

ERN eUROGEN registry

Report on Expertise Area 1.7

INTRODUCTION

This report entails the first 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.

The Clinical Practice Snapshots should only contain data about the first year of treatment. 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 'No', 'Not performed', or 'Unknown'. An example: A patient had the first contact with your HCP at 23-01-2021 (start treatment), and the reconstructive surgery took place on 08-02-2022 (more than a year after the start of treatment). This surgery should not be entered in the Clinical Practice Snapshot of the ERN eUROGEN registry. If this information was there, we interpreted it as 'No reconstructive surgery'. If it was indicated that the surgery took place but the date is unknown, we interpreted the variable as 'Unknown', because we don't know if this surgery took place within a year from the start of treatment.

Furthermore, 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. Therefore, the results cannot be equally compared between HCPs, but the analyses give an indication of trends.

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.

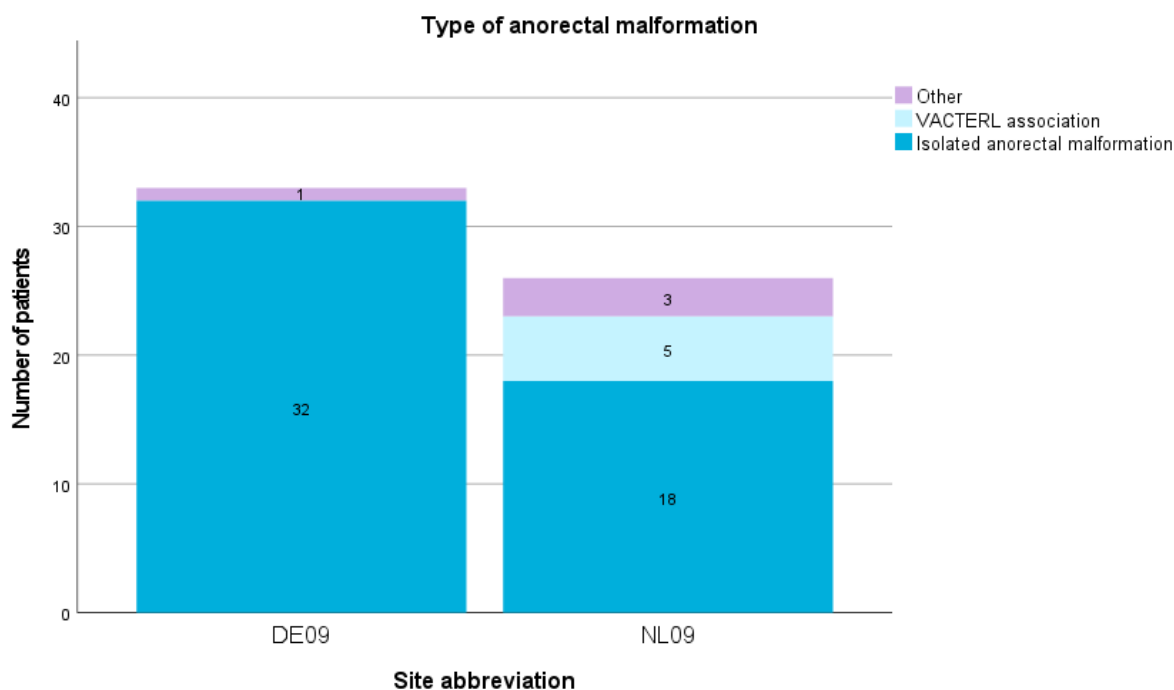
| | Total N=59 | Netherlands NL09 (N=26) | Germany DE09 (N=33) |
|--|----------------------|----------------------------|------------------------|
| Type of malformation | | | |
| Non-syndromic anorectal malformation; N (%) | 50 (84.7%) | 18 (69.2%) | 32 (97.0%) |
| Syndromic anorectal malformation; N (%) | 9 (15.3%) | 8 (30.8%) | 1 (3.0%) |
| Diagnostics, timing after first visit & abnormalities | | | |
| Ultrasound of kidney & bladder performed; N (%) | 54 (91.5%) | 24 (95.3%) | 30 (90.9%) |
| Days first visit to USK&B; Median (range)* | 1 day (0;112) | 2 days (0;70) | 1 days (0;112) |
| Abnormalities found on USK&B: N (%) | 15 (27.8%) | 5 (20.8%) | 10 (33.3%) |
| Voiding cystourethrogram performed; N (%) | 26 (44.1%) | 18 (69.2%) | 8 (24.2%) |
| Days first visit to VCUG; Median (range)* | 53 days (2;158) | 68.5 days (2;158) | 17 days (12;67) |
| VUR diagnosed; N (%) | 8 (32.0%) | 4 (22.7%) | 4 (50.0%) |
| X-ray of spine / sacrum performed; N (%) | 25 (42.4%) | 22 (84.6%) | 3 (9.1%) |
| Days first visit to X-ray; Median (range)* | 2 days (0;70) | 2 days (0;70) | 0.5 days (0.5;1) |
| Abnormalities found on X-ray: N (%) | 8 (32.0%) | 5 (22.7%) | 3 (100%) |
| Ultrasound of spine / sacrum performed; N (%) | 44 (74.5%) | 21 (79.8%) | 23 (69.7%) |
| Days first visit to USS&S; Median (range)* | 2.5 days (0;217) | 2 days (0;70) | 6.5 days (0;217) |
| Abnormalities found on USS&S; N (%) | 11 (25.0%) | 2 (9.5%) | 9 (39.1%) |
| MRI of spine / sacrum performed; N (%) | 9 (15.3%) | 2 (7.7%) | 7 (21.2%) |
| Days first visit to MRI; Median (range)* | 40.5 days (1;249) | 1 day (-) | 74 days (3;249) |
| Abnormalities found on MRI; N (%) | 7 (77.8%) | - | 7 (100%) |
| Performed echocardiogram; N (%) | 28 (47.5%) | 16 (61.5%) | 12 (36.4%) |
| Days first visit to ECG; Median (range)* | 3 days (0;70) | 2.5 days (0;70) | 6 days (0;31) |
| Abnormalities found on ECG; N (%) | 20 (71.4%) | 14 (87.5%) | 6 (50.0%) |
| Surgery and treatment | | | |
| Reconstructive surgery performed; N (%) | 49 (83.1%) | 24 (92.3%) | 25 (75.7%) |
| Performed in own HCP; N (%) | 42 (89.4%) | 23 (95.8%) | 19 (79.4%) |
| Days from diagn. to surgery; Median (range)* | 75 days (-554;44530) | 78 days (-554;44530) | 16.5 days (0;114) |
| Patients with stoma/enterostomy; N (%) | 19 (32.2%) | 6 (23.0%) | 13 (39.4%) |
| Patient who still have the stoma; N (%) | 1 (5.3%) | - | 1 (7.7%) |
| Days stoma was in place; Median (range)* | 183 days (69;303) | 118 days (69;118) | 218 days (69;303) |
| Patients undergo anal dilatations: N (%) | 31 (52.5%) | 12 (46.2%) | 19 (57.6%) |
| Treated with stool regulators; N (%) | 29 (49.2%) | 8 (30.8%) | 21 (63.6%) |

*Only procedures performed in the ERN HCP were used for the calculation.

