# ERN eUROGEN registry
# Report on Expertise Area 1.1/1.2/1.4

## INTRODUCTION

This report entails an analysis of the data in the Common Data Elements of the ERN eUROGEN registry. This report encompasses Expertise Areas 1.1: complex genital reconstructions (DSDs), 1.2 Epispadias/BEEC/cloacal exstrophy and 1.4 Non-syndromic uro-recto-genital tract malformations. Please note that for these Expertise Areas, no Clinical Practice Snapshot is available, which is why only the Common Data Elements could be used. 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 1.1, 1.2, 1.4

# Descriptive statistics

The table below provides an overview of the descriptive statistics for patients from Expertise Area 1.1, 1.2, 1.4. Corresponding figures were made of the variables, and they are displayed on the next pages.

*1.1 Complex Genital reconstructions (DSD)*

| | Total N=32 | Germany DE22 (N=30) | Netherlands NL09 (N=2) |
|---|---|---|---|
| **Type of DSD** | | | |
| 46-XX DSD induced by fetal androgens excess, N (%) | 12 (37.5%) | 12 (40.0%) | - |
| 46-XY DSD, N (%) | 11 (34.4%) | 9 (30.0%) | 2 (100%) |
| Turner syndrome, N (%) | 6 (18.8%) | 6 (20.0%) | - |
| Classic congenital adrenal hyperplasia, N (%) | 3 (9.4%) | 3 (10.0%) | - |
| | | | |
| **Moment of diagnosis** | | | |
| Antenatal, N (%) | 2 (6.3%) | 1 (3.3%) | 1 (50.0%) |
| At birth, N (%) | 4 (12.5%) | 3 (10.0%) | 1 (50.0%) |
| After birth/at later age, N (%) | 26 (81.3%) | 26 (86.7%) | - |
| Undetermined, N (%) | - | - | - |
| | | | |
| **Moment of onset** | | | |
| Antenatal, N (%) | 1 (3.1%) | - | 1 (50.0%) |
| At birth, N (%) | 5 (15.6%) | 4 (13.3%) | 1 (50.0%) |
| After birth/at later age, N (%) | 24 (75.0%) | 24 (8000%) | - |
| Undetermined, N (%) | 2 (6.3%) | 2 (6.7%) | - |

*1.2 Isolated epispadias/BEEC/cloacal malformations*

| | Total N=27 | Czechia CZ02 (N=1) | Germany | | | Spain | | Italy IT34 (N=2) | The Netherlands NL09 (N=9) | Sweden SE01 (N=1) |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | DE09 (N=1) | DE19 (N=4) | DE22 (N=4) | ES10 (N=1) | ES17 (N=4) | | | |
| **Type of bladder exstrophy** | | | | | | | | | | |
| Isolated epispadias, N (%) | 6 (22.2%) | 1 (100%) | - | 2 (50.0%) | - | - | - | - | 3 (33.3%) | - |
| BEEC, N (%) | 10 (37.0%) | - | - | 1 (25.0%) | 1 (25.0%) | 1 (100%) | 3 (75.0%) | 1 (50.0%) | 3 (33.3%) | - |
| Cloacal exstrophy, N (%) | 11 (40.7%) | - | 1 (100%) | 1 (25.0%) | 3 (75.0%) | - | 1 (25.0%) | 1 (50.0%) | 3 (33.3%) | 1 (100%) |
| | | | | | | | | | | |
| **Birth sex** | | | | | | | | | | |
| Male, N (%) | 18 (66.7%) | - | 1 (100%) | 3 (75.0%) | 1 (25.0%) | 1 (100%) | 3 (75.0%) | 2 (100%) | 6 (66.7%) | 1 (100%) |
| Female, N (%) | 8 (29.6%) | 1 (100%) | - | 1 (25.0%) | 3 (75.0%) | - | 1 (25.0%) | - | 2 (22.2%) | - |
| Undetermined, N (%) | 1 (3.7%) | - | - | - | - | - | - | - | 1 (11.1%) | - |
| | | | | | | | | | | |
| **Moment of diagnosis** | | | | | | | | | | |
| Antenatal, N (%) | 3 (12.0%) | - | - | 1 (25.0%) | - | 1 (100%) | 1 (25.0%) | - | - | - |
| At birth, N (%) | 13 (52.0%) | - | - | 2 (50.0%) | 3 (75.0%) | - | 3 (75.0%) | 2 (100%) | 2 928.6%) | 1 (100%) |
| After birth/at later age, N (%) | 7 (28.0%) | 1 (100%) | - | 1 (25.0%) | 1 (25.0%) | - | - | - | 4 (57.1%) | - |
| Undetermined, N (%) | 2 (8.0%) | - | 1 (100%) | - | - | - | - | - | 1 (14.3%) | - |

