ORIGINAL ARTICLE



Congenital anomalies in breech presentation: A nationwide record linkage study

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Abstract

Our study aimed to determine if congenital anomalies are associated with breech presentation at delivery. We conducted a nationwide, retrospective population-based record linkage study and analyzed all singleton births in Finland from 1996 to 2016 using the mandatory health register data collected by the Finnish Institute for Health and Welfare. We compared all major congenital anomalies detected during pregnancy, birth, or the first year of life according to the fetus's presentation at the time of delivery using X^2 -square statistic and Student's t test. We adjusted the results for known risk factors for congenital anomalies to estimate adjusted odds ratios and 95% confidence intervals. Fetuses in breech presentation at delivery had an increased risk for congenital anomalies (6.5%) compared with fetuses in cephalic presentation (3.6%), P < .001. Breech presentation was associated with nearly all types of examined congenital anomalies. The strongest associations were observed with congenital deformities of the hip, the central nervous system, the respiratory system, and the musculoskeletal system. Our study supports the theory that breech presentation is, in many cases, a symptom of a fundamental problem in fetal morphogenesis or function. Neonates born in the breech presentation have a higher risk of congenital anomalies and should undergo a postnatal screening.

KEYWORDS

birth defect, breech presentation, chromosomal anomaly, congenital anomaly, risk factor

INTRODUCTION 1

A congenital anomaly is a structural or functional defect detected during pregnancy, birth, or later in life.¹ Congenital anomalies are associated with stillbirth, neonatal and infant mortality, and morbidity. Almost half of all congenital anomalies cannot be linked to a specific cause.¹ Wellknown risk factors for congenital anomalies are genetic, socioeconomic, demographic, environmental factors, infections, and maternal nutritional status. They include ethnicity, increased maternal age, maternal diseases such as diabetes, family history of congenital anomalies, viral and parasitic infections during pregnancy, medications, alcohol or drug abuse, smoking, radiation, and chemical agents' exposure in pregnancy.¹⁻⁶ Globally, ~2.4% to 4.4% of all children are born with a severe congenital anomaly; 90% of congenital anomalies occur in low and middle-income countries.^{1,4-6} These anomalies can be primary abnormalities such as defects in an organ's structure or secondary abnormalities due to an

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Abbreviations: aOR, adjusted odds ratio; BMI, body mass index; CI, confidence interval; ICD, international classification of diseases system; OR, Odds ratio; RCM, National Medical Birth Register MBR and the Register on Congenital Malformations.

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interruption of an organ's normal development, caused by outer influences like teratogenic agents or a trauma. In the international classification of diseases system (ICD), congenital anomalies are classified according to organ systems or genetic defects. Preventive public health measures work to decrease the frequency of specific congenital anomalies by removing environmental risk factors or reinforcing protective factors such as preconceptionally folic acid use. Congenital anomalies can be detected through prenatal and neonatal screening.¹

Breech presentation of the fetus appears in around 2% to 4% of all births.⁷ At delivery, breech presentation is a known marker for adverse neonatal outcomes. Previous studies have shown an association between breech presentation at delivery and congenital anomalies.⁷⁻¹¹ However, these studies are limited by study size and the lack of adjustments for common risk factors of congenital anomalies. First, we hypothesize that congenital anomalies are associated with breech presentation at delivery. Second, we aimed to investigate the types of anomalies associated with breech presentation and increase awareness of the need for prenatal and neonatal screening among the children born in the breech to improve these children's treatment and care.

2 | METHODS

2.1 | Study design and data sources

We conducted a population-based record linkage study using anonymized data of mothers and infants recorded on the National Medical Birth Register (MBR) and the Register on Congenital Malformations (RCM). The registers are maintained by the Finnish Institute for Health and Welfare. All maternity hospitals are obliged to report to the MBR and the RCM. In Finland, all live-born infants receive unique personal identification numbers; these numbers can be used to trace all healthcare data and diagnoses in the case of hospitalization or death. Stillbirths can be identified by using the mother's identification number. A pediatrician routinely examines all newborns.