Type of malformation

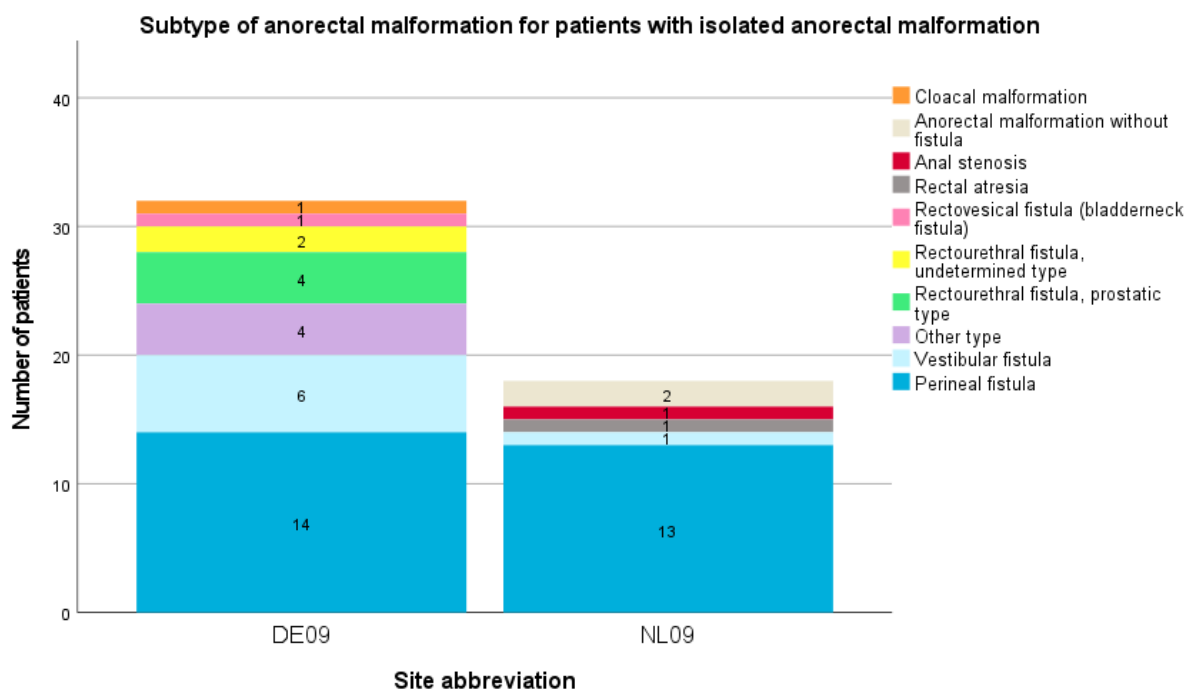
Type of anorectal malformation

The majority of patients was diagnosed with an isolated form of anorectal malformation.



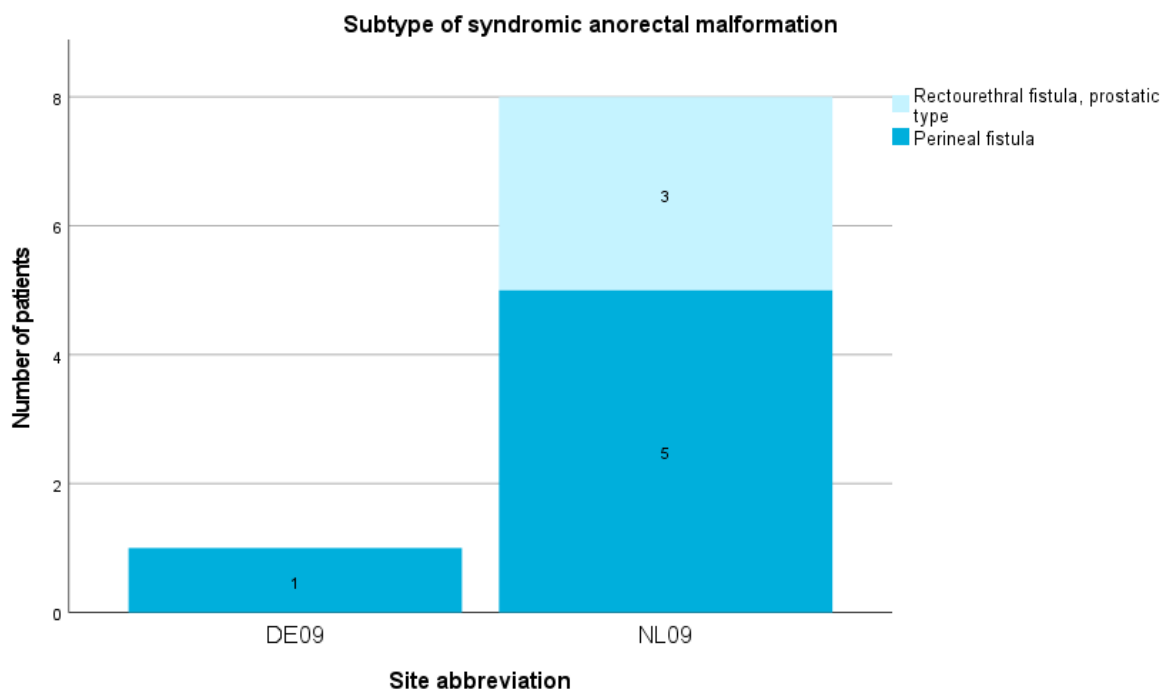
Subtype of anorectal malformation for patients with isolated anorectal malformation

Overall, there was large variability in type of isolated anorectal malformation, but most patients were diagnosed with perineal fistula.



Subtype of syndromic anorectal malformation

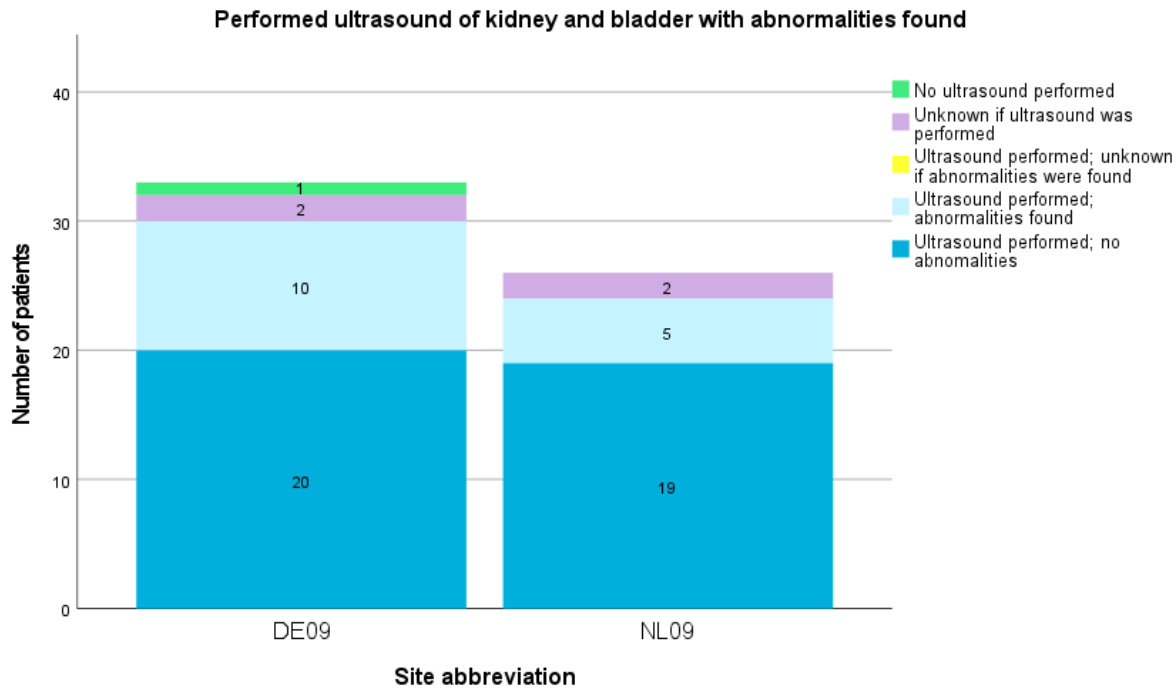
Also for patients with a syndromic type of anorectal malformation, perineal fistula was the most common type.



