

ERN eUROGEN registry

Report on Expertise Area 1.7

INTRODUCTION

This report entails the second ERN eUROGEN registry retrospective 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. As retrospective data entry is still ongoing for the majority of HCPs, not all HCPs have reached the minimum of 30 retrospective patients per Expertise Area, yet. Only HCPs which entered more than 5 patients for 1.7 Anorectal malformations were included in these analyses. As patient numbers are not similar, 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 patients 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 taken into account 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=117	Germany DE09 (N=34)	Italy IT58 (N=12)	Netherlands			Sweden SE03 (N=14)
				NL01 (N=10)	NL03 (N=19)	NL09 (N=28)	
Type of malformation (N=113 patients (96.6%))							
Non-syndromic anorectal malformation; N (%)	89 (78.8%)	32 (94.1%)	5 (41.7%)	8 (80.0%)	16 (84.2%)	19 (70.4%)	9 (81.8%)
Syndromic anorectal malformation; N (%)*	24 (21.2%)	2 (5.9%)	7 (58.4%)	2 (20.0%)*	3 (15.8%)	8 (29.6%)	2 (18.2%)
Diagnostics, timing after first visit & abnormalities							
<i>Ultrasound of kidney and bladder</i> (N=117 (100%))							
Performed; N (%)	91 (77.8%)	27 (79.4%)	10 (83.3%)	10 (100%)	18 (94.7%)	14 (50.0%)	12 (85.7%)
Days first visit to USS; Median (range)	8 days (0;112)	1 days (0;112)	0 days (0;1)	2 days (0;29)	2 days (0;25)	2 days (0;70)	4 days (0;29)
Abnormalities found on USS : N (%)	19 (27.8%)	9 (33.3%)	3 (30.0%)	1 (10.0%)	2 (11.1%)	4 (28.6%)	-
<i>Voiding cystourethrogram (VCUG)</i> (N=115 (98.3%))							
Performed; N (%)	35 (30.4%)	7 (20.6%)	4 (33.3%)	-	-	18 (64.3%)	6 (42.9%)
Days first visit to VCUG; Median (range)	34 days (2;175)	17 days (12;67)	32 days (14;175)	-	-	69 days (2;158)	26 days (3;29)
VUR diagnosed; N (%)	9 (25.7%)	3 (42.9%)	-	-	-	4 (22.2%)	2 (33.3%)
<i>X-ray of spine/sacrum</i> (N= 116 (99.1%))							
Performed; N (%)	57 (49.1%)	2 (5.9%)	10 (83.3%)	17 (89.5%)	10 (100%)	12 (42.9%)	6 (46.2%)
Days first visit to X-ray; Median (range)	2 days (0;70)	1 day (0;1)	5 days (0;12)	3 days (0;29)	2 days (0;25)	2 days (0;70)	5 days (1;15)
Abnormalities found on X-ray: N (%)	11 (19.3%)	2 (100%)	2 (20.0%)	3 (17.6%)	3 (30.0%)	2 (16.7%)	5 (83.3%)
<i>Ultrasound of spine/sacrum</i> (N=116 (99.1%))							
Performed; N (%)	82 (70.7%)	22 (64.7%)	9 (75.0%)	10 (100%)	18 (94.7%)	13 (46.7%)	10 (76.9%)
Days first visit to USS&S; Median (range)	3 days (0;217)	7 days (0;217)	9 days (0;142)	2 days (0;29)	3 days (0;25)	2 days (0;70)	4 days (2;29)
Abnormalities found on USS&S; N (%)	16 (25.0%)	8 (36.4%)	1 (11.1%)	3 (30.0%)	1 (5.6%)	1 (7.7%)	2 (20.0%)
<i>MRI of spine/sacrum</i> (N=117 (100%))							
Performed; N (%)	16 (13.7%)	5 (14.7%)	3 (25.0%)	2 (20.0%)	3 (15.8%)	1 (3.6%)	2 (14.3%)
Days first visit to MRI; Median (range)	66 days (1;249)	74 days (3;249)	30 days (30;212)	28 days (13;43)	67 days (65;71)	1 day (-)	151 days (108;194)
Abnormalities found on MRI; N (%)	12 (75.0%)	5 (100%)	2 (66.7%)	2 (100%)	1 (33.3%)	-	2 (100%)
<i>Echocardiogram</i> (N=117 (100%))							
Performed; N (%)	74 (63.2%)	11 (32.4%)	12 (100%)	10 (100%)	18 (94.7%)	11 (39.3%)	13 (92.9%)
Days first visit to ECG; Median (range)	2 days (0;102)	6 days (0;31)	1 day (0;18)	3 days (0;65)	6 days (0;102)	2 days (0;70)	0 days (0;57)
Abnormalities found on ECG; N (%)	30 (40.5%)	5 (45.5%)	8 (66.7%)	7 (70.0%)	-	8 (72.7%)	2 (15.4%)

	Total N=117	Germany DE09 (N=34)	Italy IT58 (N=12)	Netherlands			Sweden SE03 (N=14)
				NL01 (N=10)	NL03 (N=19)	NL09 (N=28)	
Surgery and treatment							
<i>Reconstructive surgery</i> (N=117 (100%))							
Performed within 1 year after 1 st visit; N (%)	101 (86.3%)	26 (76.5%)	9 (75.0%)	8 (80.0%)	19 (100%)	25 (89.3%)	14 (100%)
Performed in own HCP; N (%)	92 (91.1%)	22 (84.6%)	9 (100%)	8 (100%)	15 (78.9%)	24 (96.0%)	14 (100%)
Age at surgery; Median (range)	100 days (1;6042)	119 days (1;6042)	44 days (3;132)	128 days (101;223)	39 days (1;216)	104 days (1;253)	59 days (7;920)
<i>Stoma/enterostomy</i> (N=117 (100%))							
Patients with stoma/enterostomy; N (%)	36 (30.8%)	7 (20.6%)	5 (41.7%)	-	8 (42.1%)	7 (25.0%)	9 (64.3%)
Patient with closed stoma; N (%)	31 (86.1%)	7 (100%)	4 (80.0%)	-	8 (100%)	4 (57.1%)	8 (88.9%)
Days stoma was in place; Median (range)	173 days (69;303)	218 days (69;303)	135 days(111;224)	-	213 days (167;299)	139 days (69;160)	141 days (80;225)
<i>Anal dilations</i> (N=117 (100%))							
Treated with anal dilations; N (%)	74 (63.2%)	20 (58.8%)	12 (100%)	10 (100%)	19 (100%)	13 (46.4%)	-
<i>Stool regulators</i> (N=116 (99.1%))							
Treated with stool regulators; N (%)	69 (59.5%)	22 (64.7%)	8 (66.7%)	10 (100%)	10 (52.6%)	10 (35.7%)	9 (69.2%)