The MBR collects baseline data on pregnancies, deliveries, and newborn outcomes during the first days of life. These data include all live births and stillbirths with a birth weight of at least 500 g or a gestational age of 22 weeks or beyond. The RCM was established in 1963. The Register's primary purpose is to continuously monitor the prevalence and types of congenital anomalies for the early identification of any new environmental factors (teratogens) that potentially cause fetal defects and for the prevention of congenital anomalies by influencing these factors. Statistical data are used for monitoring congenital anomalies nationally and regionally, for planning prenatal screening and diagnostics of fetal anomalies, as well as for treatment of congenital anomalies, and for researching congenital anomalies. The RCM receives data on congenital anomalies from hospitals, health care professionals, and genetic laboratories. It also draws data from the MBR, the Care Register for Health Care, the Register on Induced Abortions, all maintained by the Finnish Institute for Health and Welfare, and from the Register of Visual Impairment, the data provided by the National Supervisory Authority for Welfare and Health, as well as from the Cause of Death Statistics, maintained by Statistics Finland. The data coverage and quality are regarded as very good.¹²

Authorization to use the data were obtained from the Finnish Institute for Health and Welfare as required by the national data protection law in Finland (Reference number THL/652/ 5.05.00/2017).

We analyzed all singleton live births in Finland from 1996 to 2016 using the Finnish Institute for Health and Welfare data. We compared all congenital anomalies detected at birth or during the first year of life according to the fetus's presentation at the time of delivery. Our study population included women with a singleton fetus either in breech or cephalic presentation at the delivery time. All other birth presentations were excluded. The classification for congenital anomalies was selected according to the ICD-10 classification (Table 1). ICD-9 codes (Atlanta modification for congenital anomalies) were matched to the ICD-10 codes. The selection of maternal variables and risk factors for congenital anomalies was based on the previous literature (Table 2).

The congenital anomalies were adjusted for all significant variables and risk factors. The calculations were performed using SAS 9.4. Statistical differences in categorical variables were evaluated with the Chi-squared test or Fisher's exact test when appropriate. We calculated odds ratios (ORs) with corresponding 95% confidence intervals using binary logistic regression. Differences were considered statistically significant, with a *P*-value of \leq .05.

3 | RESULTS

We analyzed the data on 1 184 499 women with a singleton delivery from 1996 to 2016. Women with a breech delivery numbered 34 030 (2.9%).

Among the neonates born in breech presentation, there were 2209 (6.5%) neonates presenting with at least one congenital anomaly,

TABLE 1 Accessed congenital anomalies

ICD 10 codes	Anomalies
Q00-Q07	Congenital malformations of the nervous system
Q10-Q18	Congenital malformations of eye, ear, face and neck
Q20-Q28	Congenital malformations of the circulatory system
Q30-Q34	Congenital malformations of the respiratory system
Q35-Q37	Cleft lip and cleft palate
Q38-Q45	Other congenital malformations of the digestive system
Q50-Q56	Congenital malformations of genital organs
Q60-64	Congenital malformations of the urinary system
Q65	Congenital deformities of hip
Q66-Q79	Congenital malformations and deformations of the musculoskeletal system
Q80-Q89	Other congenital malformations
Q90-Q99	Chromosomal abnormalities. Not elsewhere classified

