# A comparison of birth defects self-reported by mothers with data provided by general practitioners -

*data from the Dutch Pregnancy Drug Register*

01-09-2023

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***Version with updated data (07-09-2023)***

# Birth defects

- ≈ 170.000 children are born per year in the Netherlands
  - Every 3 in 100 children are born with a congenital birth defect
    - About 2-3% of these birth defects are caused by teratogenic medication use during pregnancy

- For only 1 in 5 medicines information on its safety during pregnancy is known

- The presence of a birth defect in a infant is often one of the primary outcomes in drugs-safety studies

# Aim of the study

- What is the best and most efficient way to retrieve information birth defects?
- Is there a golden standard?
- Who has the most accurate information?

**Aim of this study**

*to compare self-reported information on the offspring's birth defects by mothers with the information provided by GPs.*

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# The Dutch Pregnancy Drug Register

- Ongoing prospective population-based cohort study
  - Currently ≈ 20.000 inclusions
- Aim: to obtain insight into medication use among pregnant and breastfeeding women and its potential effects on maternal and fetal/infant health
- **All** pregnant women ≥18 years of age can participate
  - Independent of (chronic) medication use
  - Preferably enroll as early as possible in pregnancy

- Topics; obstetric history, (chronic) health conditions, vaccinations, vitamins, medication use, demographic factors, prenatal screening, pregnancy- birth outcomes, breastfeeding, biological father, etc.

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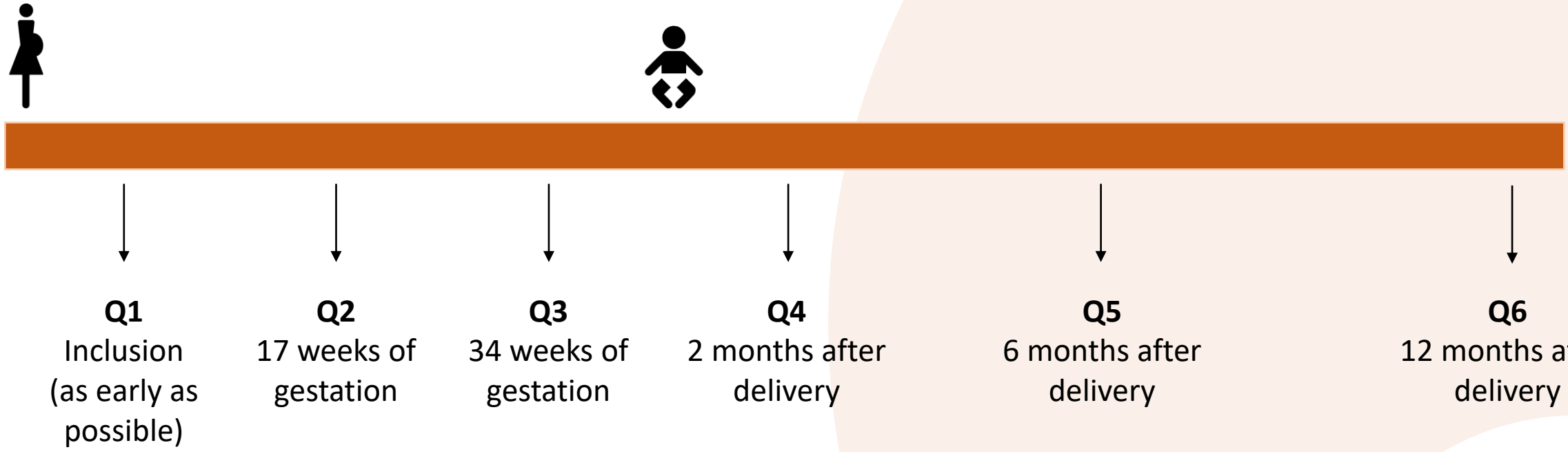
# The Dutch Pregnancy Drug Register

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# Inclusion criteria

Participants enrolled between **2014 and 2019** and adhered to the following criteria were included

1. had a singleton live-birth;
2. completed at least the fifth questionnaire of the Dutch Pregnancy Register, 6-months after expected due date;
3. gave permission to contact their GP for follow-up information and
4. provided the contact information of their GP