Diagnostics, timing after first visit & abnormalities

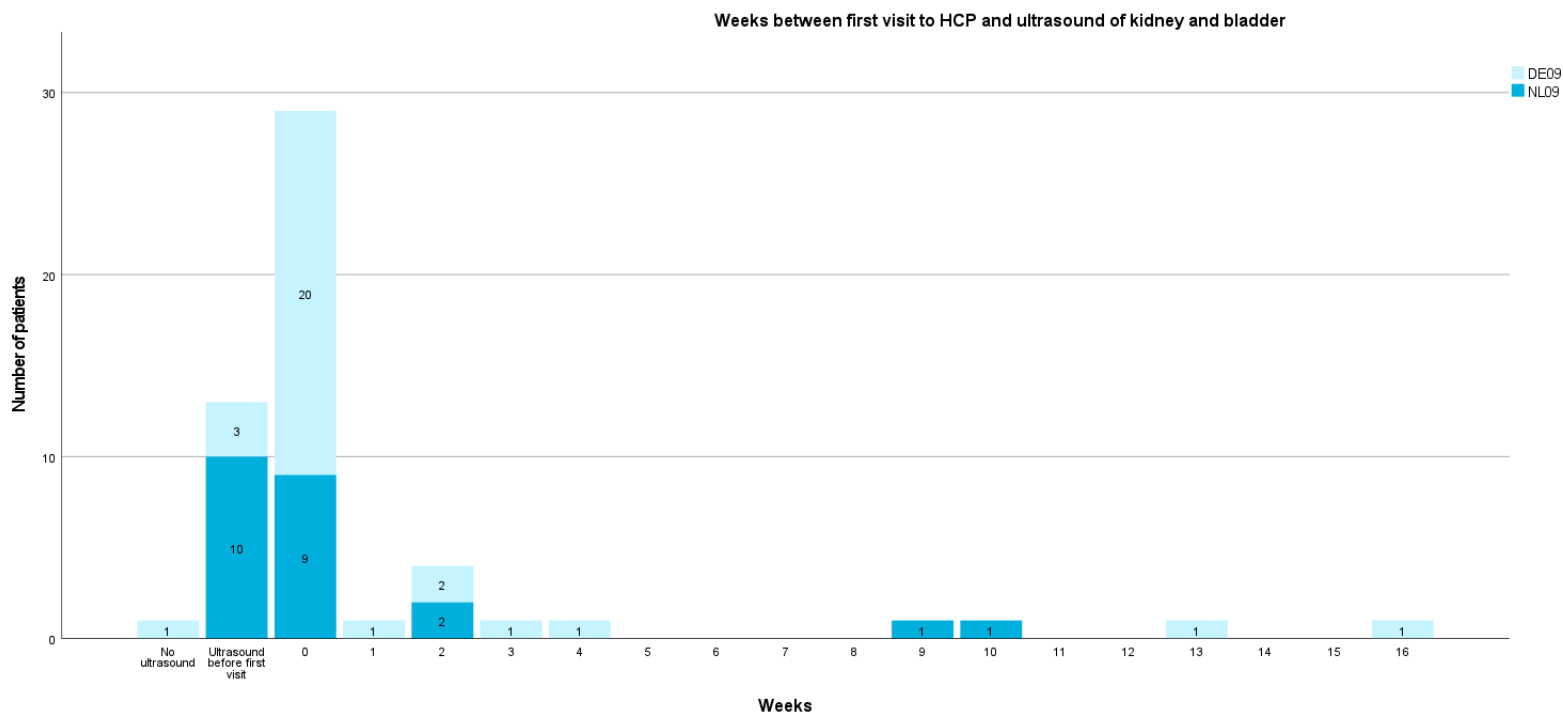
Ultrasounds of kidney and bladder with abnormalities found

An ultrasound was not performed in the majority of patients. However, when an ultrasound was performed, abnormalities were found in the majority of patients.



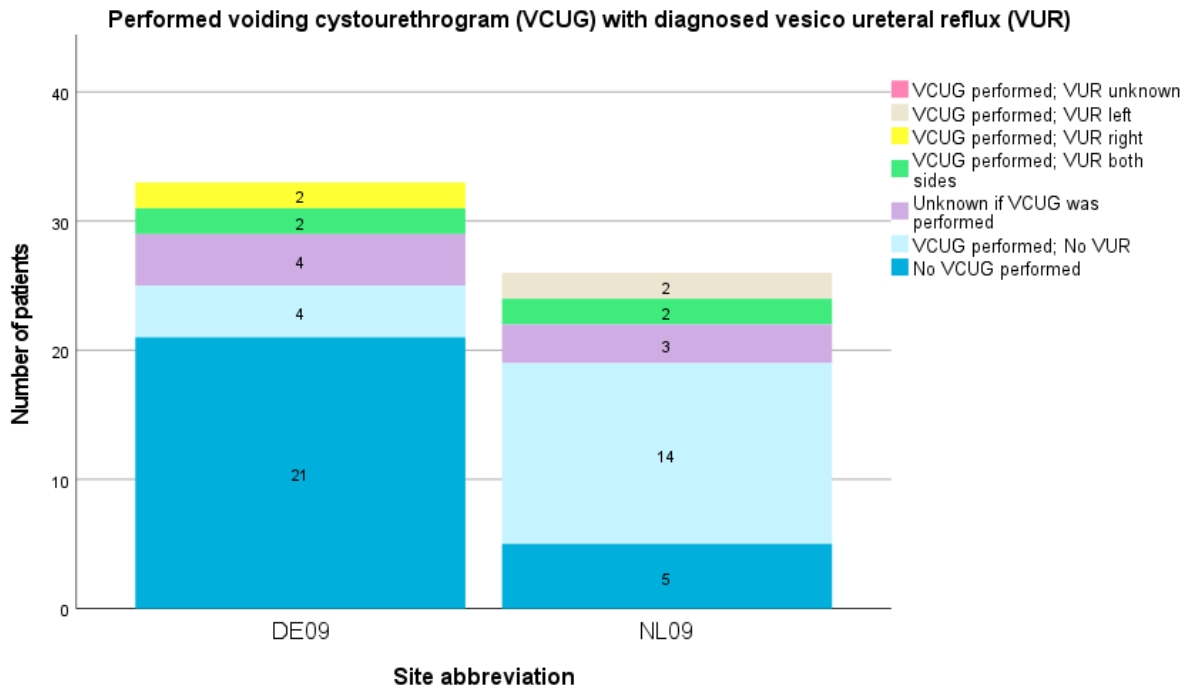
Weeks between first visit to HCP and ultrasound of kidney and bladder

The majority of ultrasounds were performed in the same week as the first visit to the HCP, if the ultrasound was not performed earlier already.