TABLE 2 Maternal characteristics and cofounders

	Breech 34 03	30	Cephalic 1 15	0 469	OR 95% CI
	Ν	%	Ν	%	
Maternal age < 25	721	2.1	28 852	2.5	0.84 (0.78-0.91)
Maternal age ≥ 35	6692	19.7	214 713	18.7	1.07 (1.04-1.10)
Smoking	5250	15.4	171 310	14.9	1.04 (1.01-1.07)
Maternal BMI ≥35	737	2.2	27 413	2.4	0.91 (0.84-0.98)
Nullipara	20 195	59.3	469 106	40.8	2.12 (2.07-2.17)
Multipara ≥3	2018	5.9	115 076	10.0	0.57 (0.54-0.59)
Maternal hypothyroidism E03.9	166	0.5	3452	0.3	1.63 (1.39-1.90)
Maternal hyperthyroidism E05.9	32	0.1	85	0.0	12.74 (8.48-19.13)
Gestational diabetes O24.4	1690	5.0	60 447	5.3	0.94 (0.90-0.99)
Diabetes mellitus type 1 and 2 O24.0/O24.1	5	0.0	88	0.0	1.92 (0.78-4.73)
Maternal care for fetal injury from alcohol O35.4	16	0.05	572	0.05	0.95 (0.58-1.55)
Maternal care for damage to fetus by drugs O35.5	25	0.07	1293	0.11	0.65 (0.44-0.97)
Maternal care for damage to fetus by radiation O35.6	15	0.04	183	0.02	2.77 (1.64-4.69)
Supervision of high-risk pregnancy due to social problems Z35.7	41	0.12	1795	0.16	0.77 (0.57-1.05)
Viral diseases complicating pregnancy O98.5	16	0.05	562	0.05	0.96 (0.59-1.58)
Protozoal diseases complicating pregnancy O98.6	0	0.00	20	0.00	
Previous congenital anomaly	1013	2.98	31 009	2.70	1.11 (1.04-1.18)
Neonatal female gender	18 219	53.5	560 297	48.7	1.21 (1.19-1.24)
Gestational age 23 + 0-36 + 6	4907	14.4	49 486	4.3	3.75 (3.63-3.87)
Gestational age 37 + 0-39 + 6	21 779	64.0	481 845	41.9	2.47 (2.41-2.52)
Gestational age 40 + 0 -	7253	21.3	615 430	53.5	0.24 (0.23-0.24)

TABLE 3 Rate of congenital anomalies according to presentation at birth

	Breech 34 030	1/10000	Cephalic 1150 469	1/10000	Р	OR 95% CI	Adjusted OR 95% Cl
Any major congenital anomaly	2209	649.1	42 089	365.8	<.001	1.83 (1.75-1.91)	1.40 (1.33-1.46)
Isolated	1438	422.6	32 945	286.4	<.001	1.50 (1.42-1.58)	1.23 (1.16-1.30)
Two major congenital anomalies	101	29.7	1295	11.3	<.001	2.64 (2.16-3.24)	1.77 (1.44-2.19)
≥ 2 major congenital anomalies	274	80.5	3011	26.2	<.001	3.09 (2.73-3.50)	1.74 (1.52-1.98)
Syndrome	396	116.4	4831	42.0	<.001	2.79 (2.52-3.10)	1.65 (1.47-1.84)
Other	0	0.0	7	0.1			

Note: Adjusted for: Maternal age < 25, maternal age \geq 35, smoking, maternal BMI \geq 35, nullipara, multipara \geq 3, maternal hypothyroidism, maternal hyperthyroidism, gestational diabetes, maternal care for damage to fetus by alcohol, maternal care for damage to fetus by radiation, previous congenital anomaly, neonatal sex, and gestational age.

whereas, among the neonates in cephalic presentation, such anomalies occurred in 42 089 (3.6%) cases. The rate of neonates with at least one congenital anomaly per 10 000 births was 649/10 000 in breech deliveries and 366/10 000 in cephalic deliveries (adjusted odds ratio [aOR] 1.40, 95% confidence interval [CI] [1.33-1.46], P < .001).

An isolated finding appeared in 1438 (65.1%) of the breech-born neonates, as compared with 32 945 (78.3%) neonates born in cephalic presentation (aOR 1.23, 95% CI [1.16-1.30], P < .001). Additionally,

children born in the breech presentation had more often multiple congenital anomalies; 101 (0.3%) of these neonates had at least two major congenital anomalies, as compared with 1295 (0.11%) children born in cephalic presentation (aOR 1.77, 95% CI [1.44-2.19], P < .001). In the breech delivery group were 274 (0.81%) neonates with at least three major congenital anomalies, while the cephalic delivery group comprised 3011 (0.26%) (aOR 1.74, 95% CI [1.52-1.98], P < .001). Of all breech-born children 396 (1.16%) had a **TABLE 4** Investigated congenital anomalies according to presentation at birth

