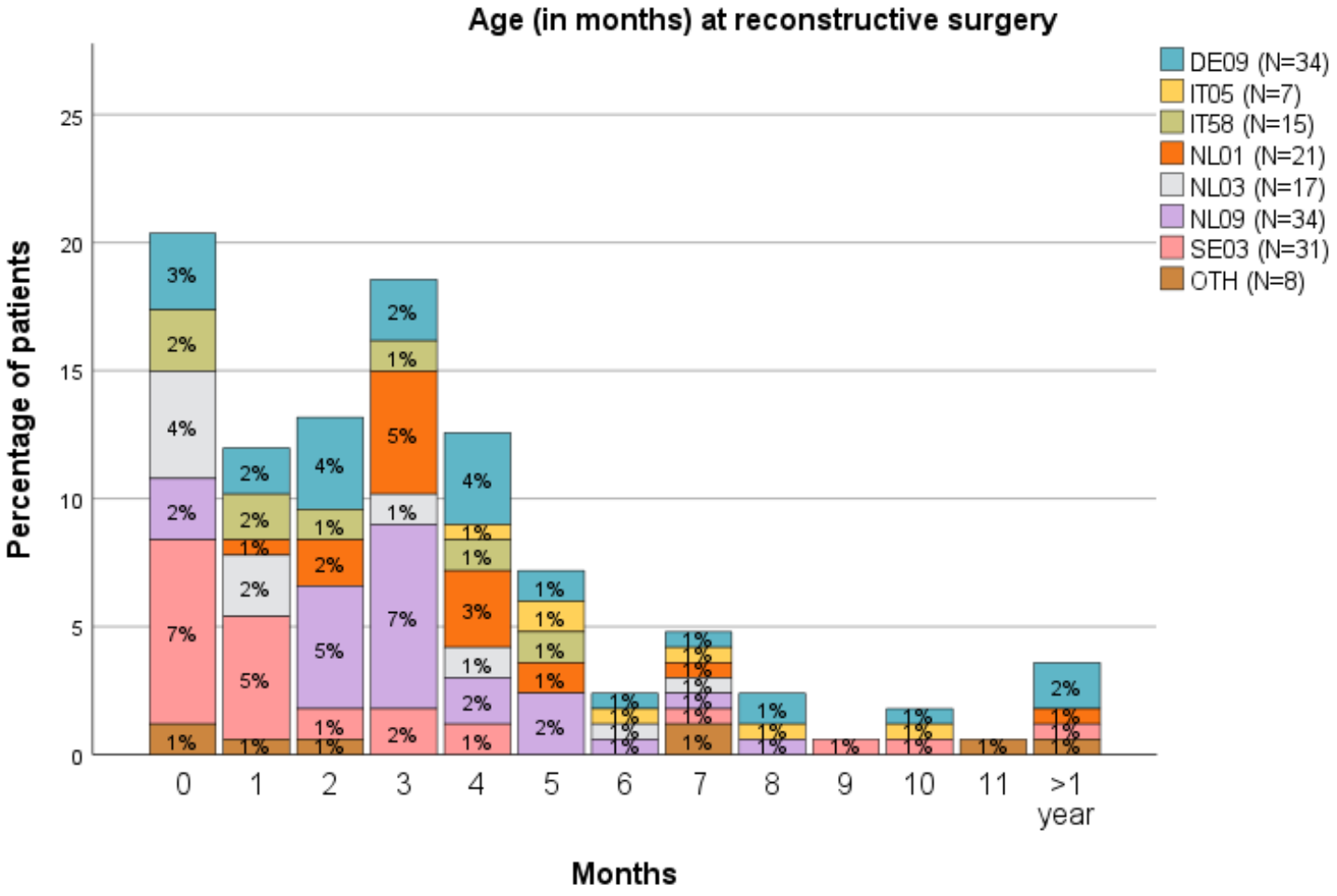
Most patients had a reconstructive surgery for their anorectal malformation within one year after their 1st visit to the HCP. A few reconstructive surgeries were performed in another HCP than the ERN eUROGEN HCP that is registering the patient.