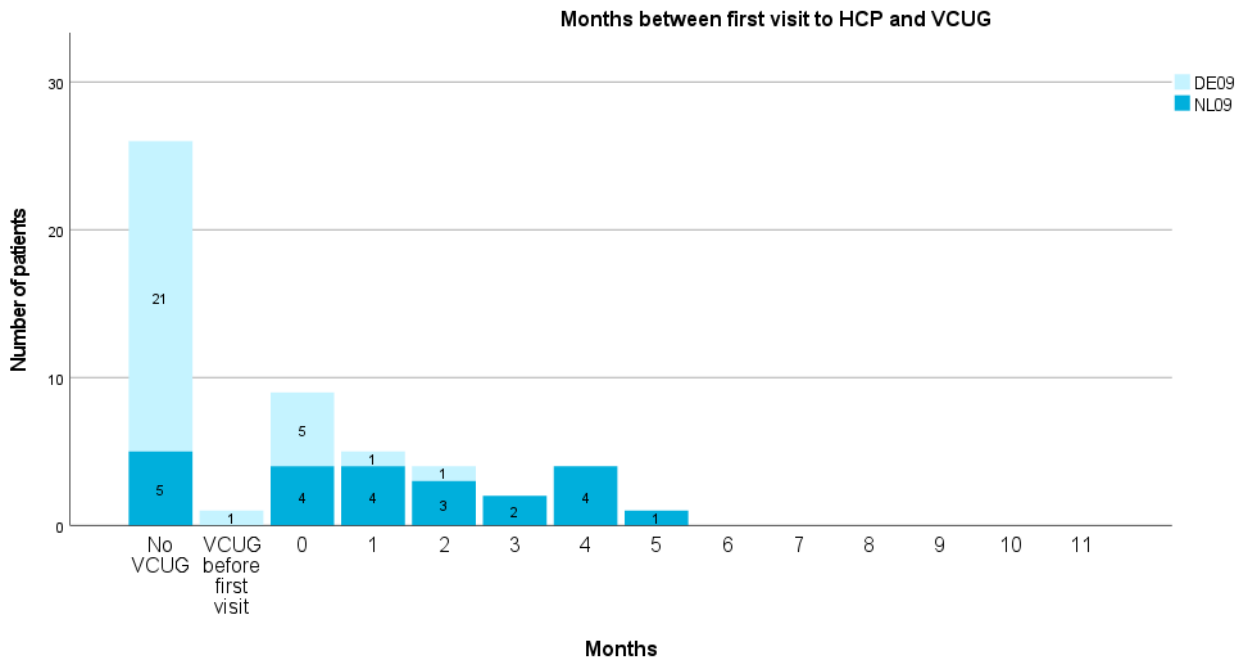
Voiding cystourethrogram (VCUG) and vesicoureteral reflux (VUR)

VCUG was not performed in the majority of DE09 patients. NL09 performed a VCUG for most patients, but VUR was diagnosed only in a minority of the patients.



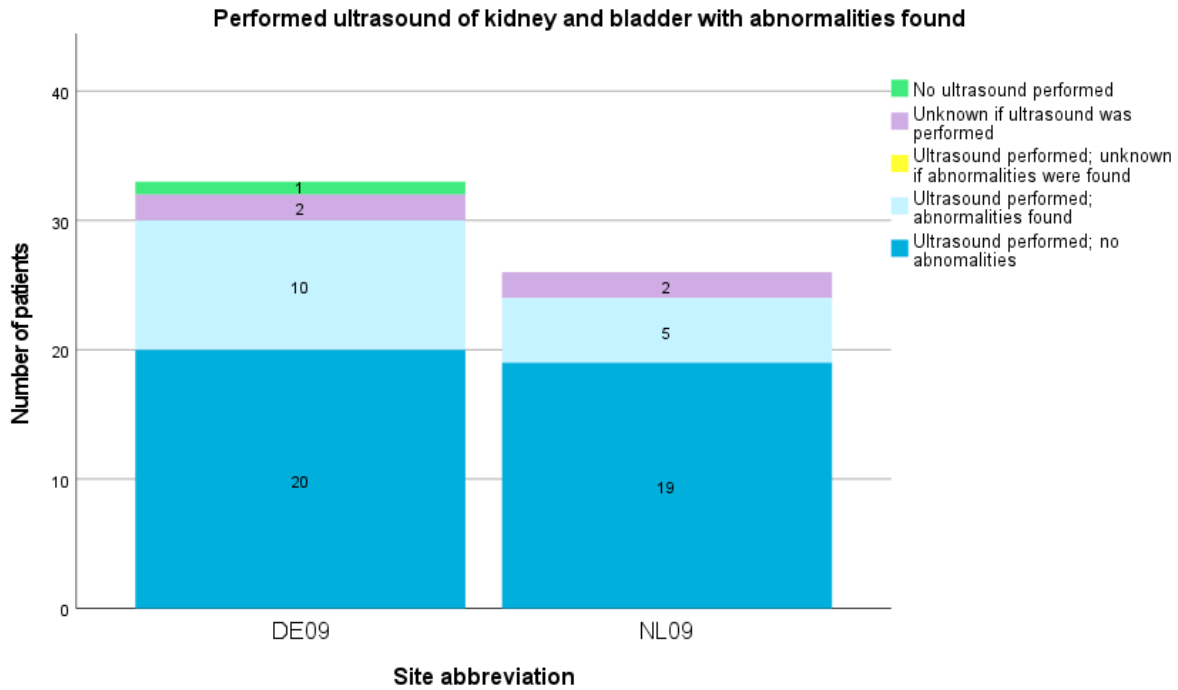
Months between first visit to HCP and VCUG

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



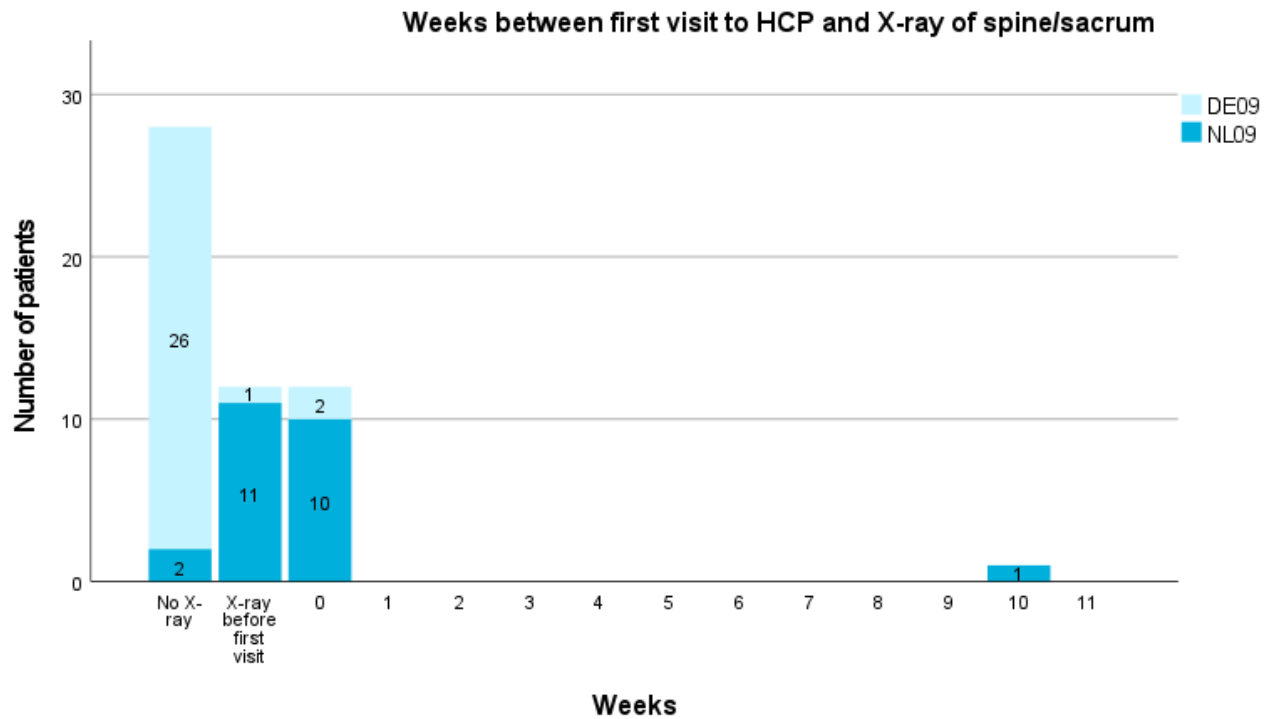
X-ray of spine/sacrum and abnormalities found

An X-ray was performed in almost all patients, but the majority of X-rays did not reveal any abnormalities.