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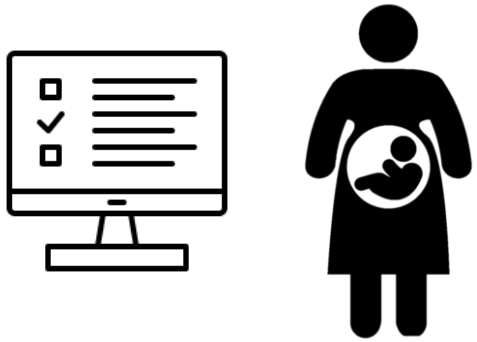
# Data collection



- **Demographics**
- **Birth defect**
  - the existence of possible birth defect
  - any other neonatal health specifics
  - reasons for a possible hospital admissions
  - final remarks

- **Birth defect**
  - the existence or suspicion of a birth defect;
  - initiation of the symptoms
  - the exact time and official diagnosis
  - if any additional diagnostic tests were performed

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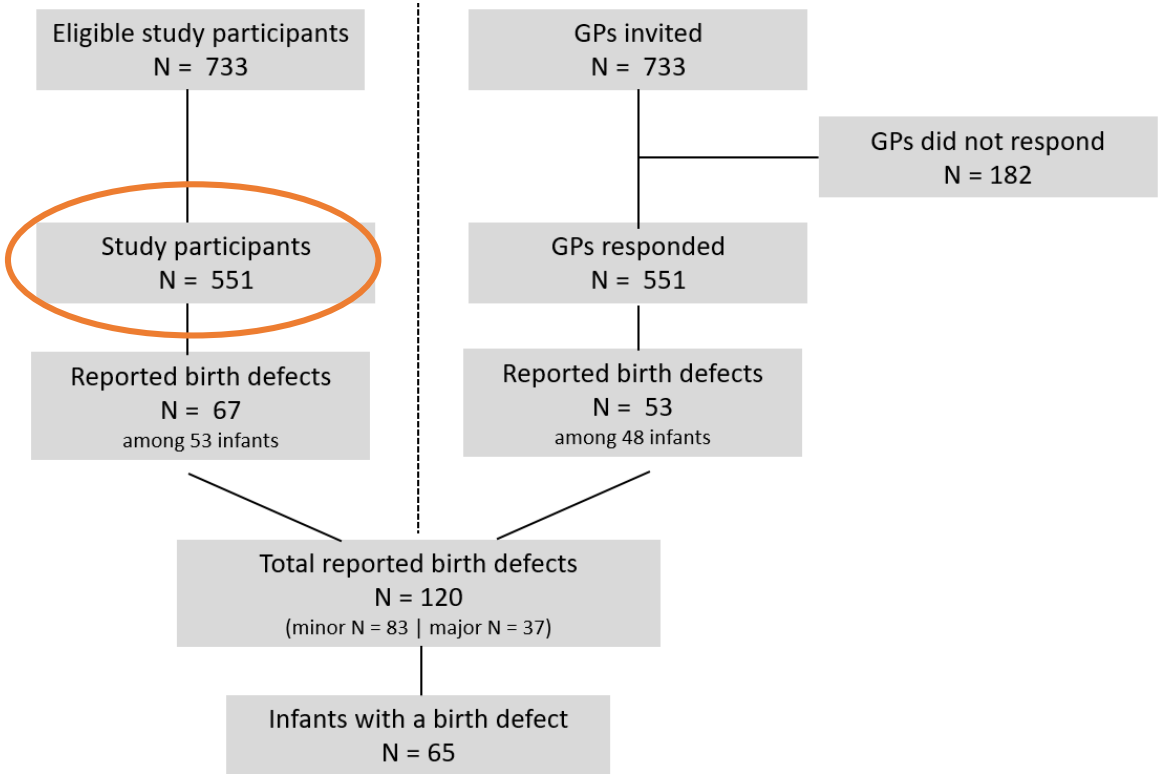
# Data analysis