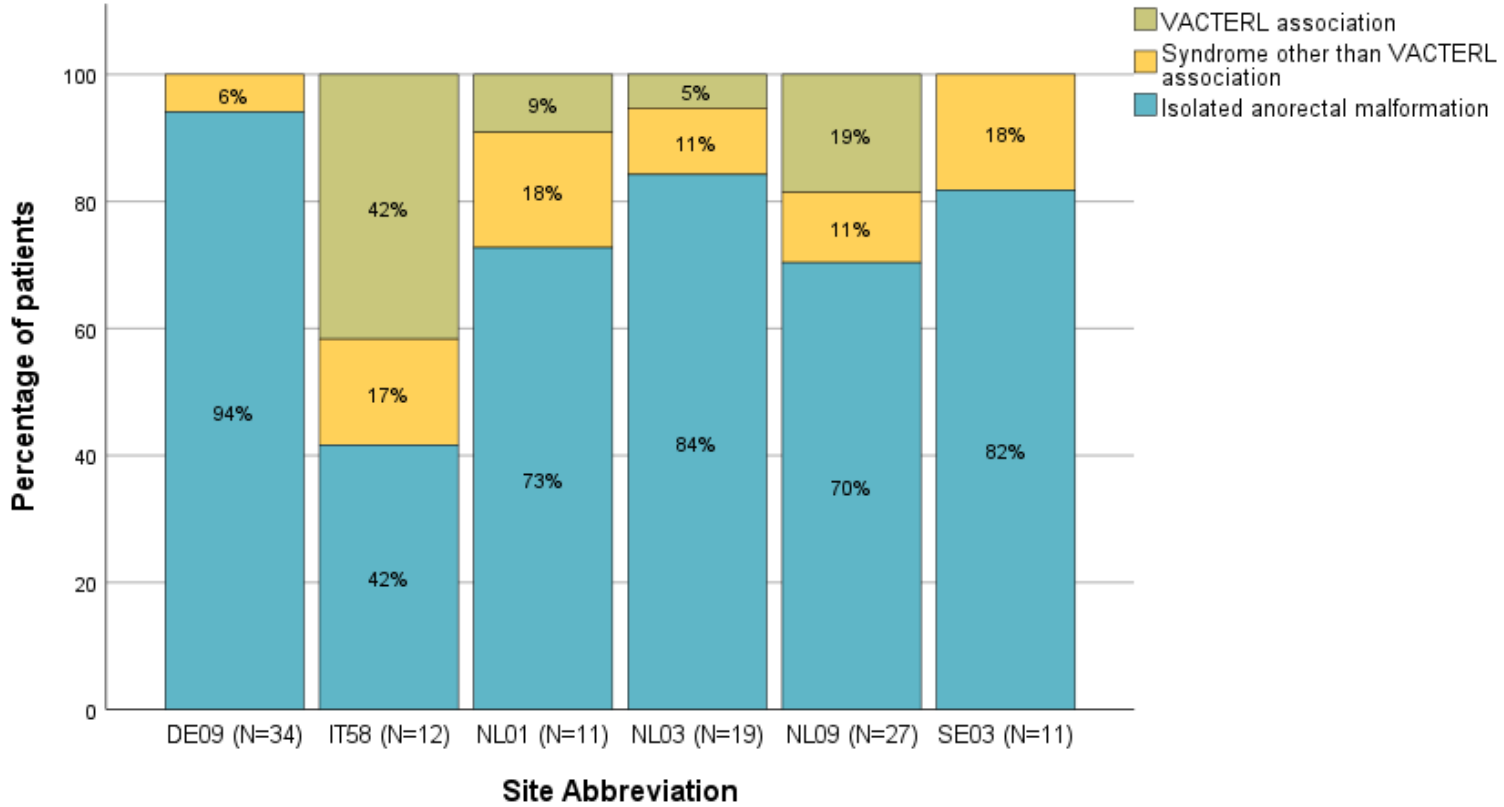
* in the corresponding figure (Type of anorectal malformation), one patients is represented twice, as this patient has been diagnosed with both a VACTERL association and another syndrome.

Type of malformation

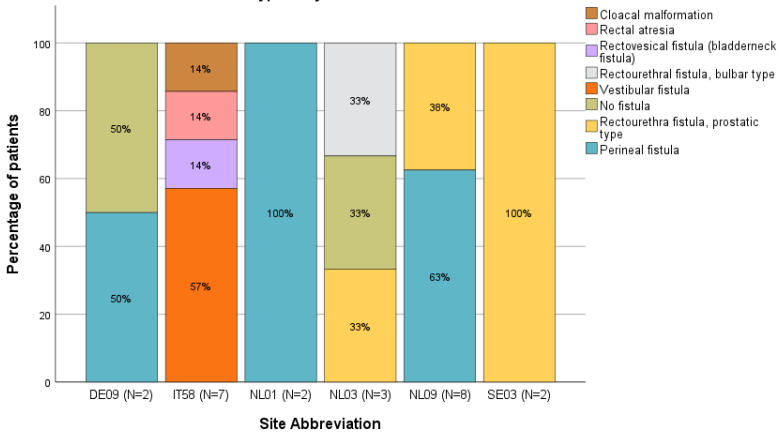
Type of anorectal malformation

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

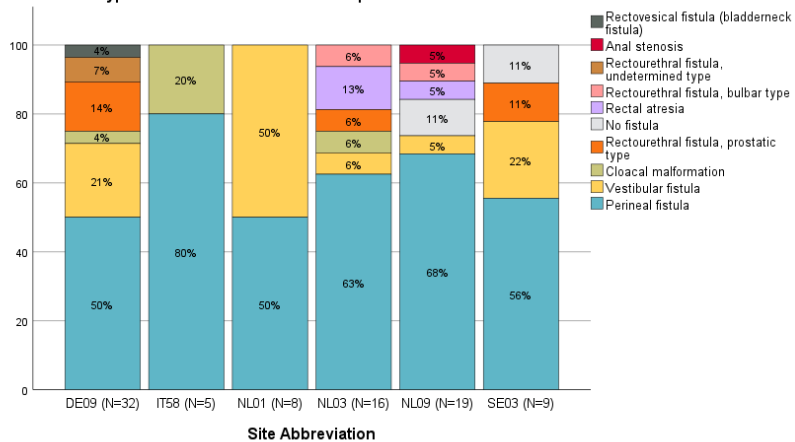
Type of anorectal malformation



Subtype of syndromic anorectal malformation



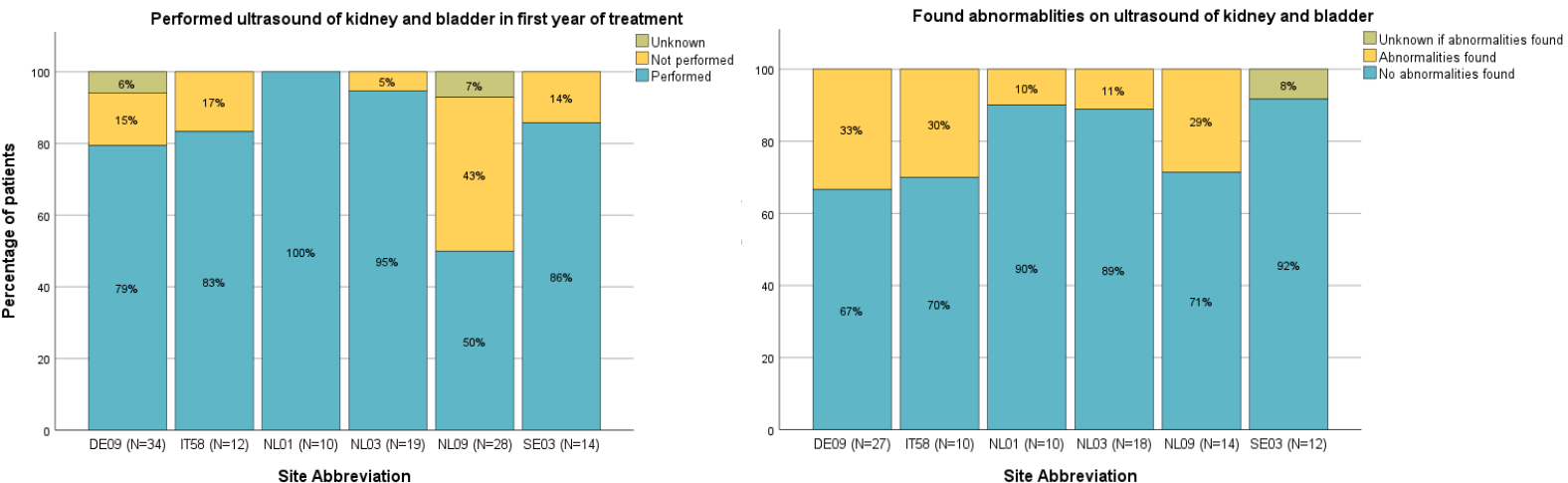
Subtype of anorectal malformation for patients with isolated anorectal malformation