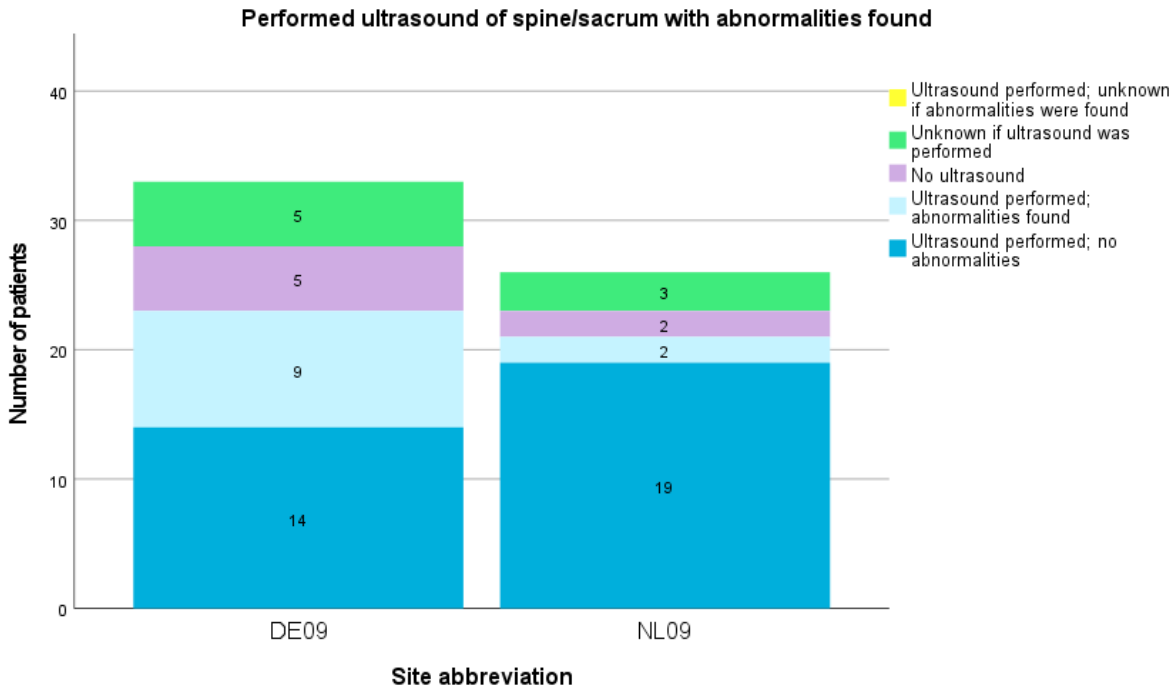
Months between first visit to HCP and X-ray of spine/sacrum

If the X-ray was not performed before the date of the first visit to the ERN eUROGEN HCP, the X-ray was almost always performed in the same week as the first visit to the HCP.



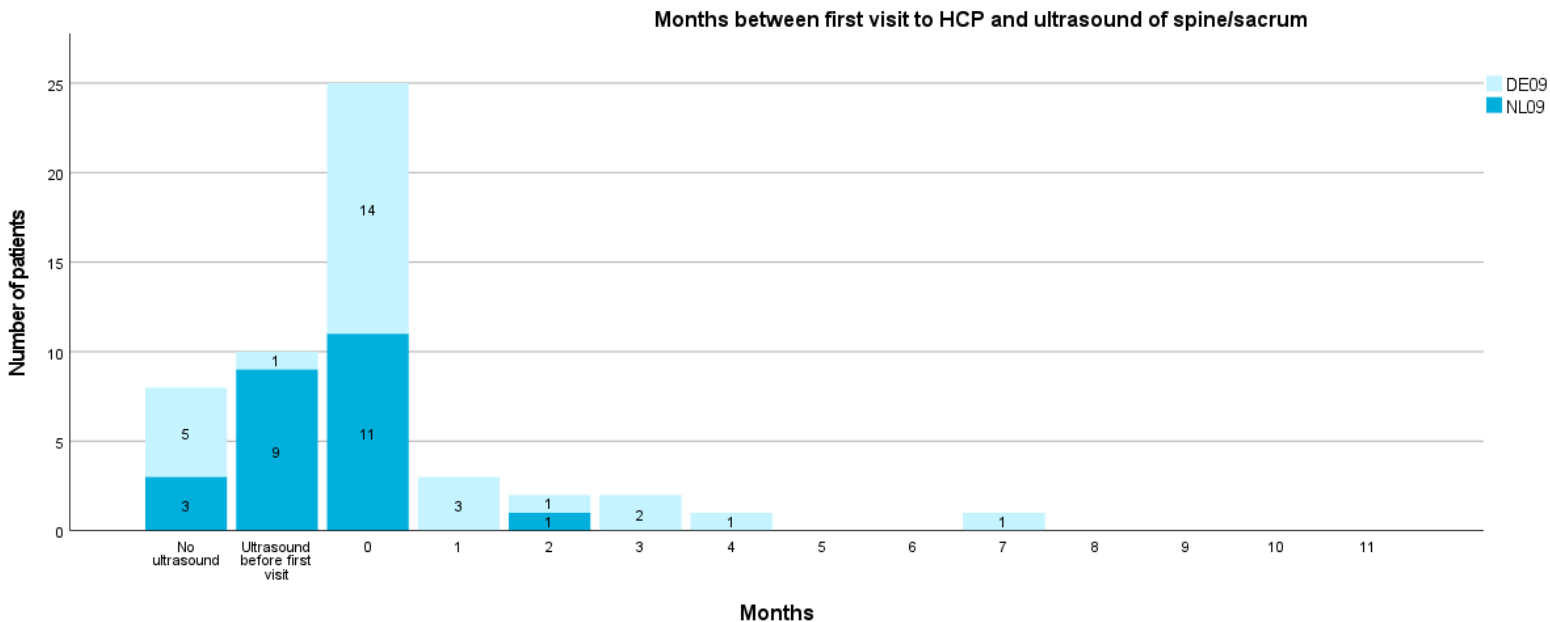
Ultrasound of spine/sacrum and abnormalities found

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



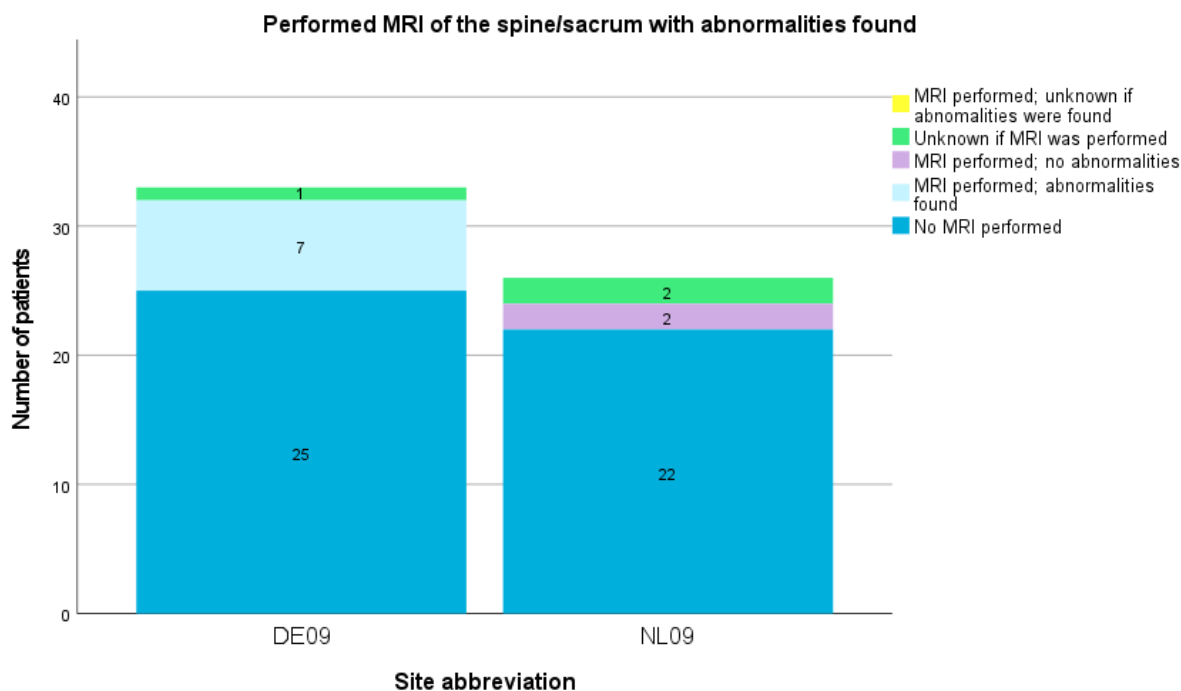
Months between first visit to HCP and ultrasound of spine/sacrum

If the X-ray was not performed before the date of the first visit to the ERN eUROGEN HCP, the ultrasound was almost always performed in the same month as the first visit to the HCP.