*1.4 Non-syndromic uro-recto-genital tract malformations*

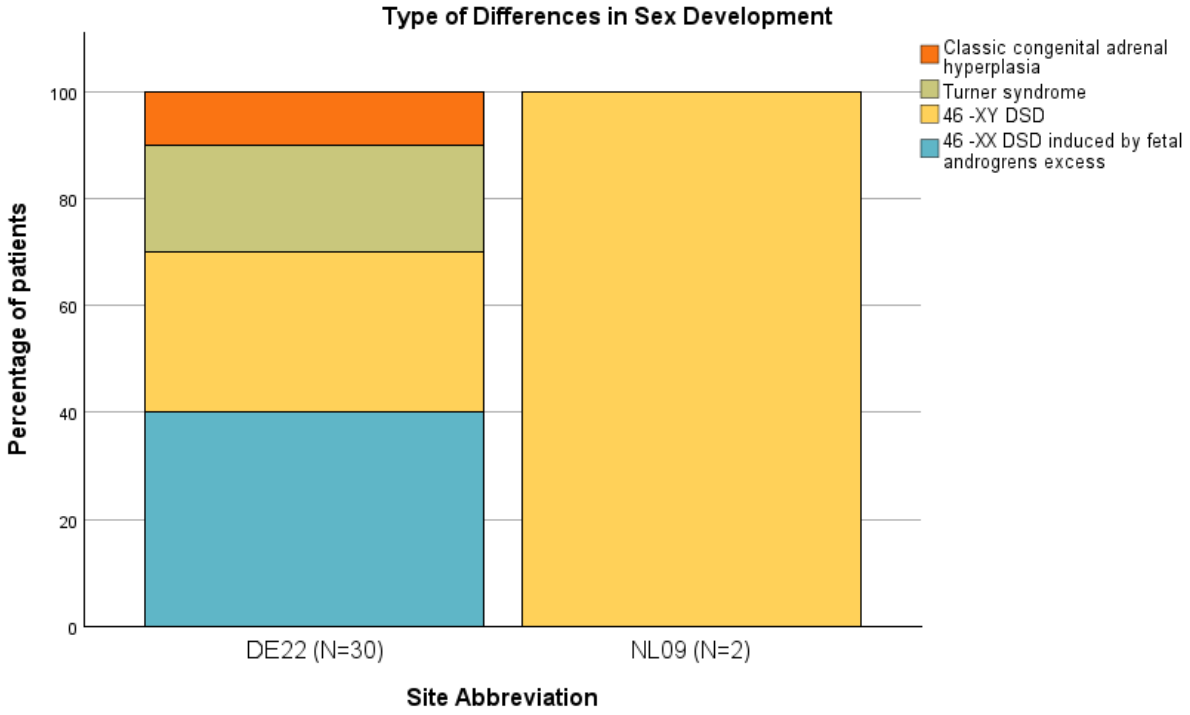
| | Total N=89 | Germany DE22 (N=88) | Netherlands NL09 (N=1) |
|---|---|---|---|
| **Type of non-syndromic malformation*** | | | |
| Septate vagina | 8 (9.0%) | 8 (9.1%) | - |
| Vaginal atresia | 5 (5.6%) | 5 (5.7%) | - |
| Congenital primary megaureter | 3 (3.4%) | 2 (2.3%) | 1 (100%) |
| Penoscrotal transpositions | 1 (1.1%) | 1 (1.1%) | - |
| Fetal lower urinary tract obstruction | 1 (1.1%) | 1 (1.1%) | - |
| MRKH (Mayer-Rokitansky-Küster-Hauser) syndrome | 71 (79.8%) | 71 (80.7%) | - |
| | | | |
| **Moment of diagnosis** | | | |
| Antenatal, N (%) | - | - | - |
| At birth, N (%) | 3 (3.4%) | 3 (3.4%) | - |
| After birth/at later age, N (%) | 84 (96.6%) | 84 (96.6%) | 1 (100%) |

*Only options that are selected are displayed.