Diagnostics, timing after first visit & abnormalities

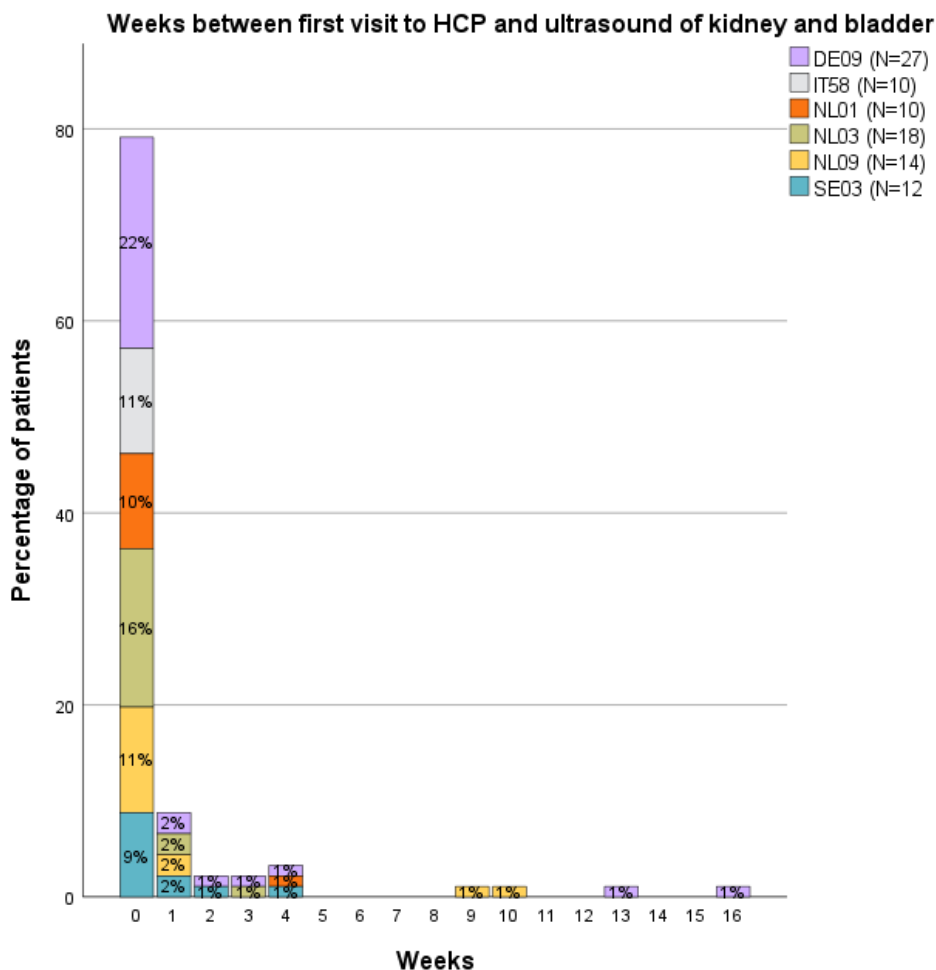
Ultrasounds of kidney and bladder with abnormalities found in the first year of treatment

An ultrasound of the kidney and bladder was performed in the majority of patients. When an ultrasound was performed, no abnormalities were found in the majority of patients.



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

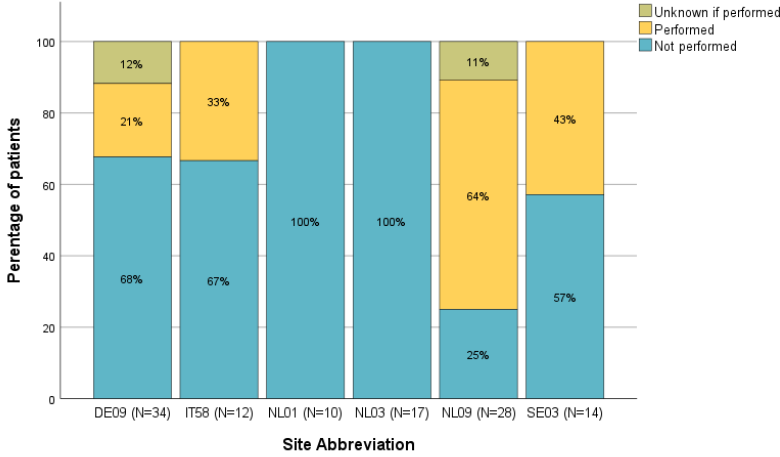
The majority of ultrasounds were performed in the same week as the first visit to the HCP, with ultrasounds being performed until 16 weeks after their first visit to the HCP.



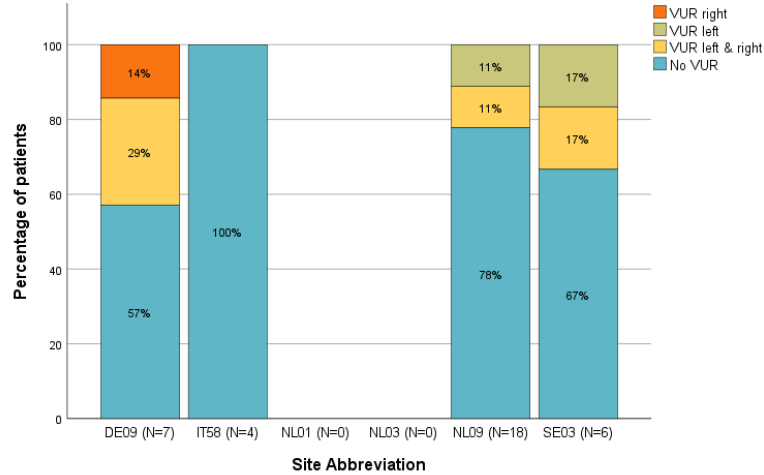
Voiding cystourethrogram (VCUG) and vesicoureteral reflux (VUR) in the first year of treatment

VCUG was not performed in the majority of patients. If a VCUG was performed, no VUR was diagnosed in most cases. If VUR was diagnosed, it occurred mostly on both sides.

Performed voiding cystourethrogram (VCUG) in first year of treatment



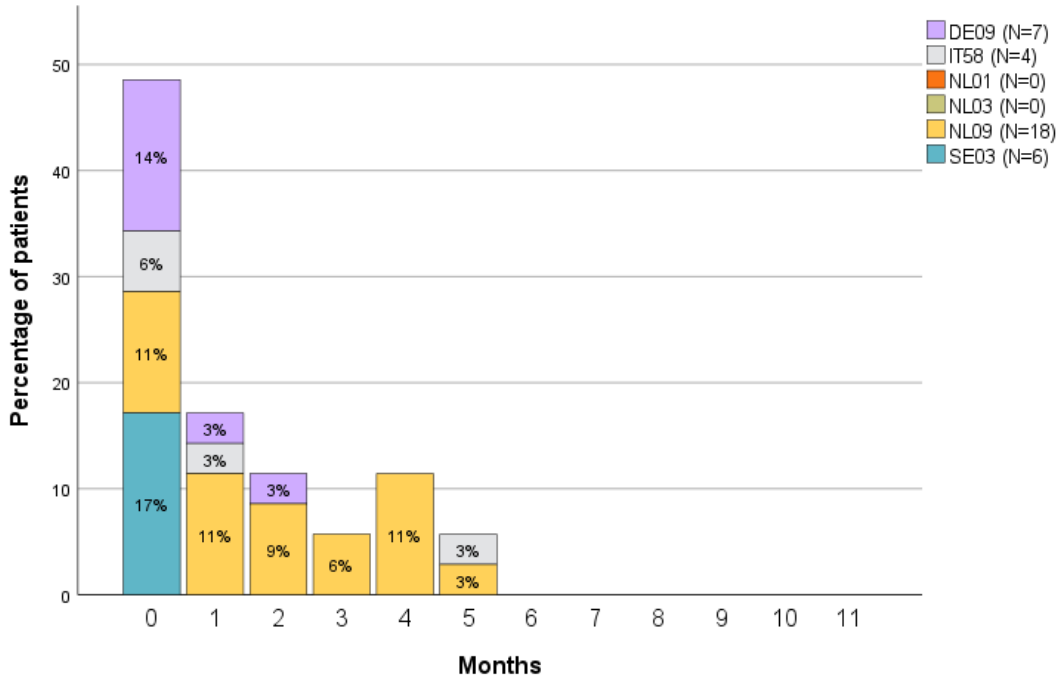
VCUG with diagnosed vesico-ureteral reflux (VUR)