	Breech 34 030		Cephalic 1 150 469		Р	OR 95% CI	Adjusted OR 95% CI
	Ν	1/10000	Ν	1/10000			
Congenital malformations of the nervous system	191	56.1	1948	16.9	<.001	3.33 (2.87-3.86)	1.89 (1.61-2.22)
Congenital malformations of eye, ear, face, and neck	124	36.4	2707	23.5	<.001	1.55 (1.29-1.86)	1.22 (1.02-1.48)
Congenital malformations of the circulatory system	818	240.4	17 935	155.9	<.001	1.56 (1.45-1.67)	1.14 (1.05-1.22)
Congenital malformations of the respiratory system	152	44.7	1091	9.5	<.001	4.73 (3.99-5.60)	2.08 (1.72-2.50)
Cleft lip and cleft palate	134	39.4	2499	21.7	<.001	1.82 (1.53-2.16)	1.45 (1.22-1.74)
Other congenital malformations of the digestive system	144	42.3	2550	22.2	<.001	1.91 (1.62-2.26)	1.01 (0.85-1.21)
Congenital malformations of genital organs	153	45.0	2535	22.0	<.001	2.05 (1.74-2.41)	1.37 (1.16-1.63)
Congenital malformations of the urinary system	251	73.8	4306	37.4	<.001	1.98 (1.74-2.25)	1.40 (1.23-1.60)
Congenital deformities of the hip	157	46.1	623	5.4	<.001	8.55 (7.18-10.19)	7.50 (6.24-9.02)
Congenital malformations and deformations of the musculoskeletal system	648	190.4	8874	77.1	<.001	2.50 (2.30-2.71)	1.88 (1.72-2.04)
Other congenital malformations	237	69.6	3441	29.9	<.001	2.34 (2.05-2.67)	1.52 (1.33-1.75)
Chromosomal abnormalities not elsewhere classified	190	55.8	2401	20.9	<.001	2.68 (2.32-3.11)	1.40 (1.20-1.65)

Note: Adjusted for: Maternal age < 25, maternal age \geq 35, smoking, maternal BMI \geq 35, nullipara, multipara \geq 3, maternal hypothyroidism, maternal hyperthyroidism, gestational diabetes, maternal care for damage to fetus by alcohol, maternal care for damage to fetus by radiation, previous congenital anomaly, neonatal sex, and gestational age.

genetic syndrome, compared with 4831 (0.42%) children born in cephalic presentation (aOR 1.65, 95% CI (1.47-1.84), P < .001]. (Table 3).

The incidence of nearly all congenital malformation types evaluated in this study was higher among children born breech than among children born in cephalic presentation (Table 4).

The rate of congenital deformities of the hip was exceptionally high in the breech group, occurring in 157 (0.46%) neonates compared with 623 (0.05%) cases in cephalic presentation. The children born in breech presentation also had more often congenital malformations and deformations of the musculoskeletal system; in 648 (1.9%) neonates vs 8874 (0.77%) neonates in cephalic presentation. Additionally, significantly higher rates of congenital malformations of the central nervous system, the circulatory and respiratory systems, genital organs, and the urinary system appeared among breech deliveries (Table 4). Furthermore, malformations of the eye, ear, face, and neck, such as cleft lip and cleft palate, were more frequent among breech-born neonates than among cephalic-born neonates (Table 4). There were some differences in the maternal characteristics and the exposures to other possible risk factors for congenital anomalies between the study groups. The study groups differed in the following variables: maternal age, smoking, maternal BMI, maternal hypothyroidism, maternal hyperthyroidism, gestational diabetes, maternal care for fetal injury by alcohol, maternal care for fetal injury by radiation, previous congenital anomaly, neonatal sex, and gestational age. (Table 2).

4 | DISCUSSION

Our study demonstrates a significant correlation between breech presentation at delivery and congenital anomalies. Breech presentation at delivery is associated with most of the congenital anomalies examined. Our study supports the theory that breech presentation is, in many cases, a symptom of a fundamental problem in fetal morphogenesis or function.^{7,8,13} We confirmed and built on existing evidence from earlier smaller studies in which breech presentation at delivery

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has been demonstrated as a marker for congenital anomalies.^{8,10,11,14} However, our results were based on a large sample size.

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Moreover, we were able to adjust our results for common confounders for congenital anomalies. Our study results provide a new insight into the relationship between congenital anomalies and breech presentation at delivery. We were also able to estimate the increase in risk for the various congenital anomalies in breech presentation. The analysis indicates the importance of prenatal and neonatal anomaly screening of children born in breech presentation, as such presentation may be a sign of congenital anomalies.