## 1.1 Complex genital reconstructions (DSDs)

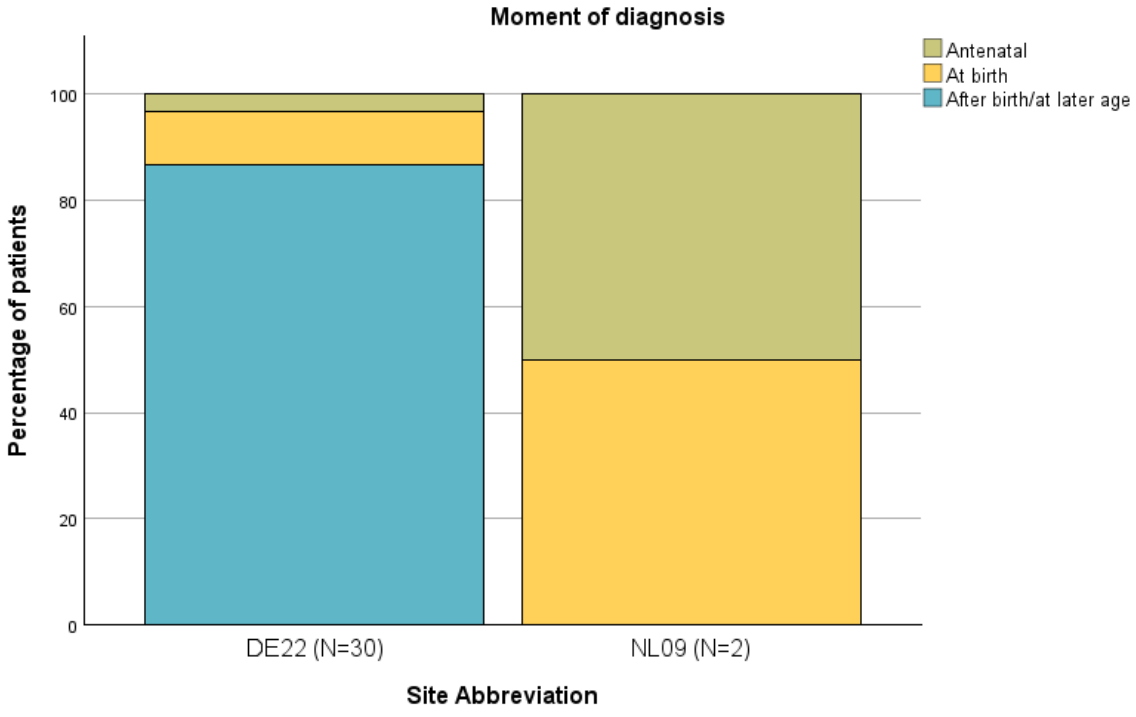
*Type of DSD*

Most patients included had 46-XX DSD induced by fetal androgens excess. Classic congenital adrenal hyperplasia occurred in the least patients.