Months between first visit to HCP and VCUG

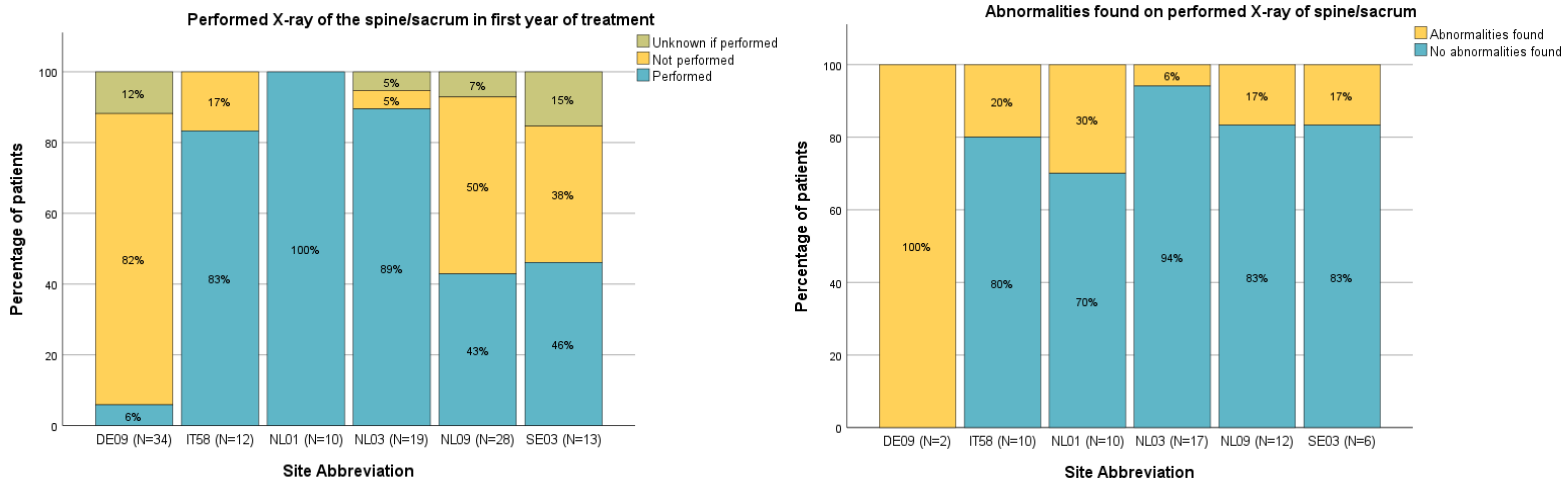
All VCUGs were performed within the first half year after the first visit to the HCP.

Months between first visit to HCP and performed VCUG



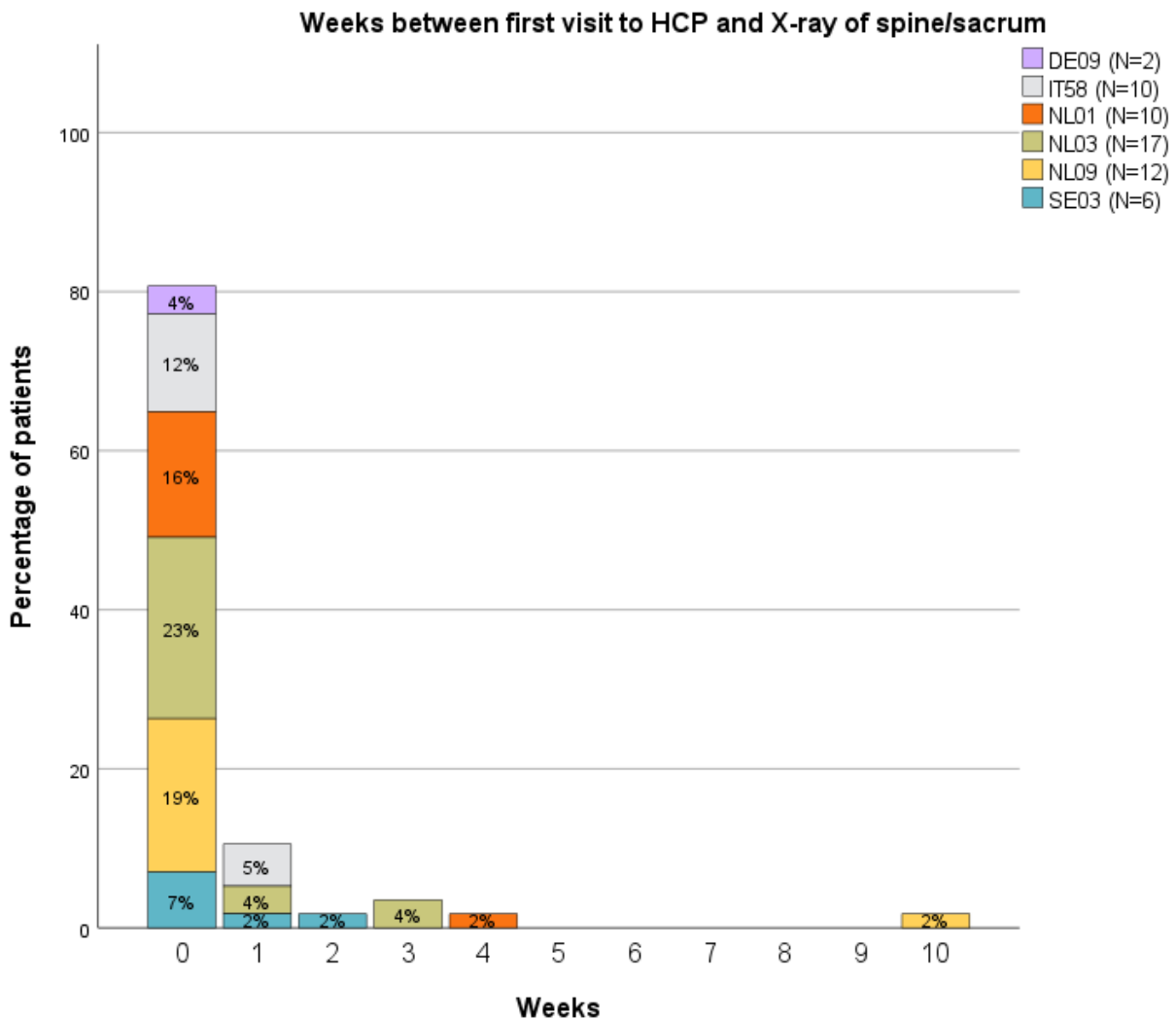
X-ray of spine/sacrum and abnormalities found in the first year of treatment

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



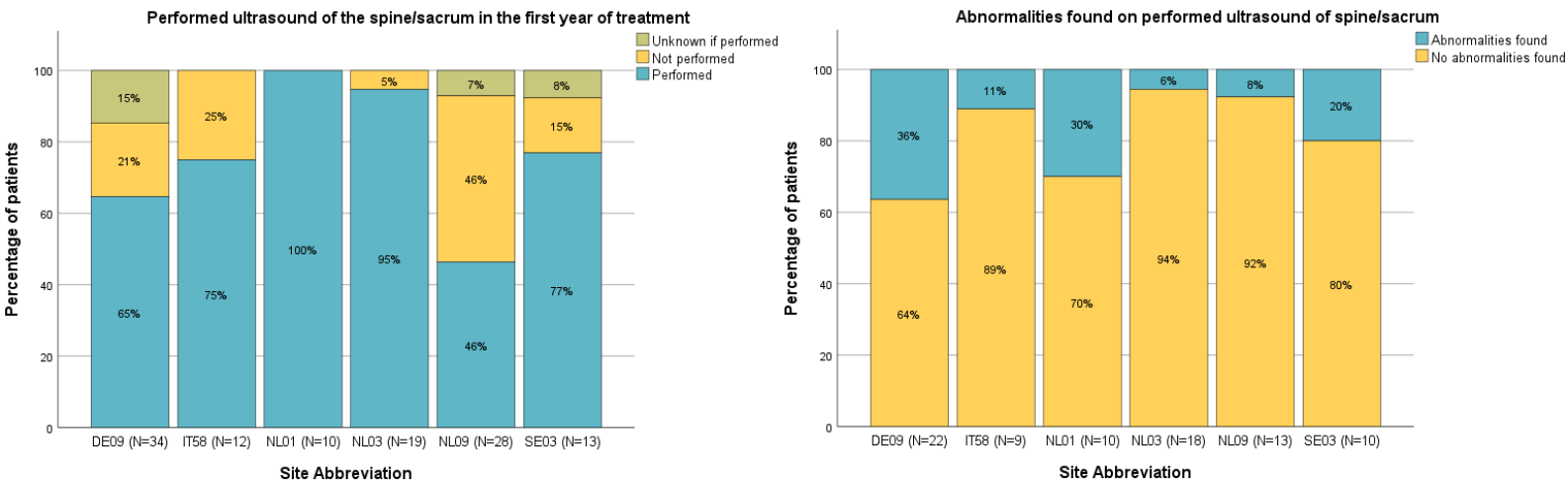
Months between first visit to HCP and X-ray of spine/sacrum