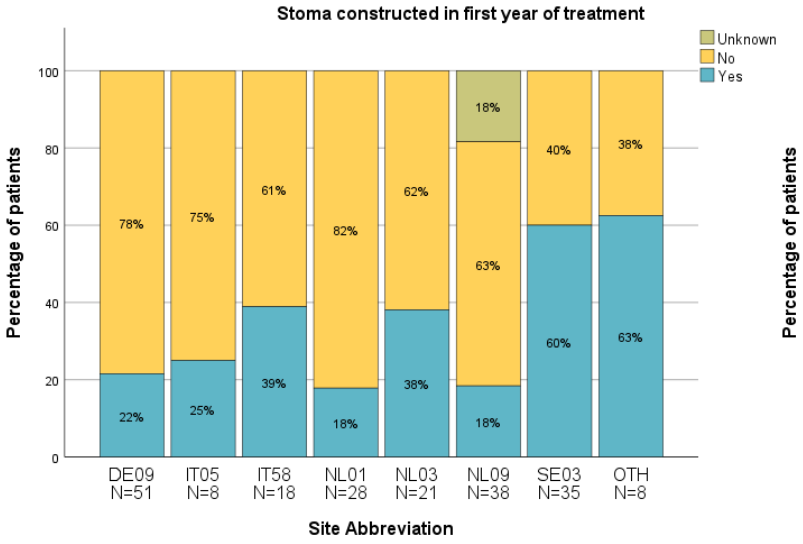
Age at reconstructive surgery when performed in the first year of treatment in own HCP

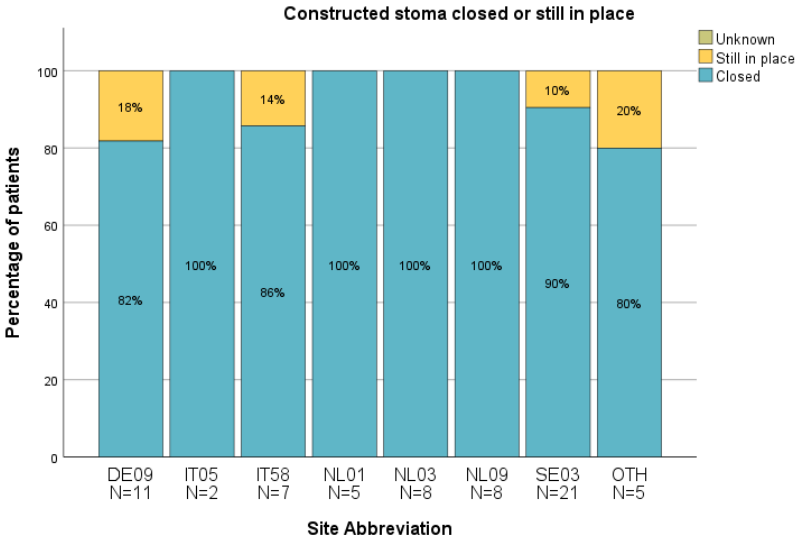
Most patients had the reconstructive surgery within the first half year of their life.



Stoma construction in the first year of treatment

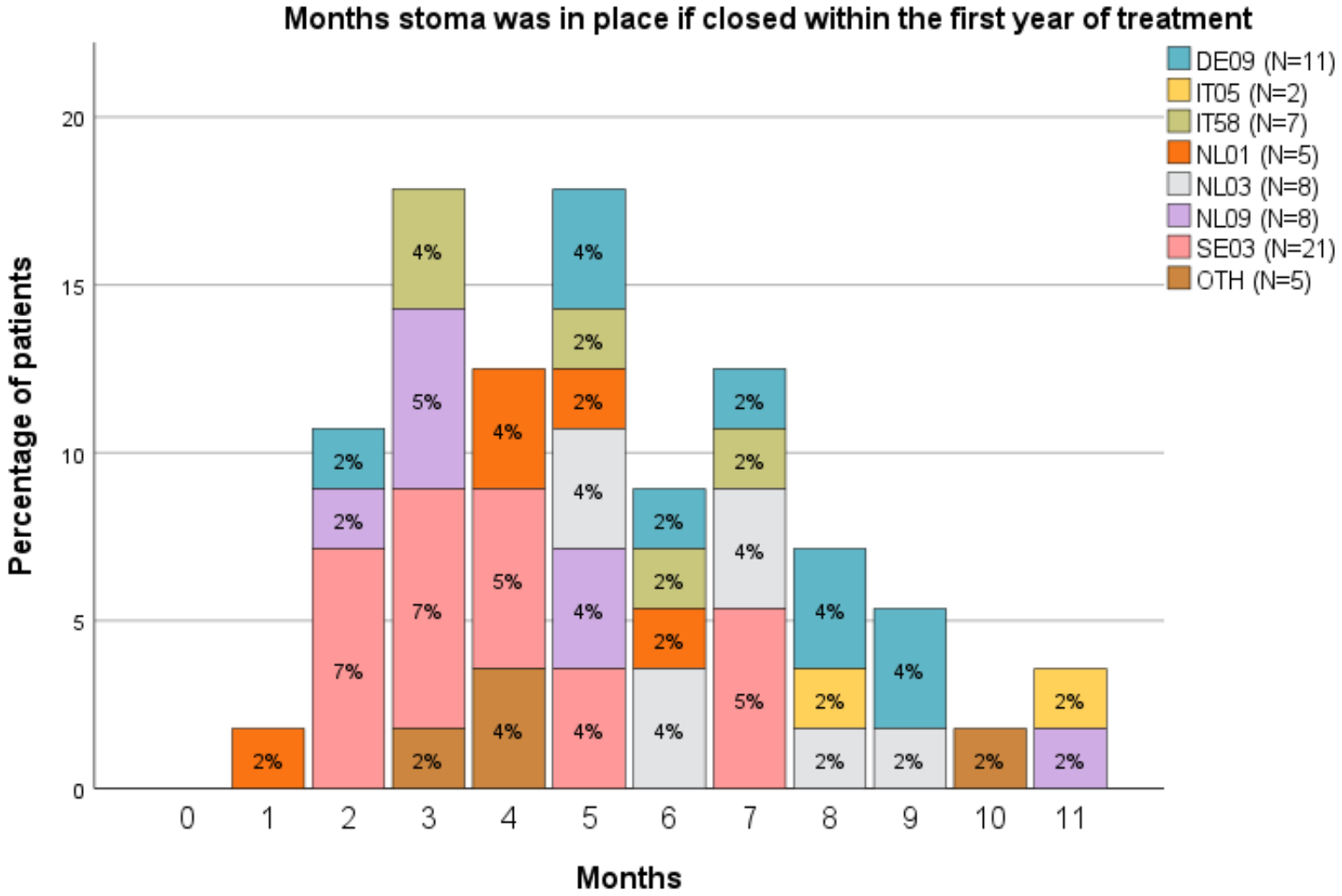
Most of the stomas that were constructed, were already closed during the first year of treatment.