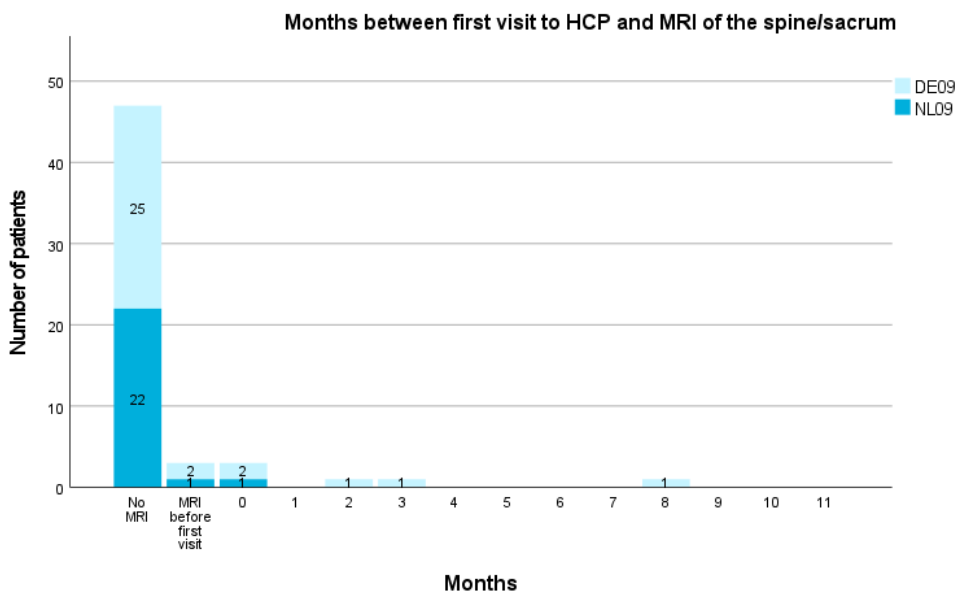
MRI of spine/sacrum and abnormalities found

For the majority of patients, no MRI was performed. For all DE09 patients with an MRI performed, abnormalities were found, while no abnormalities were found on MRIs performed in NL09.



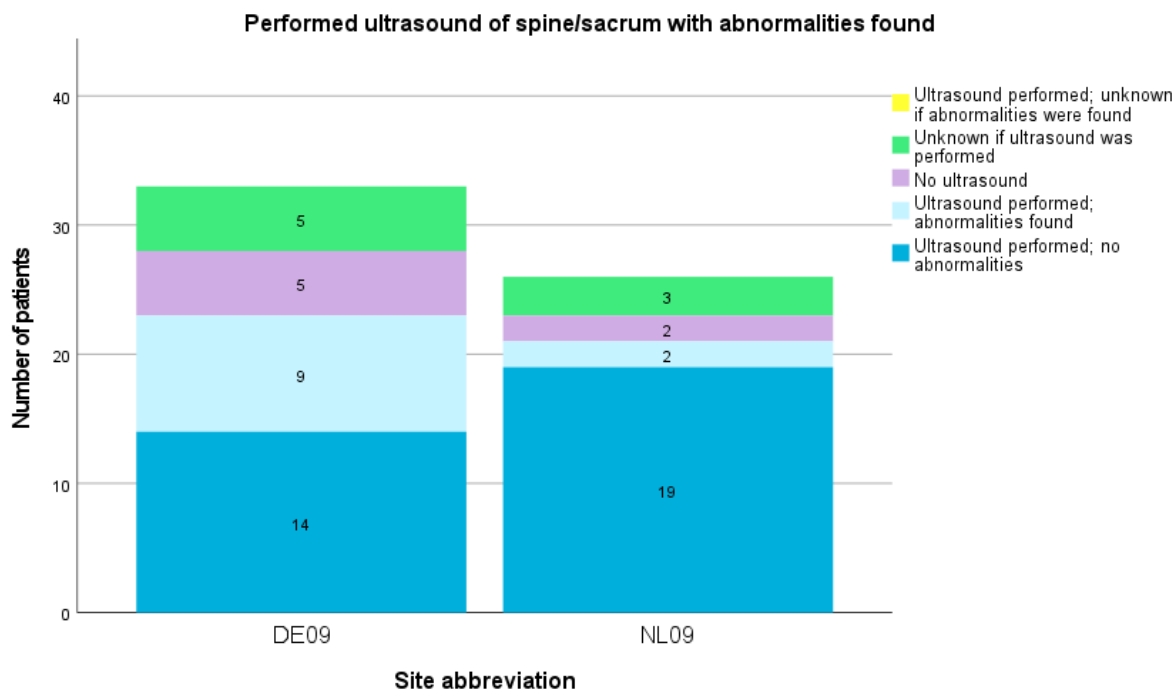
Months between first visit to HCP and MRI of spine/sacrum

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



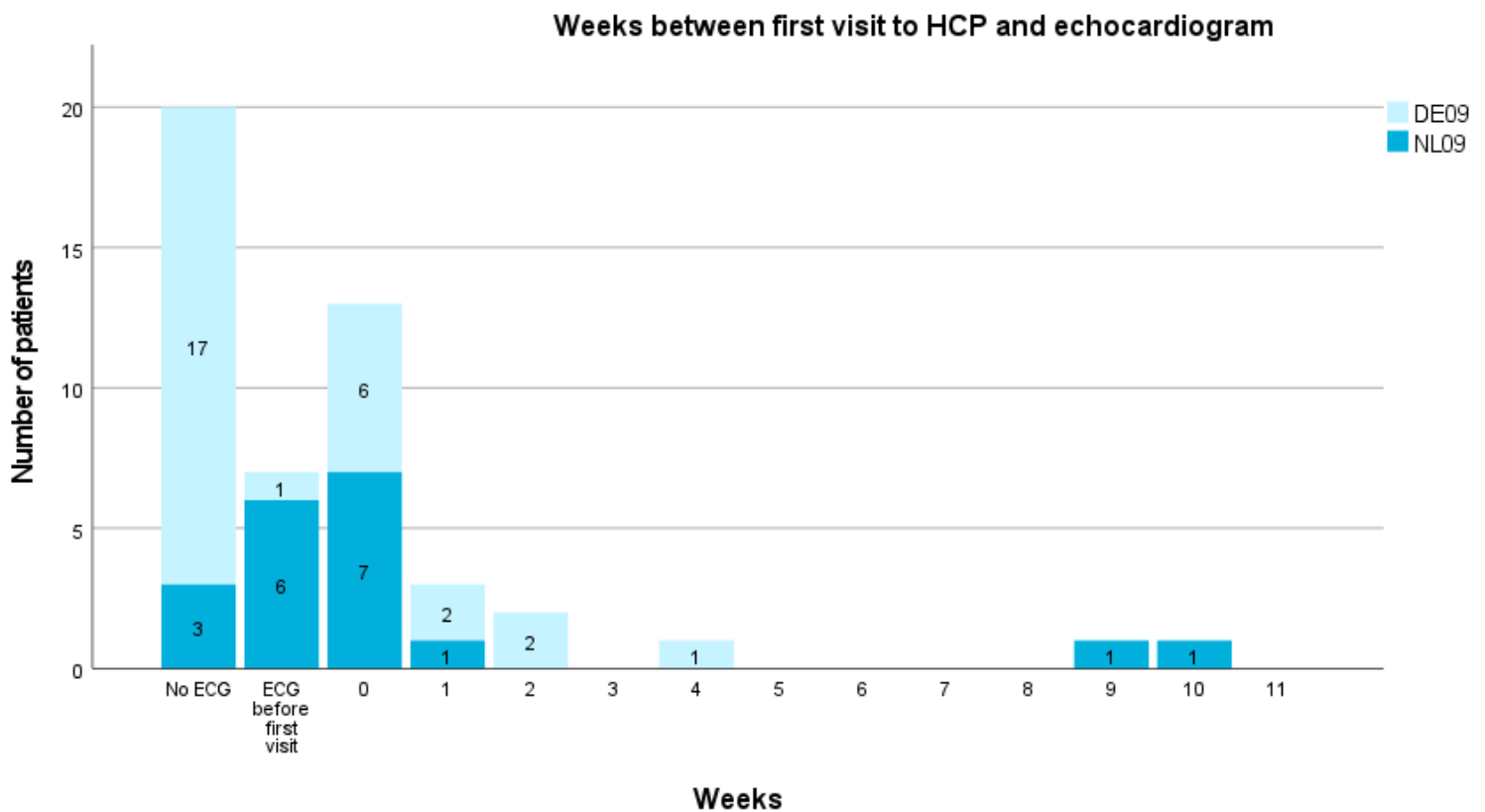
Echocardiogram (ECG) of spine/sacrum and abnormalities found