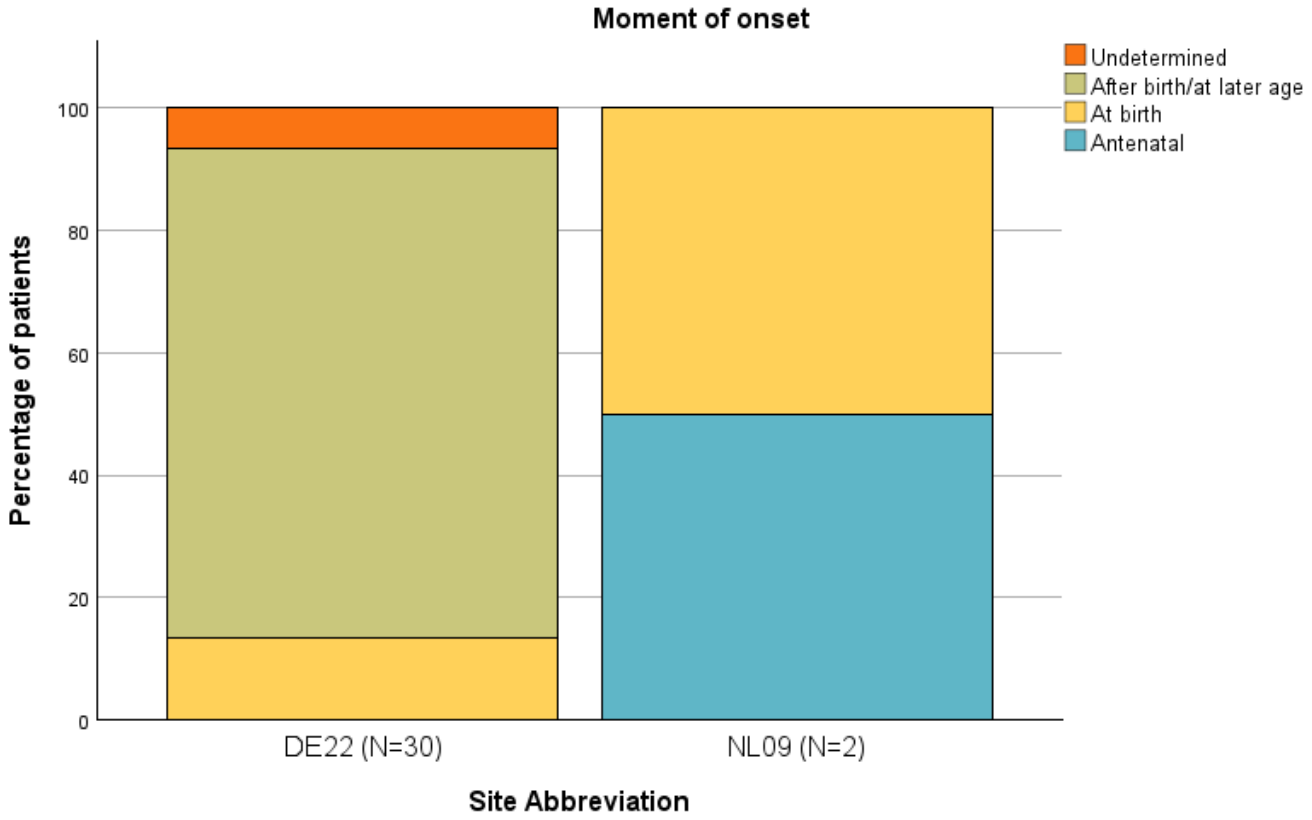

*Moment of diagnosis*

Most patients were diagnosed after birth or at later age in DE22, where in NL09, diagnoses was either made antenatal or at birth.