- All reported congenital birth defects were collected
- Blindly assessed by two independent researchers + clinical geneticist
  - Coded using the ICD-10 Index
  - Classified using the EUROCAT classifications
- Paired back to the same infant
- Differences in reported birth defects between the participant and their GP were assessed

| Information | ICD-10 code | EUROCAT classification |
|---|---|---|
| Hip dysplasia | Q56.8 - Other congenital deformities of hip | Minor |
| Tongue tie too short | Q38.1 - Ankyloglossia | Minor |
| Atrial septal defect | Q21.1 - Atrial septal defect | Major |
| Missing a toe on his right foot | Q72.3 - Congenital absence of foot and toe(s) | Major |

# Study participants




| Participants | N (%) |
|---|---|
| | **N = 551** |
| **Age** (Median (IQR)) | 31 (29-34) |
| **Educational level** | |
| High | 430 (78.3) |
| Low/Medium | 119 (21.7) |
| Missing | 2 |
| **Self-defined ethnicity** | |
| Dutch | 524 (95.3) |
| Non-Dutch | 26 (4.7) |
| Missing | 1 |
| **Pre-pregnancy BMI** (Median (IQR)) | 22.8 (20.8 – 25.7) |
| Underweight (≤19.0) | 25 (5.0) |
| Normal weight (19.0 – 24.9) | 330 (66.0) |
| Overweight (25.0 – 29.9) | 106 (21.2) |
| Obese (≥30.0) | 39 (7.8) |
| Missing | 51 |
| **Parity** | |
| Primipara | 252 (46.4) |
| Multipara | 291 (53.6) |
| Missing | 8 |

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# Classifications of infants

None

≥1 Minor

≥1 Major

94%

None

≥1 Major

98%

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# Reported birth defects

| | Both GP and participant reported a birth defect N = 78 (65.0%) | | Either GP or participant reported a birth defect N = 42 (35.0%) |
|---|---|---|---|
| | ICD-10 code identical | | |
| | Yes N = 60 (76.9%) | No N = 18 (23.1%) | |
| **Participant** | | | |
| Minor (N=48) | 20 (41.7%) | 4 (8.3%) | 24 (50.0%) |
| Major (N=19) | 10 (52.6%) | 5 (26.3%) | 4 (21.1%) |
| | | | |
| **GP** | | | |
| Minor (N=35) | 21 (60.0%) | 3 (8.6%) | 11 (31.4%) |
| Major (N=18) | 9 (50.0%) | 6 (33.3%) | 3 (16.7%) |

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# Reported birth defects

| ICD-10 code | | | ICD-10 code | | |
|---|---|---|---|---|---|
| Q65.8 | Q65.2 | ✗ | Q65 | Q65 | ✓ 56% |
| Q74.8 | Q72.8 | ✗ | Q74 | Q72 | ✗ |

| | **Both GP and participant reported a birth defect N = 78 (65.0%)** | | Either GP or participant reported a birth defect N = 42 (35.0%) |
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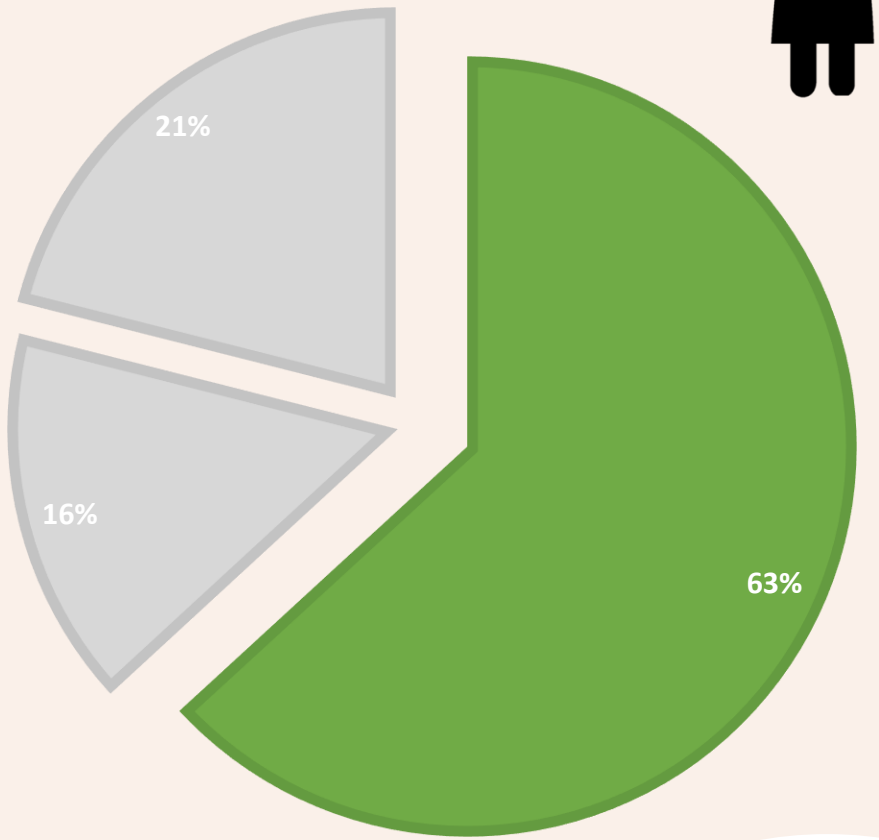
# Major birth defects