The prevalence of congenital anomalies in breech (6.5%) was twofold compared with the cephalic presentation (3.7%). Our rates are lower than the rates found by Mostello.⁸ but differences in the healthcare systems might explain these variations. Since 2010, every woman in Finland can participate in a nuchal translucency screening scan during gestational weeks 11 to 13 and a second-trimester anomaly scan during gestational weeks 19 to 21, free of charge. The nuchal translucency scan is used to detect chromosomal abnormalities, and the second-trimester scan is primarily used to assess fetal anatomy and detect the presence of any fetal anomalies. About 70% of all Finnish women participate in the scans. If one of the ultrasound examinations reveals some abnormality, the parents will have the opportunity to meet a specialist physician for further examinations and discussions about the significance of the findings and further examination at a fetomaternal medical center. The family will then be allowed to prepare for an afflicted child's birth or decide on the termination of the pregnancy (depending on the severity of the disorder), permitted under Finnish law until the 24th week of pregnancy.¹⁵

Breech presentation is a known risk factor for dysplasia of the hip.¹⁶ We also expected in our data this association. The children born in breech presentation had a significantly increased risk of having a congenital hip deformity. This is most likely one of the rare congenital anomalies caused by breech presentation. Breech presentation is one of the well-known causes for the hip's developmental dysplasia, as the fetal breech position can limit the fetal movements in the womb, particularly when the fetus's knees are extended (frank breech).^{16,17} In this case, breech presentation is most likely the cause of a congenital anomaly and not a symptom. Fortunately, this association is recognized, and children born in breech presentations are in many countries routinely screened during the neonatal period for developmental dysplasia of the hip, and therefore early detected and treated.

Our data show that breech presentation at delivery increased the risk of the nervous system's congenital malformations and a higher risk for congenital malformations and deformations of the musculo-skeletal system. These results are in line with the hypothesis that some fetuses in breech presentation lack movements or have the reduced ability to turn them into cephalic presentation. This reduced ability might be caused by neurological and musculoskeletal malformations.^{18,19}

We found that breech presentation at delivery was associated with various congenital malformations of different organ systems. After adjusting, the only congenital malformation that was not associated with breech presentation was the digestive system's malformations. Our results confirm the previous findings from Mostello's work.⁸ Some congenital anomalies are most likely associated with breech presentation as they impeach a fetal rotation into a cephalic presentation as the fetal head might not fit in the maternal pelvis through malformations of the fetal eye, ear, face, or neck. Many congenital malformations such as malformations of the respiratory system, the circulatory system, and cleft lip or cleft palate, are associated with polyhydramnios,^{20,21} which is associated with abnormal fetal presentation.²¹ Malformations of the genital organs and the urinary system might be associated with polyhydramnios^{20,21} and oligohydramnios, both known risk factors for breech presentation.^{9,21} Chromosomal abnormalities are often linked to intrauterine growth restriction, which is significantly associated with breech presentation.⁹

This study is an extensive population-based record-linkage study on breech presentation and the risk of congenital malformations. Our research presents valuable information for evaluating newborns in the nursery. The study population includes 1 184 499 deliveries from Finland. We could link the neonatal outcome to maternal data and adjust our outcomes accordingly for known cofounders. A further strength relates to the critical information on the mother's characteristics and risk factors for congenital anomalies. There are, however, some limitations in our study. The study's retrospective character constrained the methodological choices. Another limitation is the design as a recordlinkage study, which restricted the variables to data availability. As an example, we could not see which women have participated in the nuchal translucency and anatomy scan during pregnancy. An additional limitation is that we do not know which pregnancies underwent an external cephalic version, and the question remains whether the children who have been turned into cephalic presentation have a higher risk of congenital anomalies. In addition, the connection of congenital anomalies with preterm breech deliveries remains unclear and would need further research and investigations in the future.

According to our study, breech presentation at birth may be a marker of congenital anomalies. Thus, we recommend careful postnatal screening for all children born in breech presentation.

CONFLICT OF INTERESTS

The authors state explicitly that there are no conflicts of interest connected with this article. All authors declare independence from any funding agency for this work.

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