*Moment of onset*

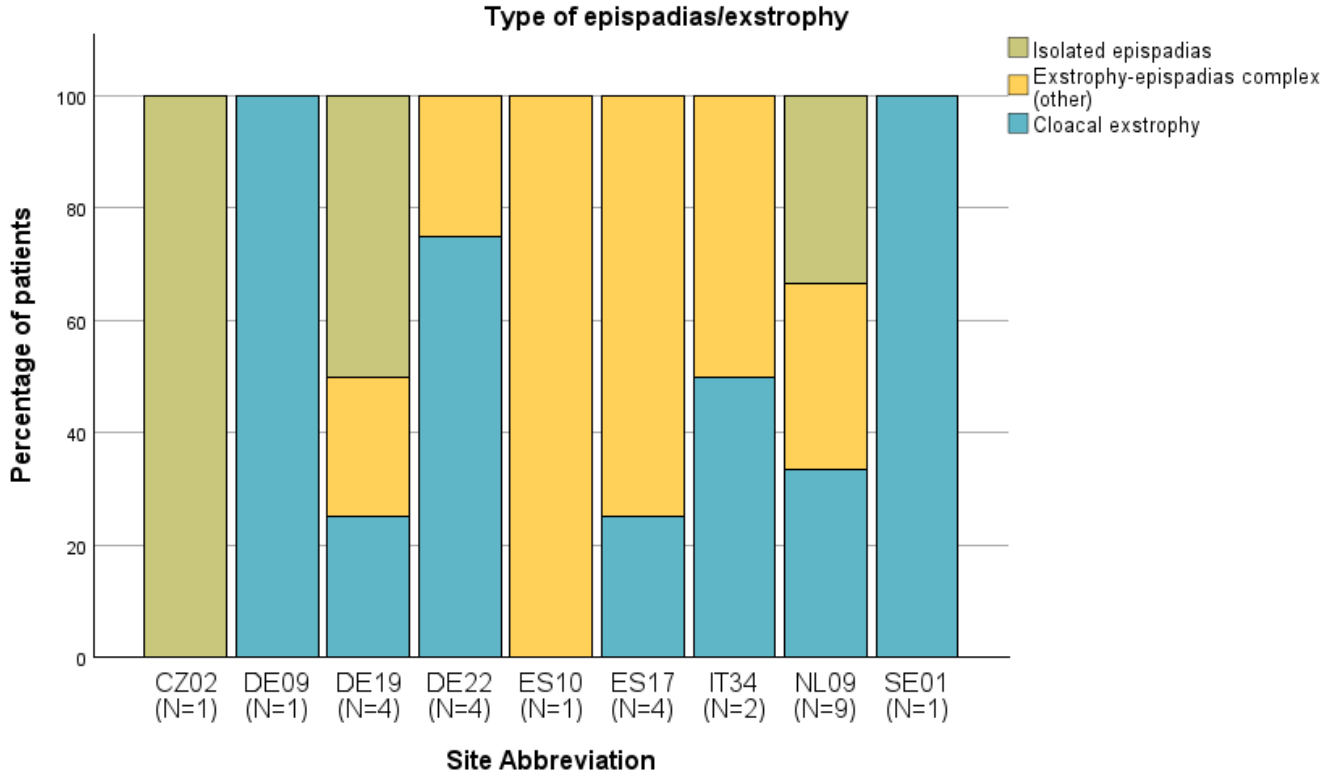
For most patients onset was after birth or at later age in DE22, where in NL09, onset was either antenatal or at birth.