In 12 cases (63%) identical birth defects reported by GP

| Participant | | GP | |
|---|---|---|---|
| | EUROCAT | EUROCAT | |
| Pulmonary valve stenosis (mild) | **Major** | **Major** | Mild pulmonary valve stenosis |
| Missing a toe on his right foot | **Major** | **Major** | Missing 1 toe on right foot |
| Ventricular septal defect | **Major** | **Major** | VSD |
| Etc. | | | |

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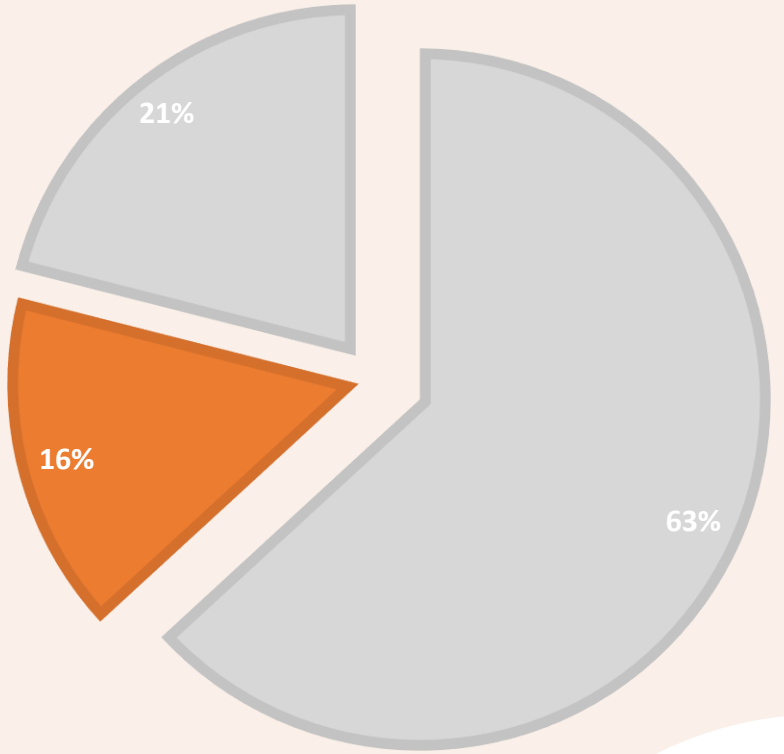
# Major birth defects

In 3 cases (16%) GP reported a minor birth defect

| Participant | | GP | |
|---|---|---|---|
| | EUROCAT | EUROCAT | |
| Only 1 functioning testis | **Major** | Minor | Antenatal torsion testis |
| Atrial septal defect | **Major** | Minor | Open foramen oval |
| Hypospadias light form | **Major** | Minor | Congenital penile curvature |