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



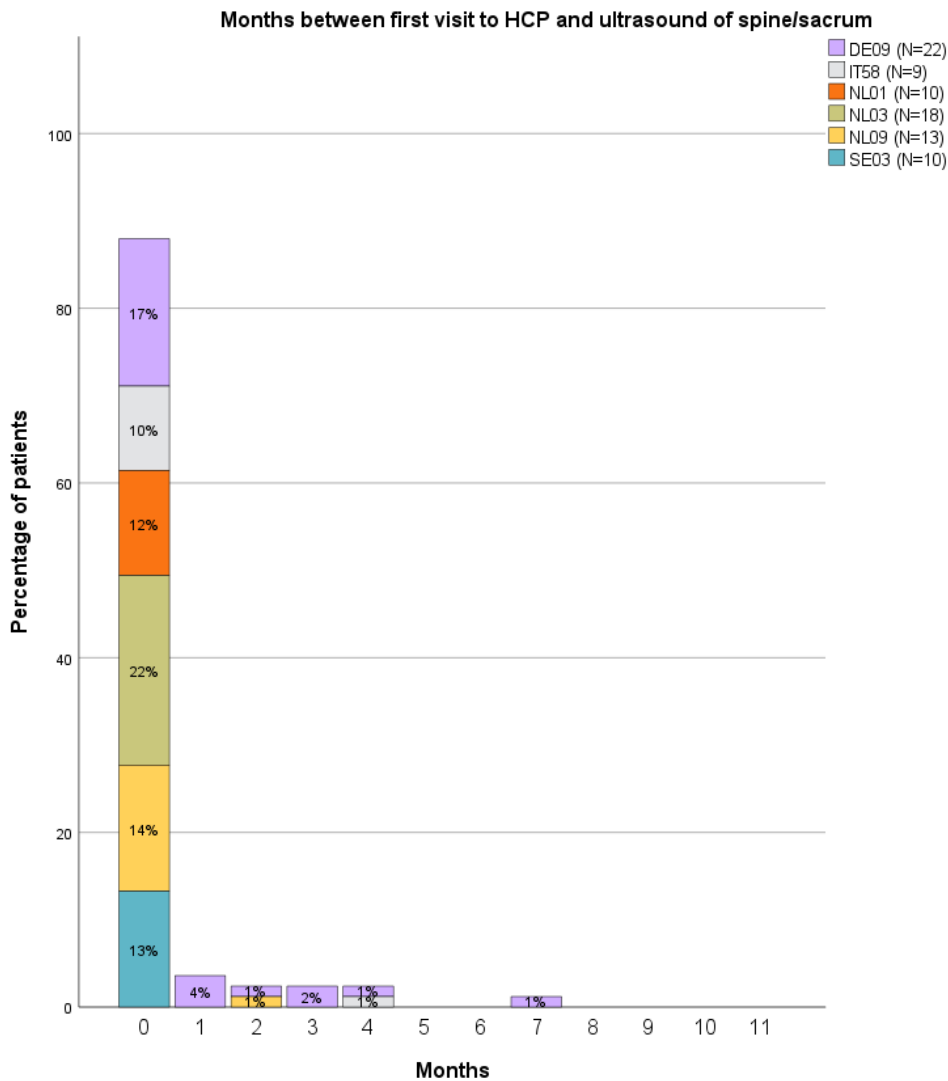
Ultrasound of spine/sacrum and abnormalities found in the first year of treatment

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



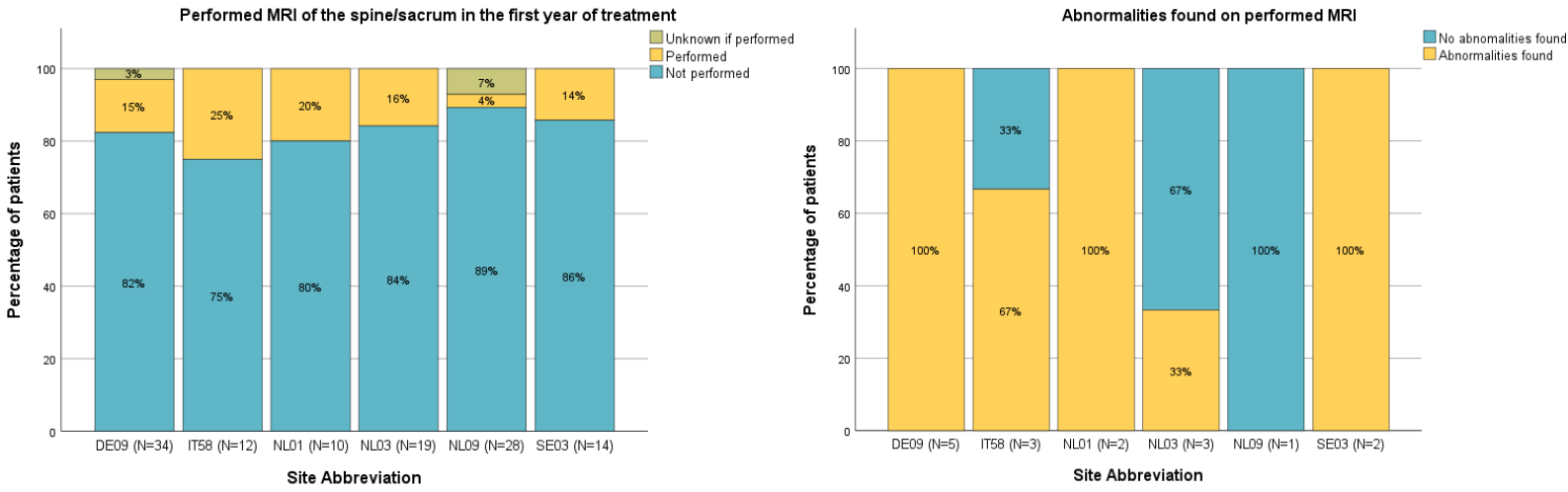
Months between first visit to HCP and ultrasound of spine/sacrum

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



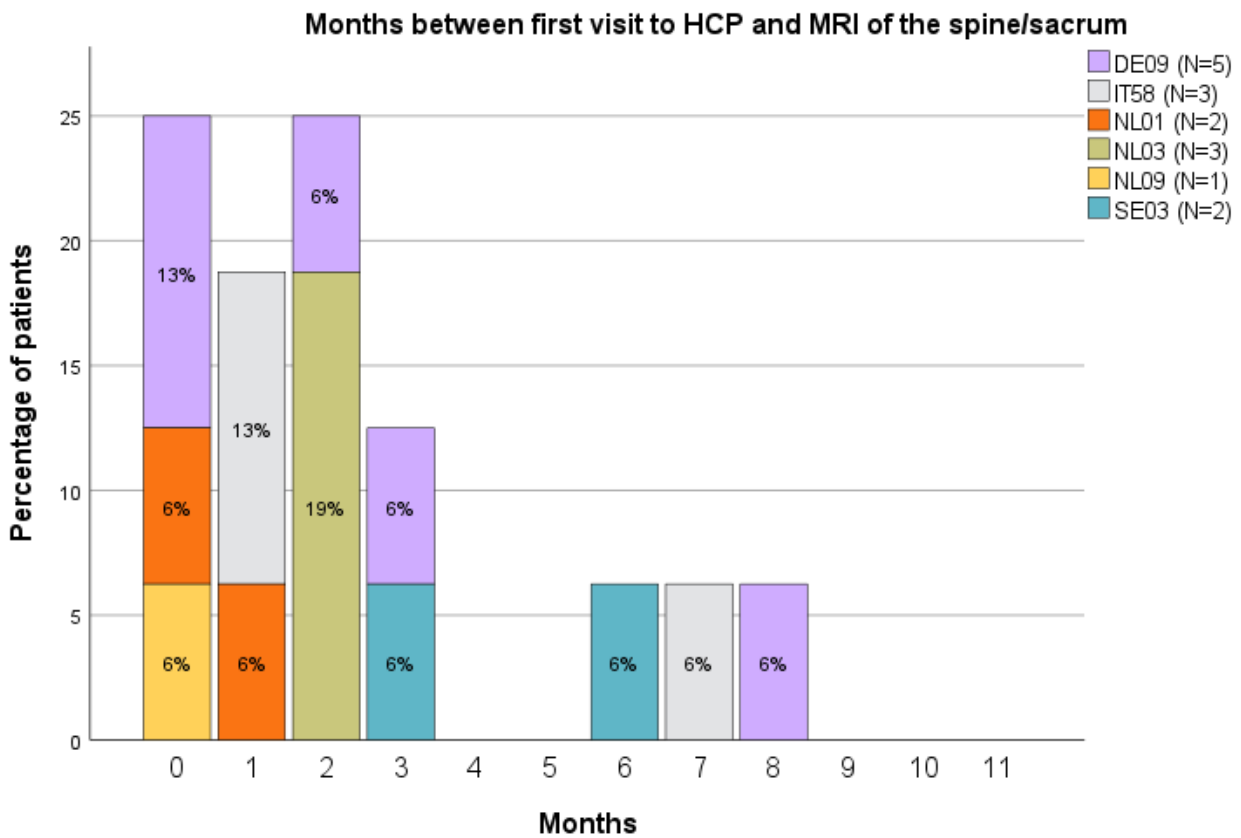
MRI of spine/sacrum and abnormalities found in the first year of treatment