For the majority of patients, an ECG was performed. Most of the time, no abnormalities were found.



Months between first visit to HCP and ECG

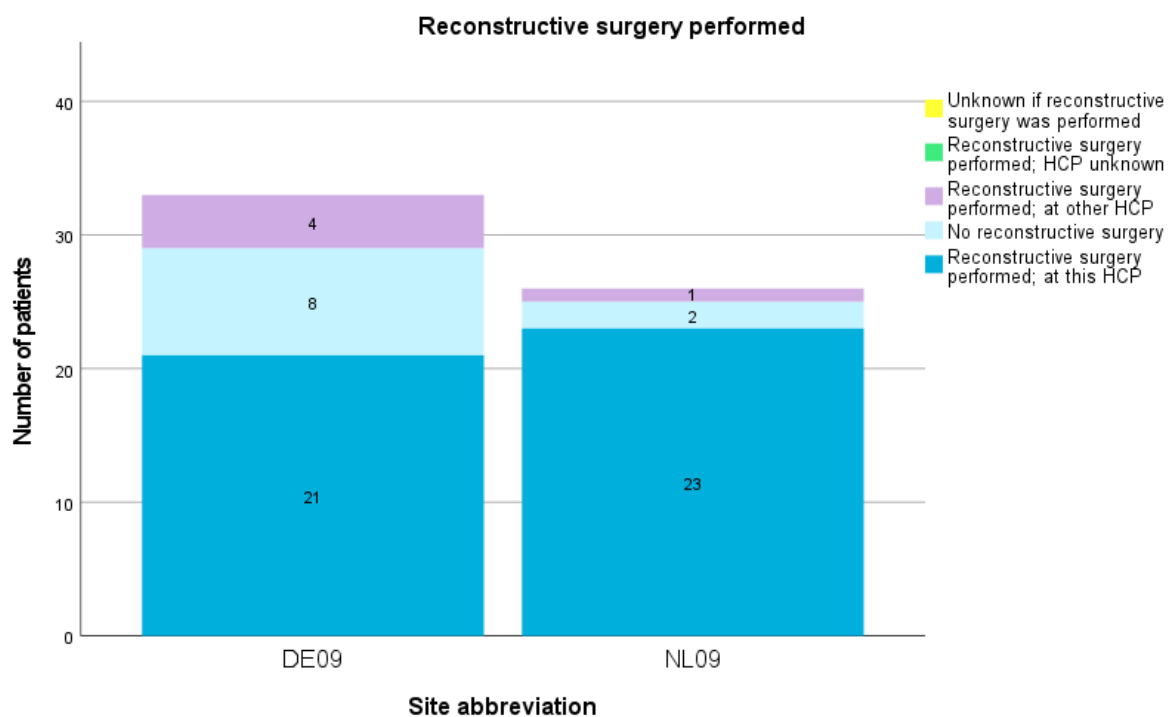
If the ECG was not performed before the first visit to the ERN eUROGEN HCP, the majority of ECGs was performed in the same week as the first visit to the HCP.



Surgery and treatment

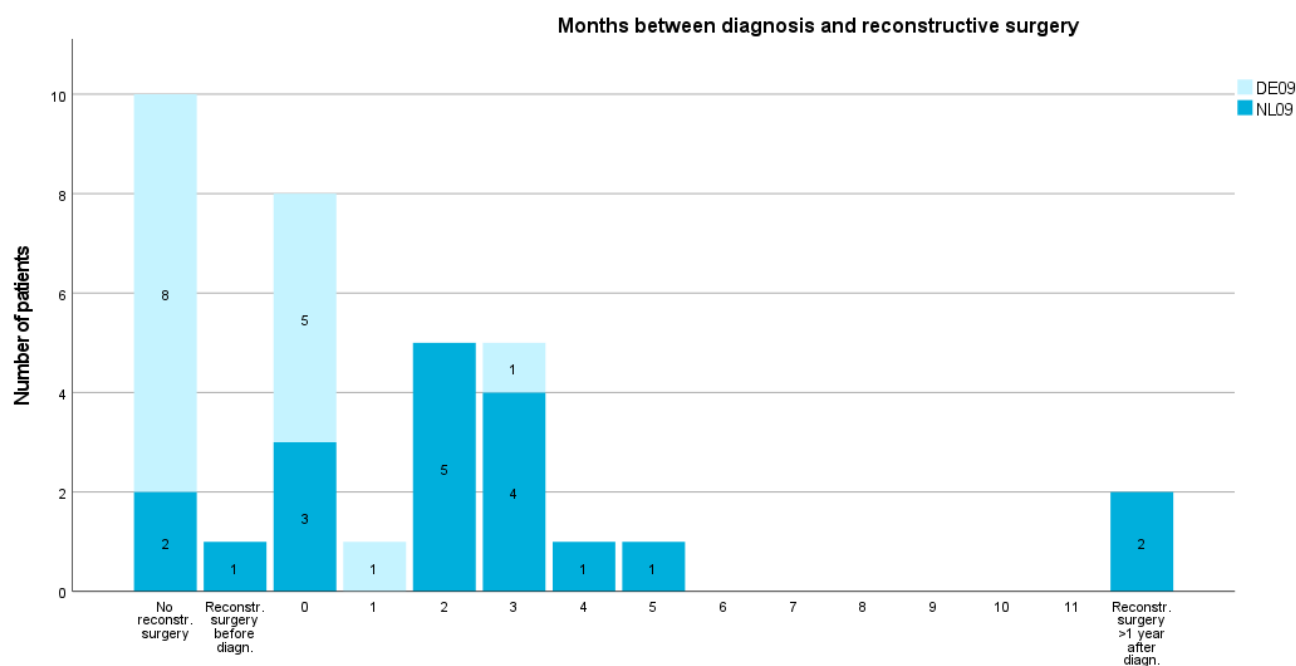
Reconstructive surgery

If the reconstructive surgery was performed, it was performed at the ERN eUROGEN HCP.



Months between diagnosis and reconstructive surgery

The majority of patients had the reconstructive surgery within 6 months after the diagnosis.