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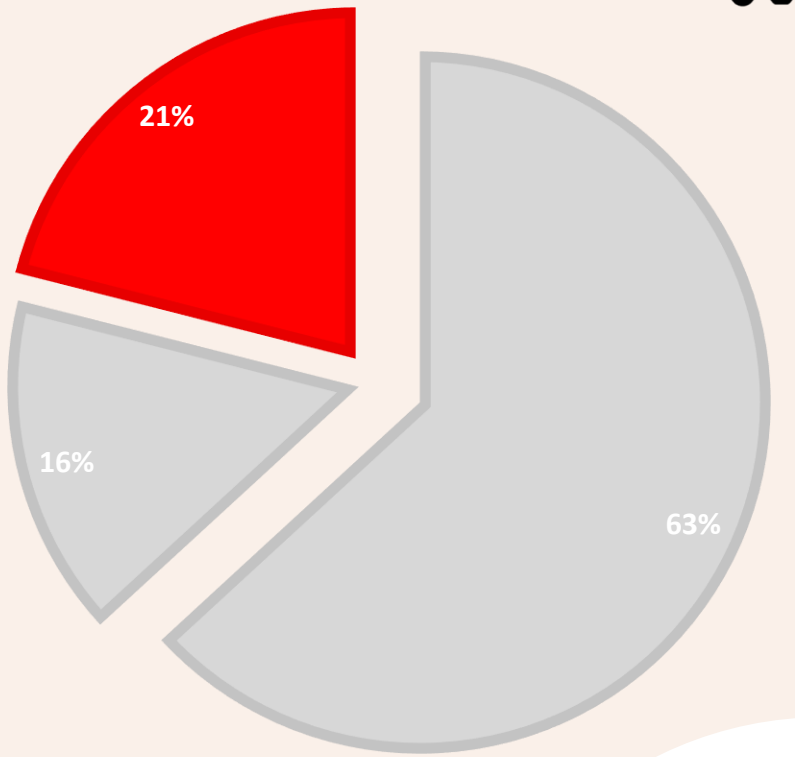
# Major birth defects

In 4 cases (21%) GP reported no birth defect

| Participant | | GP |
|---|---|---|
| | EUROCAT | |
| Hip luxation right | **Major** | |
| Pulmonary stenosis | **Major** | |
| Ventricular septal defect | **Major** | |
| Ventricular septal defect | **Major** | |

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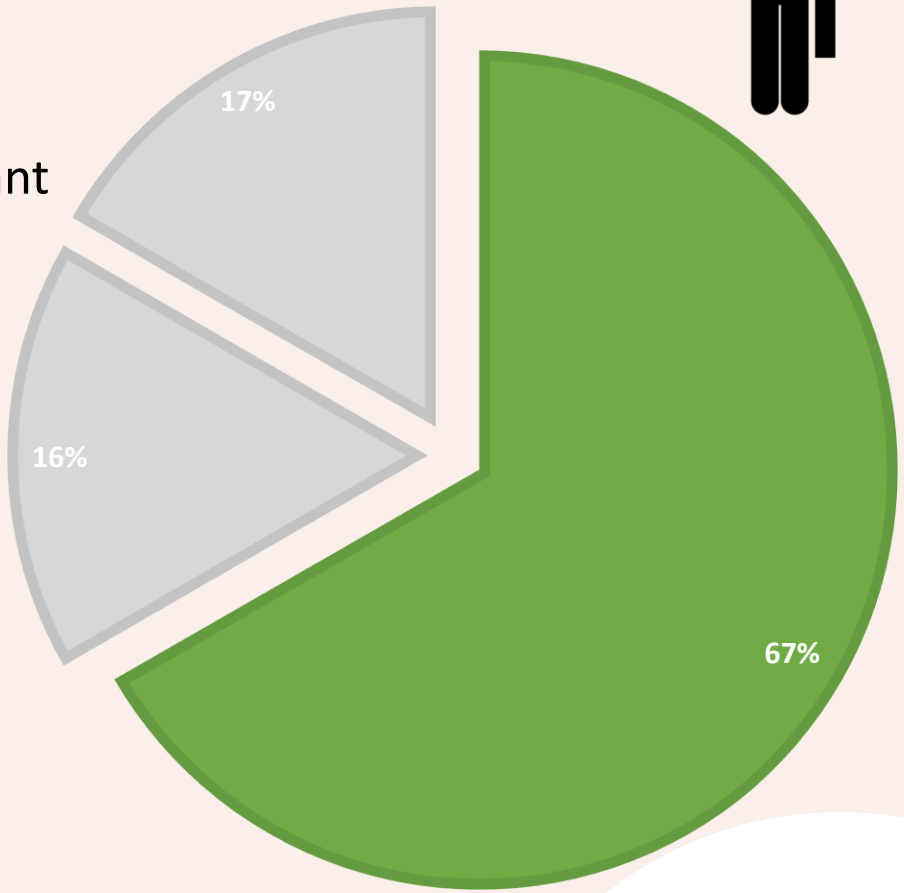
# Major birth defects