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



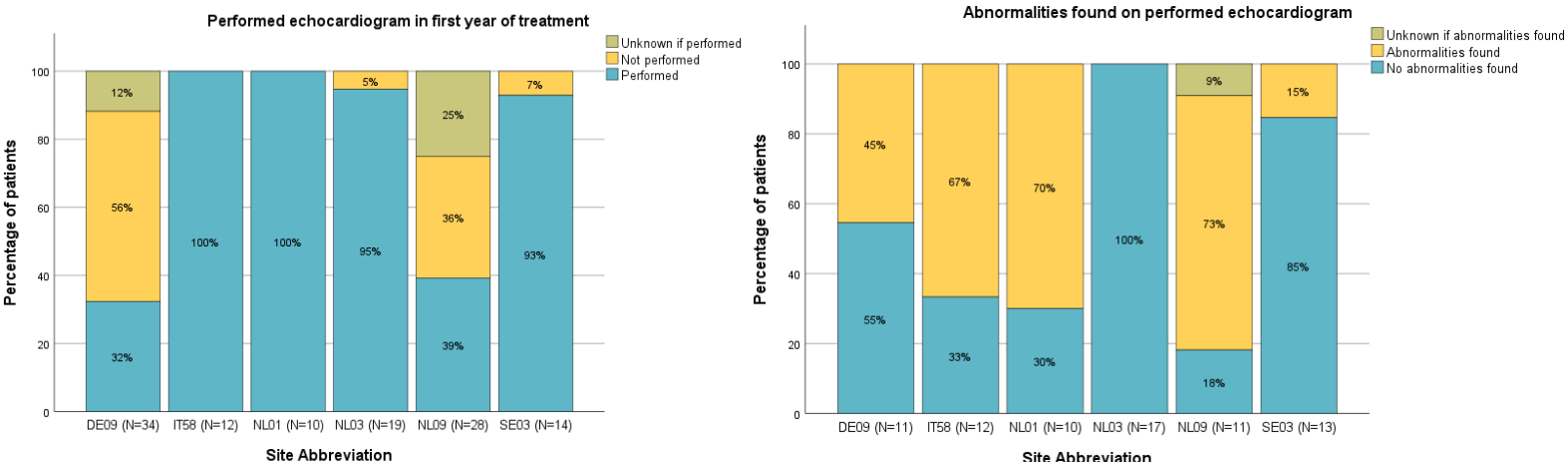
Months between first visit to HCP and MRI of spine/sacrum

Only a few MRIs were performed, and most of these were performed within the first half year after the first visit to the HCP.



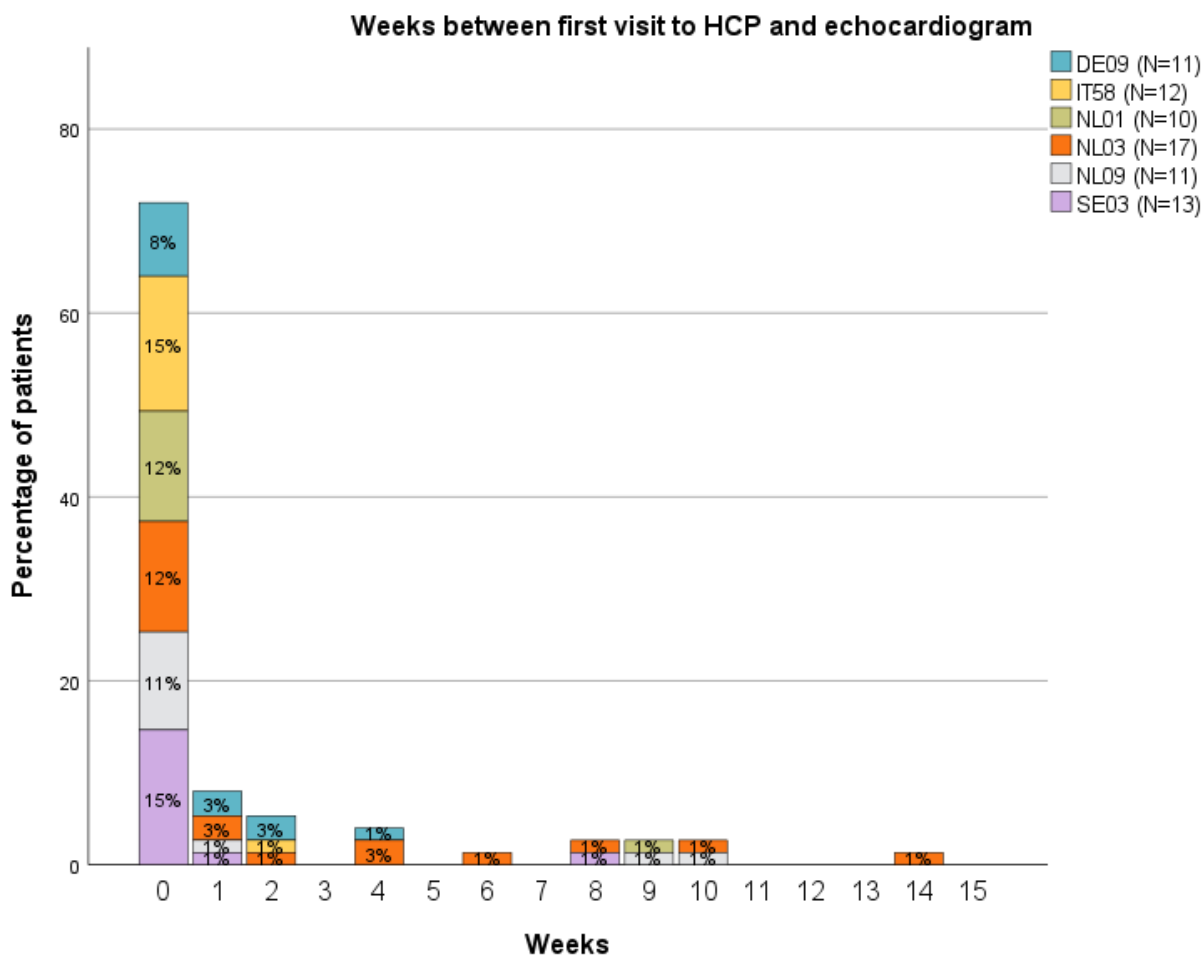
Echocardiogram (ECG) of spine/sacrum and abnormalities found in the first year of treatment

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



Weeks between first visit to HCP and ECG

The majority of ECGs was performed in the same week as the first visit to the HCP.