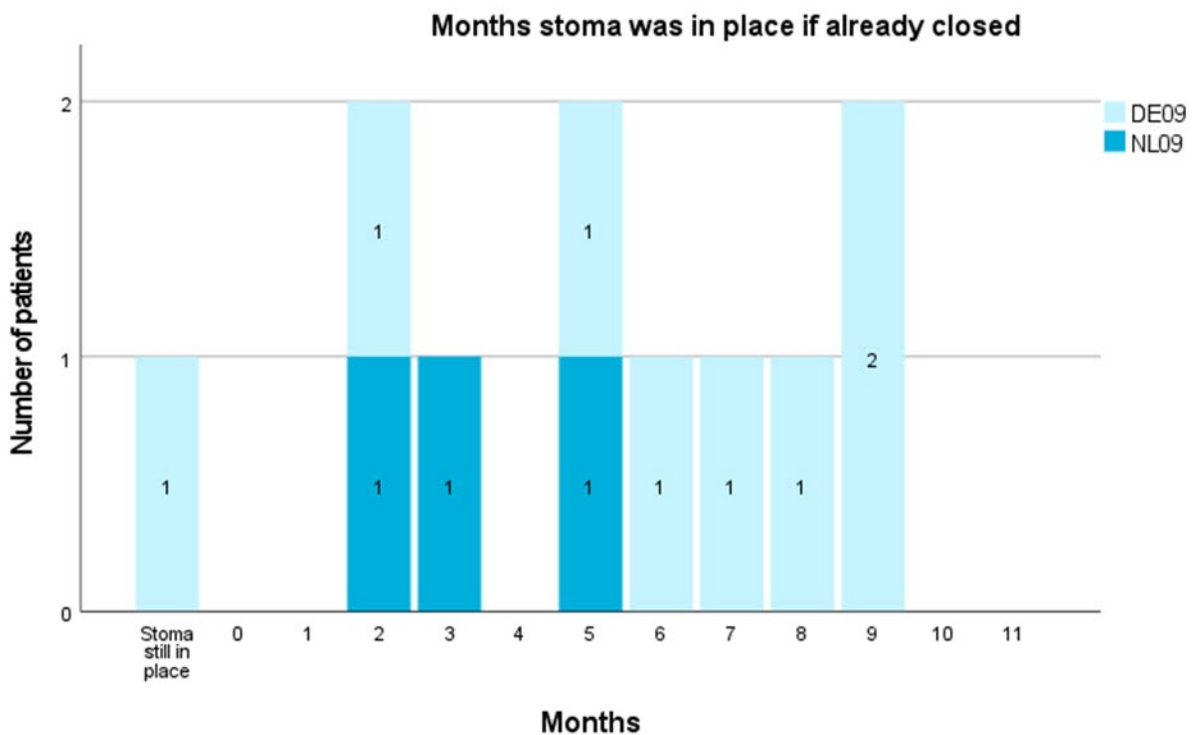
Stoma construction

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



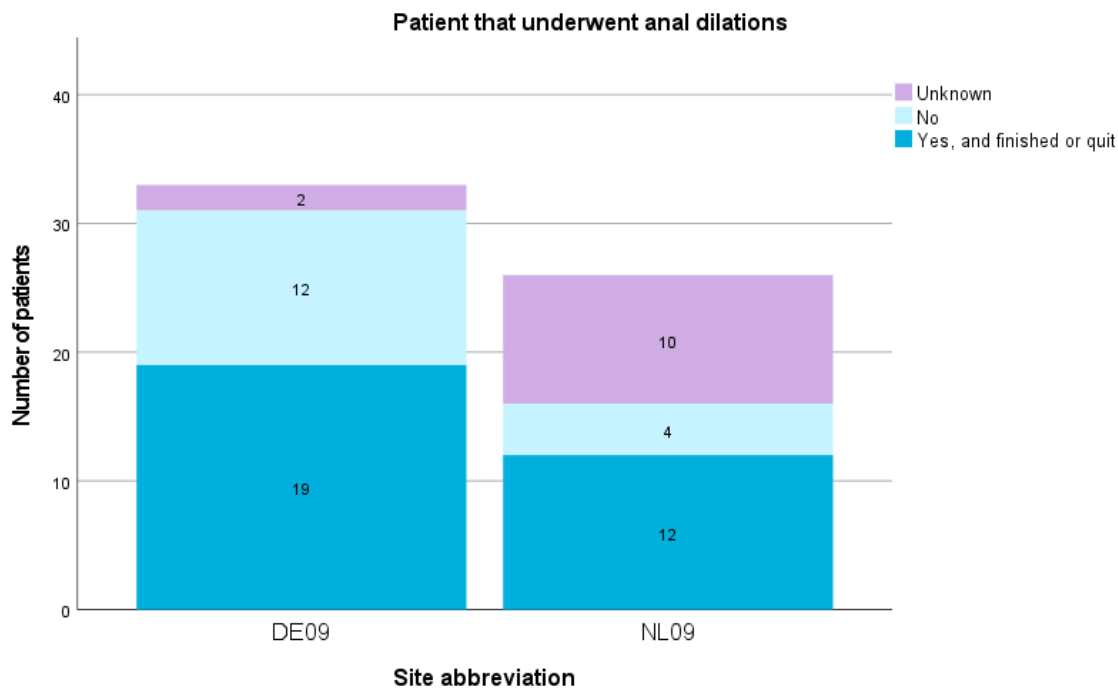
Months stoma was in place if already closed

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 months to 9 months.



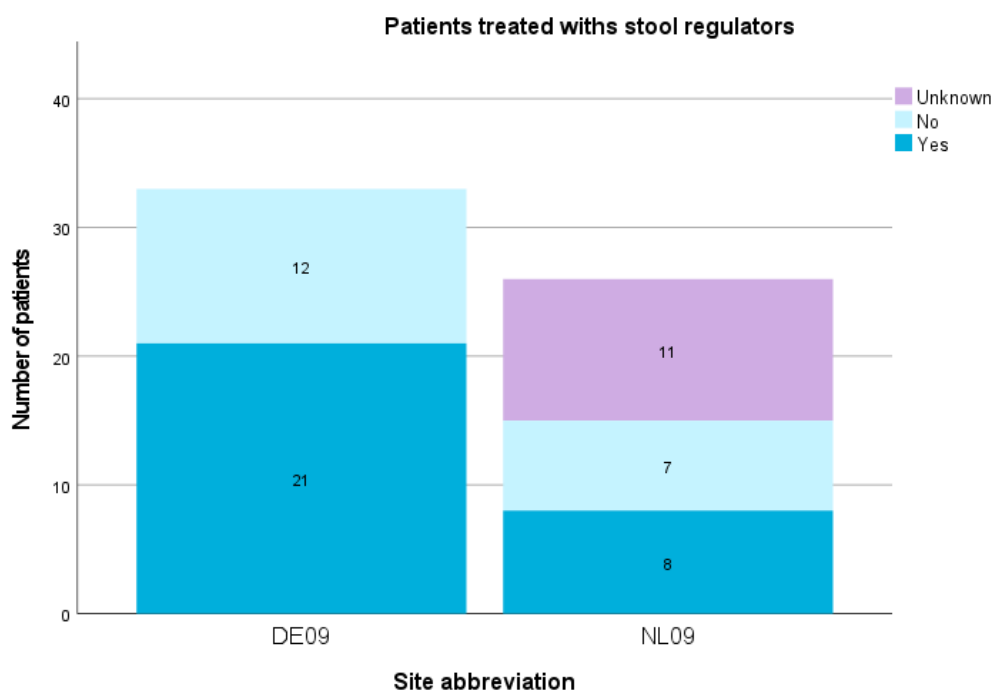
Anal dilations

The majority of patients needed anal dilations. For most patients, the dilations were either stopped or finished in the first year of treatment.



Treatment with stool regulators

Most patients were treated with stool regulators.





<http://euogen-ern.eu/>

Co-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>