In the **same** 12 cases (67%) identical birth defect reported by participant

| Participant | | GP | |
|---|---|---|---|
| | EUROCAT | EUROCAT | |
| Hypospadias | **Major** | **Major** | Glandular hypospadias |
| Esophageal atresia with tracheal fistula | **Major** | **Major** | Oesophageal atresia |
| Polydactyly | **Major** | **Major** | Polydactyly |
| Etc. | | | |

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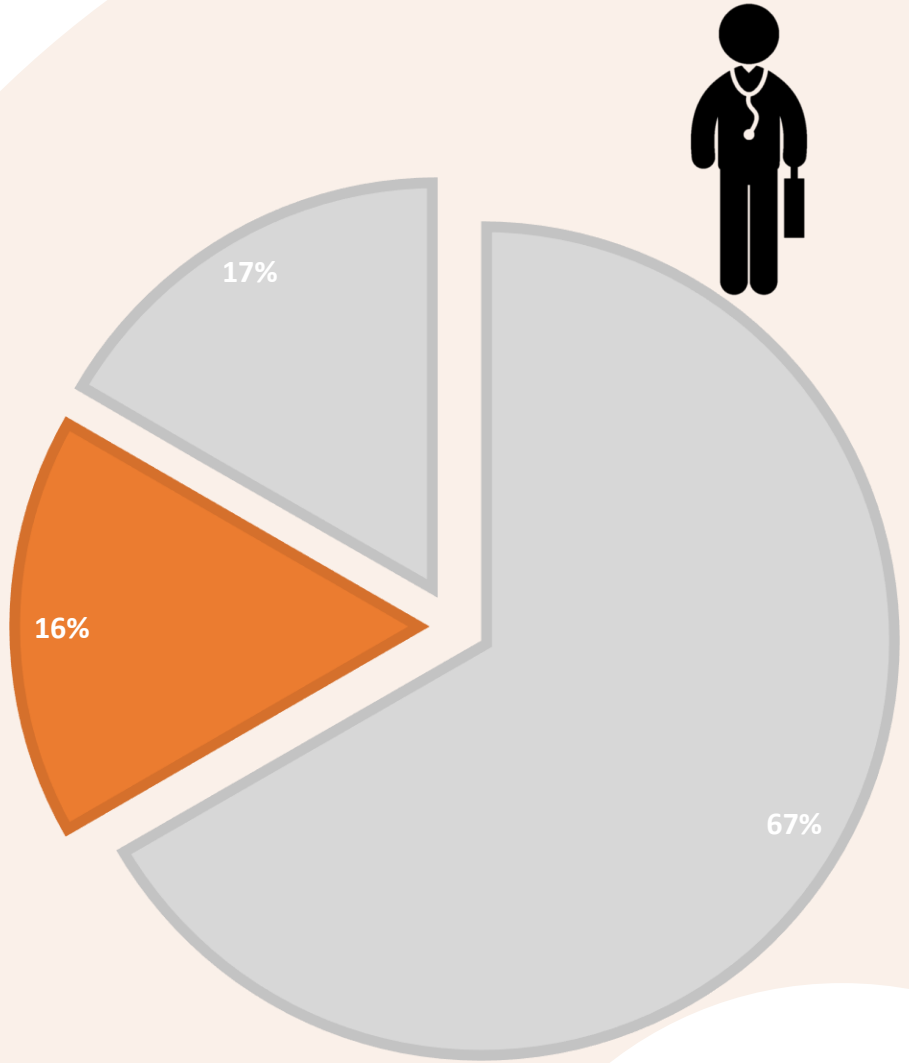
# Major birth defects