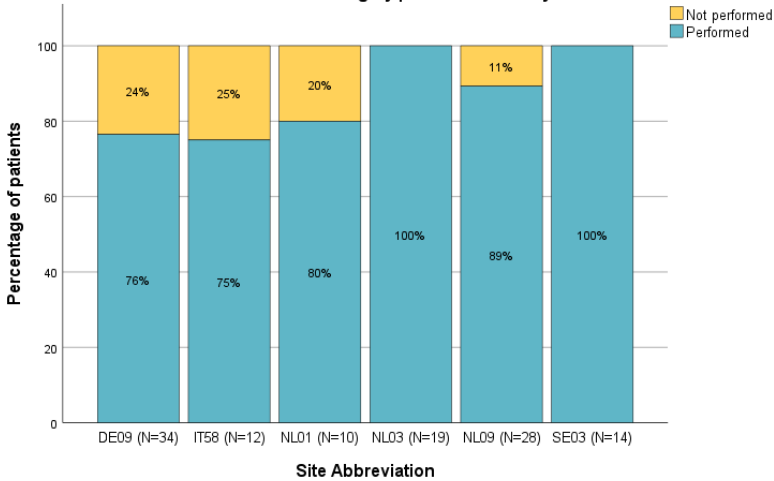


Surgery and treatment

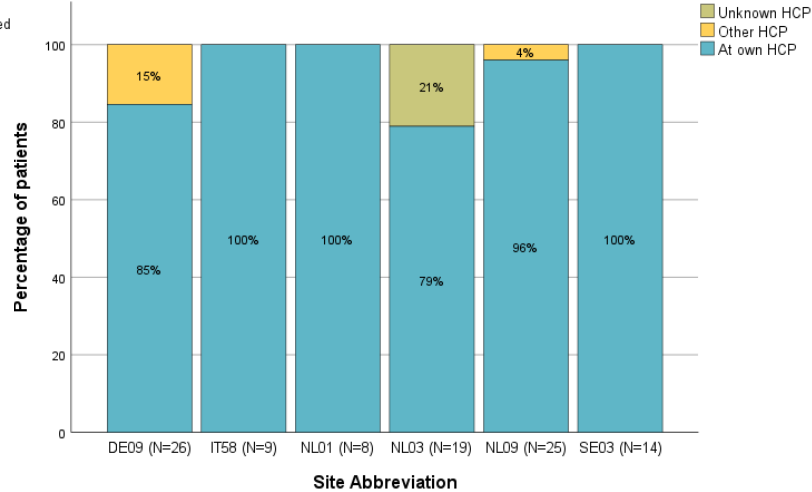
Reconstructive surgery in the first year of treatment

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.

Reconstructive surgery performed in first year of treatment



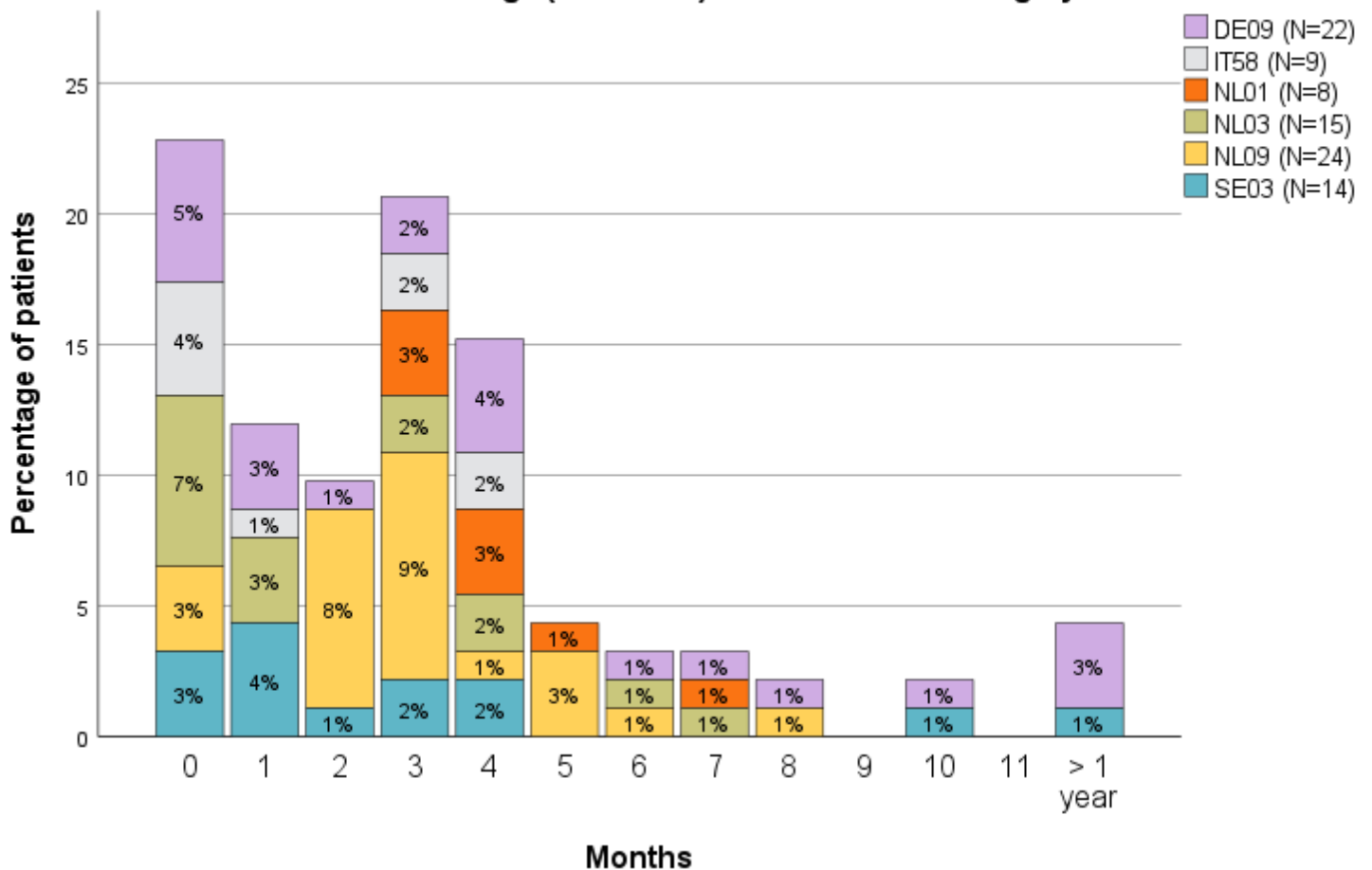
Location performed reconstructive surgery



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

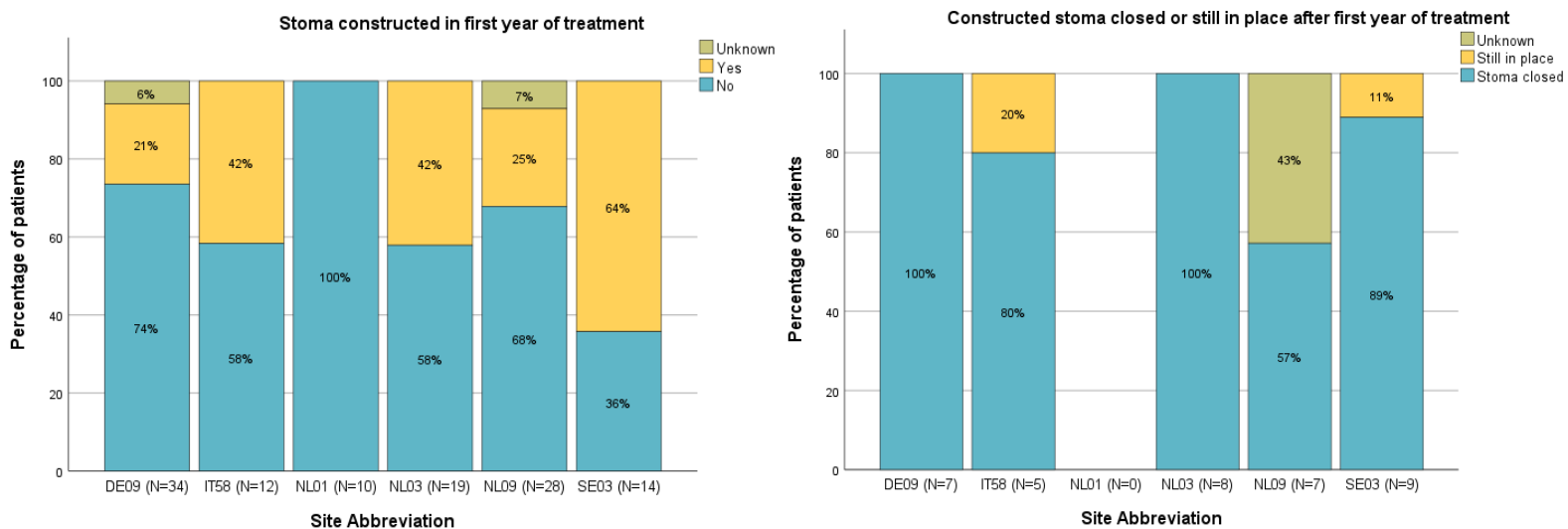
The majority of patients had the reconstructive surgery within the first half year of their life.