## 1.2 Isolated epispadias/BEEC/cloacal malformations

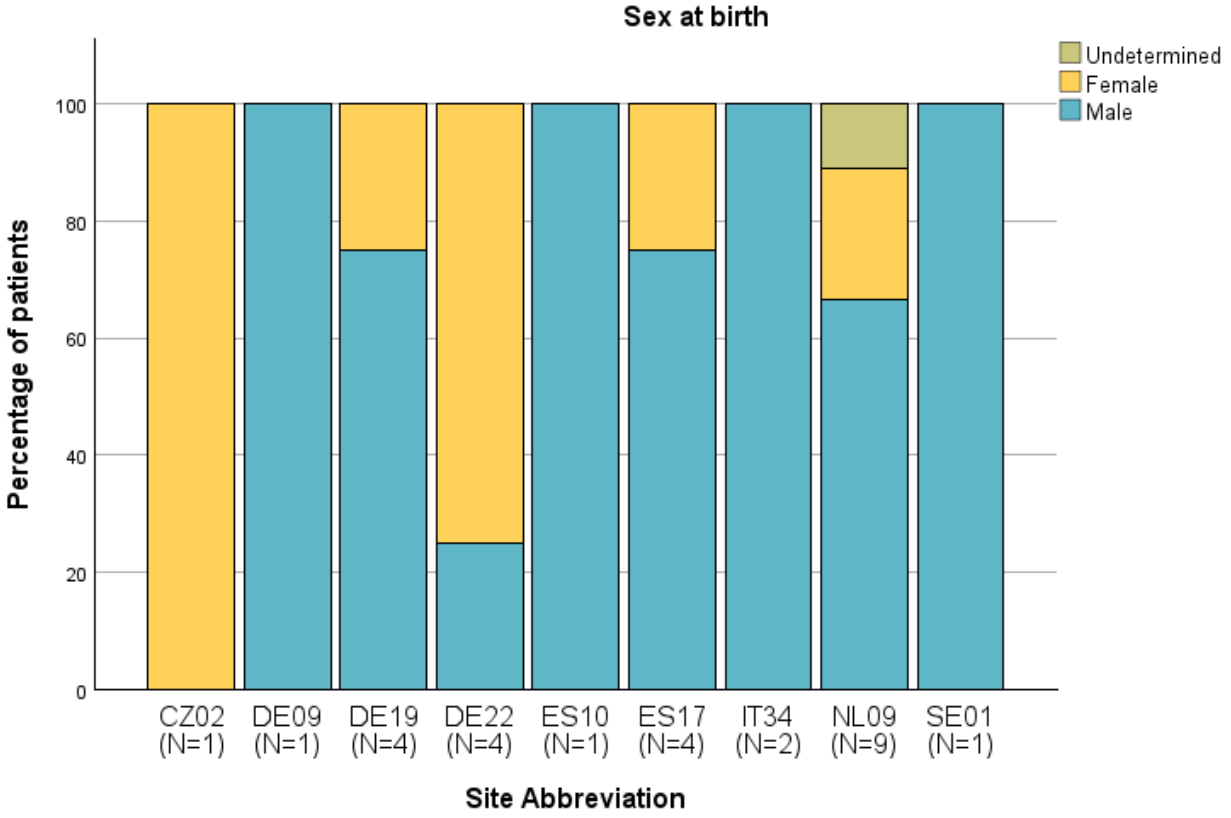
*Type of epispadias/exstrophy*

The type of epispadias/exstrophy differs per HCP. Most patients had a cloacal exstrophy, followed closely by isolated epispadias.