In 3 cases (16%) participant reported a minor birth defect

| Participant | | GP | |
|---|---|---|---|
| | EUROCAT | EUROCAT | |
| Hip dysplasia | Minor | **Major** | Congenital hip dislocation |
| A number of cysts with lymphatic fluid in the back of the neck | Minor | **Major** | Occipital encephalocele at the level of the cerebellum |
| Urine flowed back from the bladder to the kidney (vesicoureteral reflux) | Minor | **Major** | Congenital double system left kidney |

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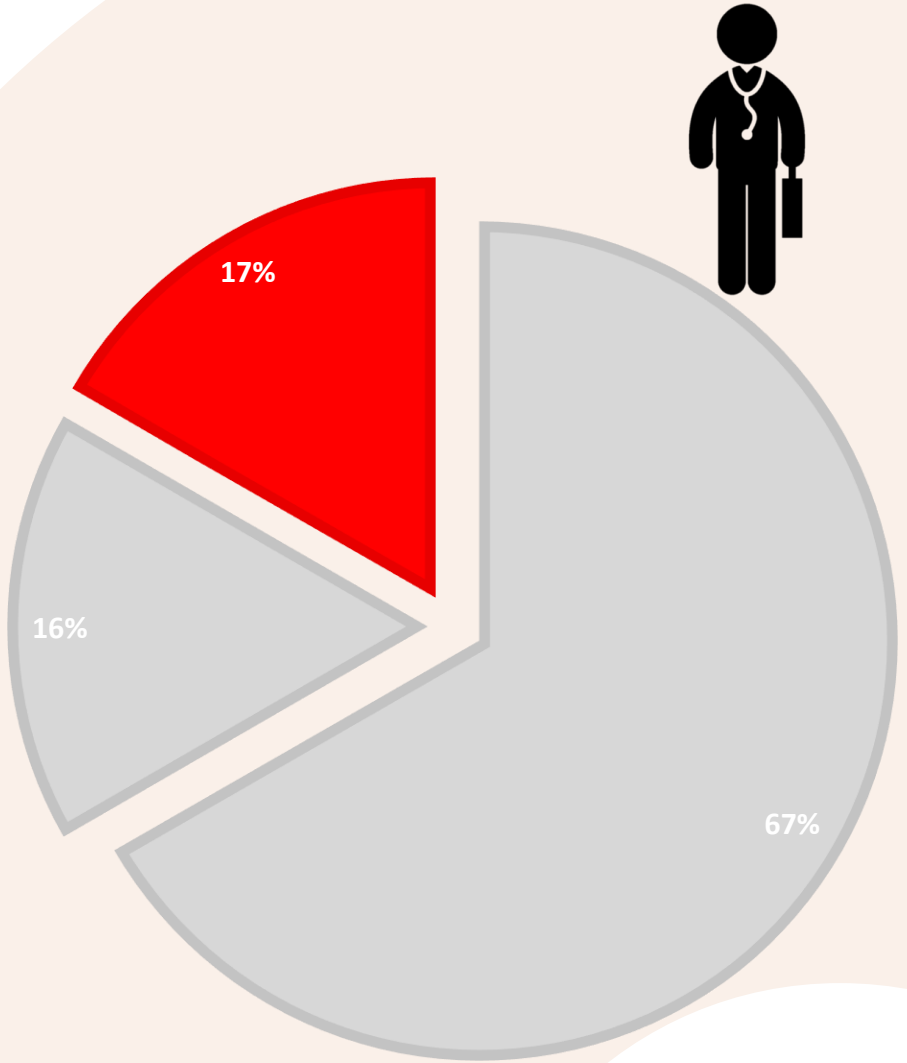
# Major birth defects

In 3 cases (17%) participant reported no birth defect

| Participant | GP | |
|---|---|---|
| | EUROCAT | |
| | **Major** | Unilateral stable right pyelum dilatation |
| | **Major** | Midseptal ventricular septal defect |
| | **Major** | Congenital syndactyly 3 toes left and right |