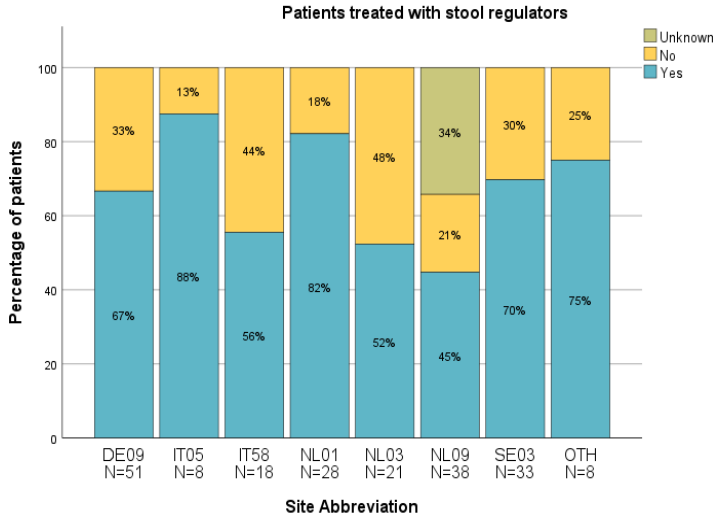
Months stoma was in place if already closed

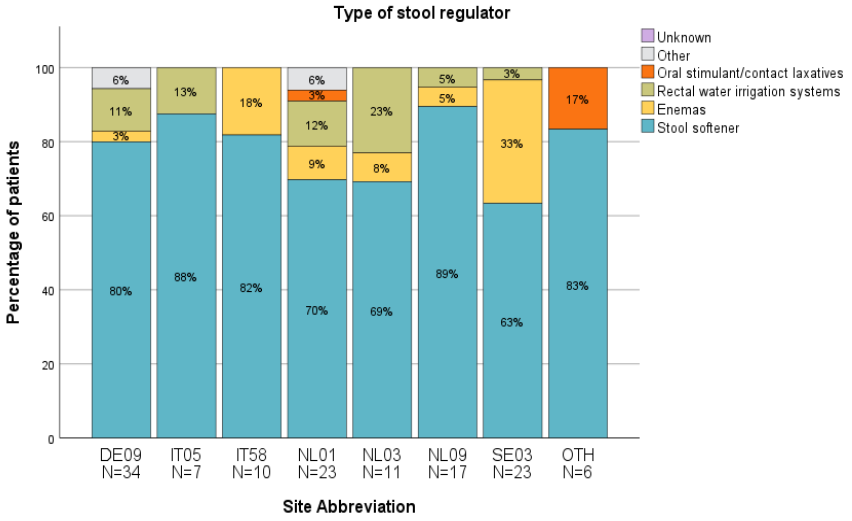
There is a large variety in how long the stoma was in place for patients with a closed stoma.



Treatment with stool regulators

Most patients were treated with stool regulators. If treated with a stool regulator, stool softener was the most common used regulator.





European Reference Networks

European Reference Network for rare or low prevalence complex diseases

Network Urogenital Diseases (ERN eUROGEN)

http://eurogen-ern.eu/

Funded by the European Union

ERN eUROGEN is one of the 24 European Reference Networks (ERNs) approved by the ERN Board of Member States. The ERNs are co-funded by the European Commission. For more information about the ERNs and the EU health strategy, please visit http://ec.europa.eu/health/ern