Age (in months) at reconstructive surgery



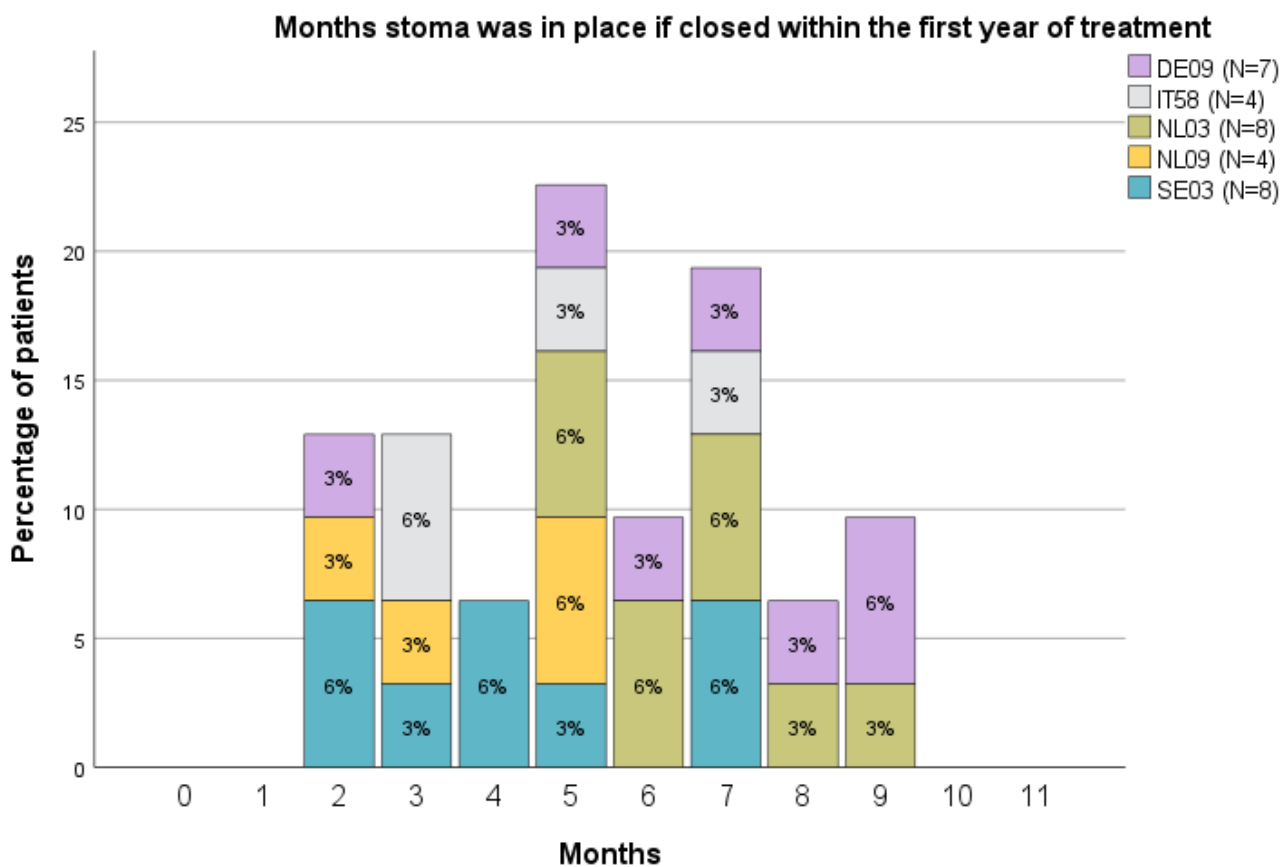
Stoma construction in the first year of treatment

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



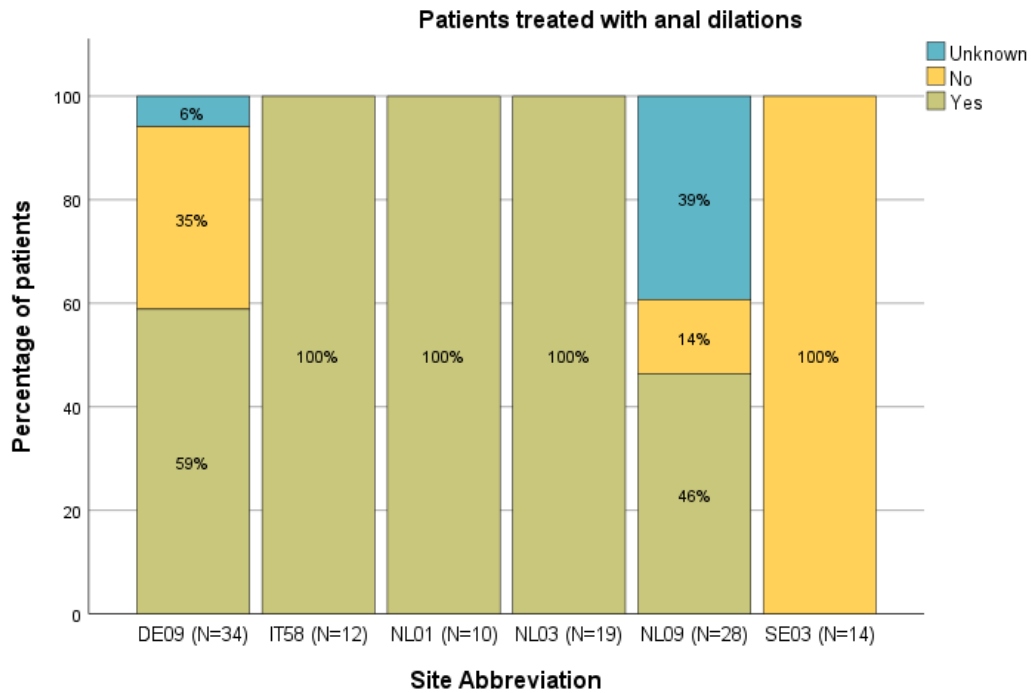
Months stoma was in place if already closed

There is a large variety in how long the stoma was in place for patients with a closed stoma. The time of having a stoma varies from 2 to 9 months.



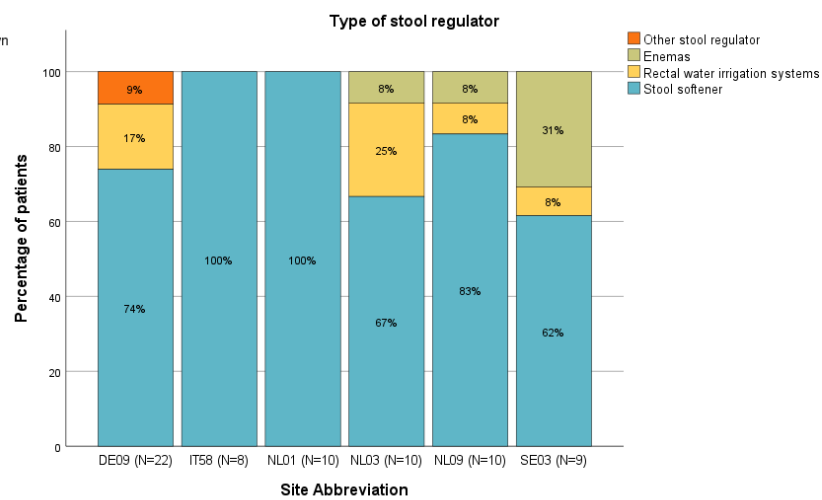
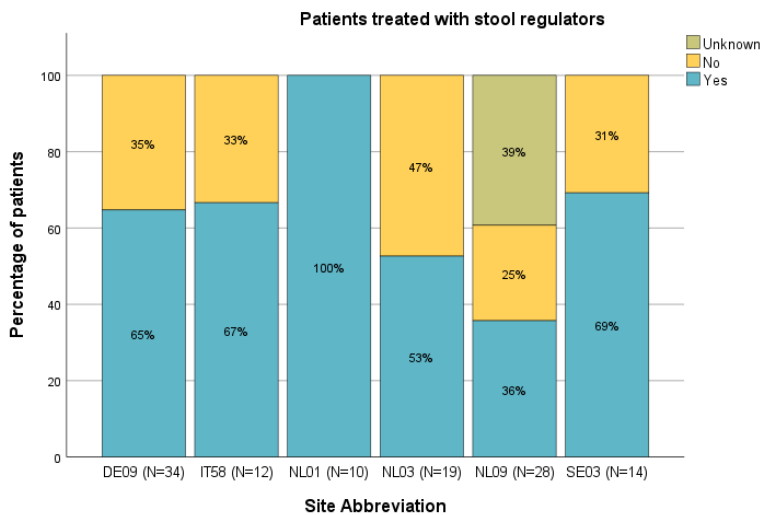
Anal dilations

The majority of patients were treated with anal dilations.



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.





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