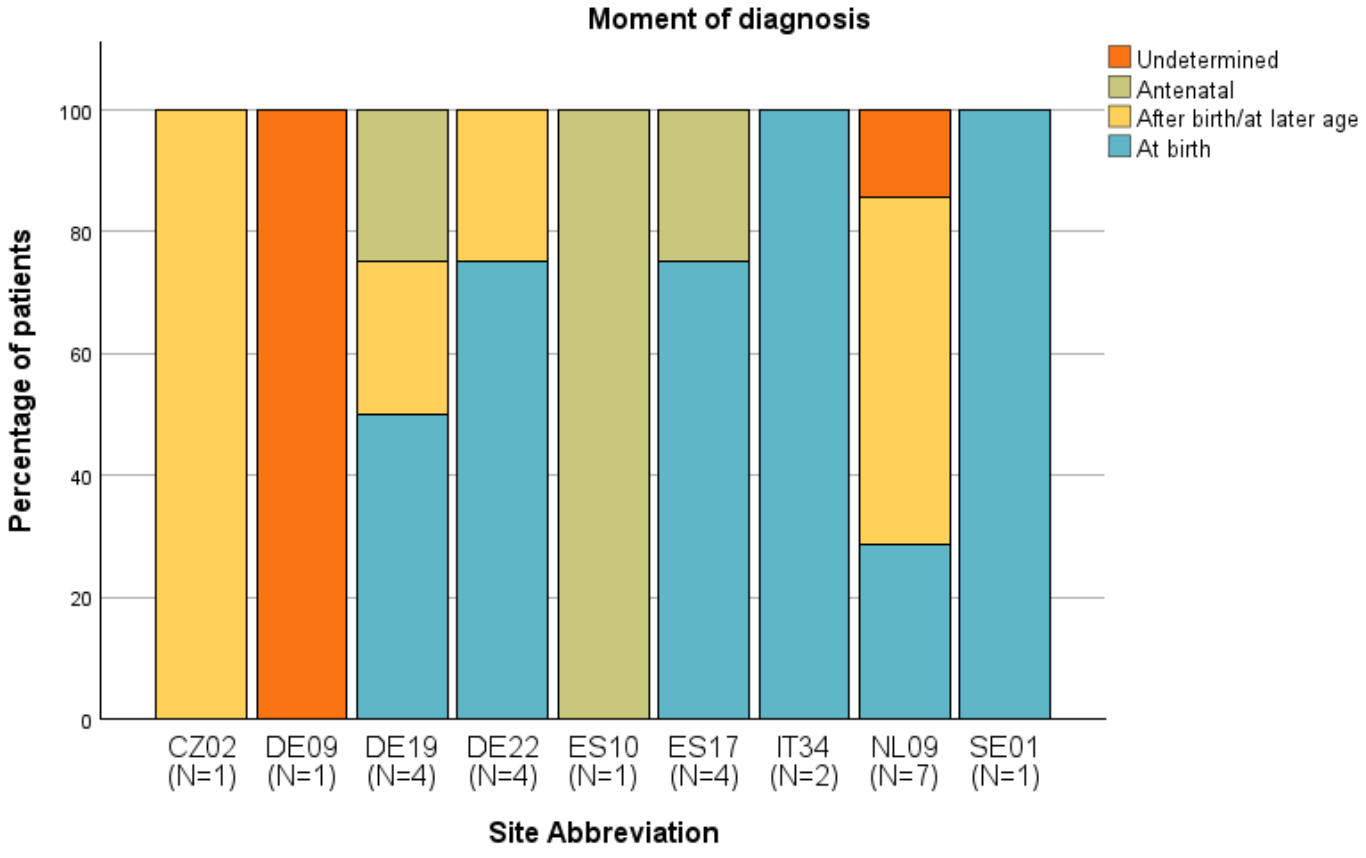
*Sex at birth*

Most patients were male, only at CZ02 and DE22, females were in majority.




*Moment of diagnosis*

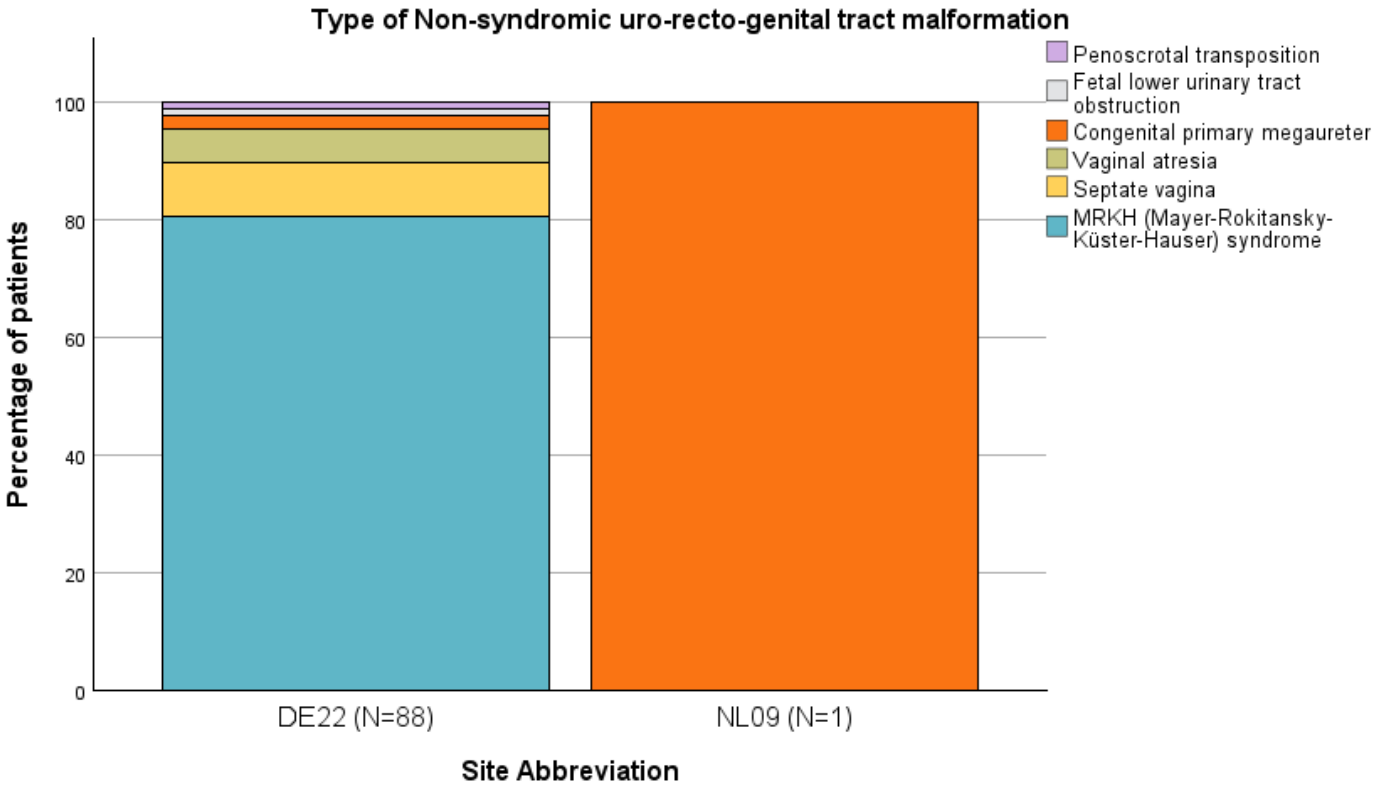
Most patients were diagnosed at birth.