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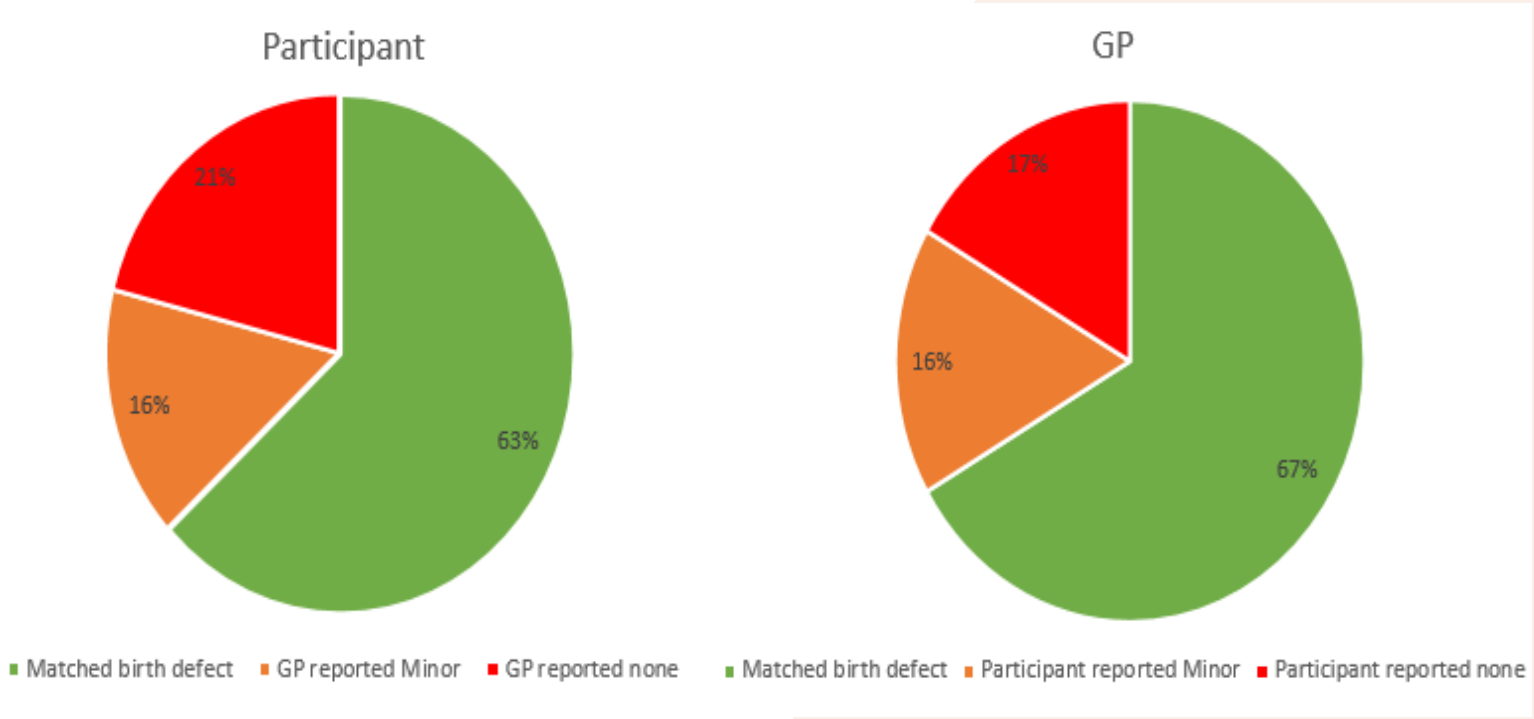
# Differences in reported Major birth defects

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# Conclusion

- Self-reported questionnaire data on infants' birth defects from mothers yields **similar information** compared to information obtained through GPs
- Mothers can adequately report the birth defects from their infant in detail since **3 out of 4** of their reported birth defects are ICD-10 coded identically with their GPs
- Some birth defects remain unreported by both the participants (N = 14) and GPs (N = 28), particularly **minor** birth defects.
- **≈35%** of reported major birth defects by either GP or participant remain **unreported or were classified as minor.**
- Almost all **infants (98.2%)** with at least one major or no birth defect were classified the same by the participant and GP.

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# Thank you for your attention!

Questions/remarks?
Contact me at v.maas@lareb.nl