## 1.4 Non-syndromic uro-recto-genital malformations

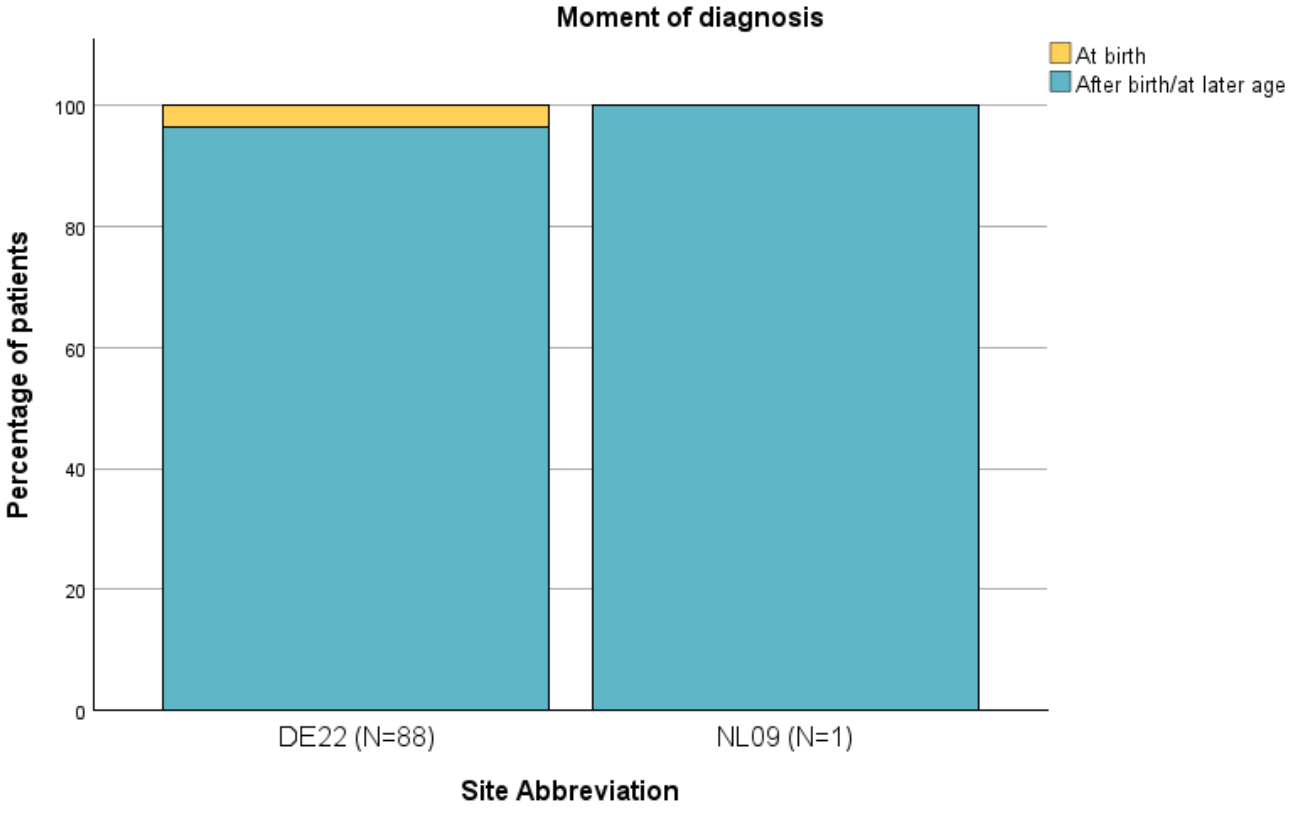
*Type of non-syndromic malformation*

Most patients were diagnosed with MRKH syndrome, followed by septate vagina.




*Time between first contact with Health Care Provider and adrenal surgery*

Almost all patients were diagnosed after birth or at later age.

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