Algorithms to identify major congenital malformations in routinely collected healthcare data: A systematic review

Drug Safety

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Supplemental Table 5. Algorithms for MCM among studies using linked databases of more than one type, 2010-2025

Author, Year	Country	Data source name	Data source types	Case ascertainment denominator	Timing of MCM assessment	Algorithm rules	Coding system	Code location in manuscript	Prevalence of MCM in unexposed or general population
Putignano 2019 [1]	Italy	Administrative health databases of Italy's Lombardy Region	Hospital discharge data and vital records	Live births	At birth or within first year of life	Recorded on birth certificates and/or ≥1 code on the infant's hospitalization record	ICD-9-CM	Methods	5.1%
Cohen, 2024 [2]	Canada	Canadian Institute for Health Information's Discharge Abstract Database (CIHI-DAD) and OHIP	Claims and hospital discharge data	Live and stillbirths	At birth	≥1 code	ICD-10-CA	eTable 1	1.0%
Ray, 2023 [3]	Canada	Canadian Institute for Health Information's Discharge Abstract Database (CIHI-DAD) and OHIP	Claims and hospital discharge data	Live and stillbirths	At birth	≥1 code in DAD; ≥1 code in addition to diagnosis billed by pediatrician in OHIP	ICD-10-CA and ICD-9	Table S2	No unexposed group

Ahmed, 2022 [4]	Canada	Canadian Institute for Health Information's Discharge Abstract Database (CIHI-DAD) and OHIP	Claims and hospital discharge data	Live births	At birth or within first six years of life	≥1 inpatient or outpatient diagnosis in the infant claims or hospitalization discharge records	ICD-10-CA for inpatient; ICD-9 for outpatient	Supplemental Material, Table S2	11.0%
Davidson 2020 [5]	Canada	Canadian Institute for Health Information's Discharge Abstract Database (CIHI-DAD) and OHIP	Claims and hospital discharge data	Live and stillbirths	At birth or within first year of life	≥1 inpatient or outpatient diagnosis in the infant claims or hospitalization discharge records	ICD-10-CA for inpatient; ICD-9 for outpatient	Supplemental Material (eTable 1)	Not provided, no unexposed population available
Ray 2016 [6]	Canada	Canadian Institute for Health Information's Discharge Abstract Database (CIHI-DAD) and OHIP	Claims and hospital discharge data	Live births	At birth or within first four years of life	≥1 inpatient diagnosis or ≥1 outpatient diagnosis in infant record by a pediatrician (Specialty Code 26)	ICD-10-CA for inpatient; ICD-9 for outpatient	Supplemental Material (eTable 1)	7.7%
Feig 2014 [7]	Canada	Canadian Institute for Health Information's Discharge Abstract Database (CIHI-DAD) and OHIP	Claims and hospital discharge data	Live births	At birth or within first year of life	≥1 code in infant record	ICD-9 and ICD-10	Methods and Appendix	2.9%
Riley 2023 [8]	UK	CPRD and HES	EMR and hospital discharge data	Live births	Birth through first 3 months of life	≥1 code in infant record	READ and ICD10	Supplemental Table 2	2.7%-3.0%
Peppa 2020 [9]	UK	CPRD, HES, and ONS	EMR, hospital discharge data, and vital records	Live births	At birth or within first year of life	≥1 code in infant record	ICD-10 (hospital), OPCS-4 (hospital), and Read (primary care) codes	Supplemental Material	7.3%
Ferrer 2025 [10]	France	EFEMERIS	Claims, EMR, hospital discharge data, and vital records	Live births, fetal deaths, and medical terminations of pregnancy	Pregnancy and at birth	≥1 code	ICD-10	Methods	2.4%
Benevent 2022 [11]	France	EFEMERIS	Claims, EMR, hospital discharge data, and vital records	Live birth, stillbirths and TOPFAs	At birth or within first two years of life	≥1 code from health certificates filled out by pediatricians or general practitioners (during the compulsory medical examinations at birth, 9 and 24 months) or prenatal diagnosis specialists	ICD-10	Methods	2.1%
Araujo 2022 [12]	France	EFEMERIS	Claims, EMR, hospital discharge data, and vital records	Live birth, stillbirths and TOPFAs	At birth or within first two years of life	≥1 code from health certificates filled out by pediatricians or general practitioners (during the compulsory medical examinations at birth, 9 and 24 months) or prenatal diagnosis specialists	ICD-10	Methods	2.6%

Vabre 2021 [13]	France	EFEMERIS	Claims, EMR, and hospital discharge data	Live birth, stillbirths and TOPFAs	At birth or within first two years of life	≥1 code from health certificates filled out by pediatricians or general practitioners (during the compulsory medical examinations at birth, 9 and 24 months) or prenatal diagnosis specialists	ICD-10	Methods	2.4%
Araujo 2021 [14]	France	EFEMERIS	Claims, EMR, and hospital discharge data	Live birth, stillbirths and TOPFAs	At birth or within first two years of life	≥1 code from health certificates filled out by pediatricians or general practitioners (during the compulsory medical examinations at birth, 9 and 24 months) or prenatal diagnosis specialists	ICD-10	Methods, Table 4	2.5%
Kharbanda, 2020 [15]	USA	Integrated health system primarily in Minnesota	Claims and EHR	Live born infants	Birth through 1 year of life	Required infants have 2 or more outpatient diagnoses or that the defect was noted in the problem list	ICD-10-CM	Appendix 1	Not reported
Süüden, 2023 [16]	Estonia	Medical Birth Register, Abortion Registry, Health Insurance Fund and hospital records	National register, claims, and hospital discharge	Pregnancies ≥ 12 weeks gestation	Birth through 1 month of life	≥1 code	ICD-10	Methods, Supplemental Table 1	3.8%
Eltonsy 2017 [17]	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Live and stillbirths	At birth or within first year of life	Three algorithms were implemented across two classification methods: (1) ≥1 major malformation diagnosis recorded in the hospital database; (2) ≥2 major malformation diagnoses recorded in the medical claims database or ≥1 major malformation diagnosis recorded in the hospital database; (3) ≥1 major malformation diagnosis recorded in the medical claims database or ≥1 major malformation diagnosis recorded in the hospital database	ICD-9 and ICD-10	Supplemental Material (Table e1)	5.1-7.1% and 7.0-10.6 % across algorithms using the two classification methods
Zhao 2015 [18]	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Live births	At birth or within first year of life	≥2 diagnostic codes for the same category in infant records	ICD-9 and ICD-10	Methods	3.7%
Blais 2015 [19]	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Live and stillbirths	At birth or within first year of life	≥1 code in the infant record (if live born) or in the maternal record (if stillborn). Malformations that were initially undetermined with regard to major or minor were reclassified as major if there was ≥1 hospitalization with an admission or a primary diagnosis of the malformation recorded in the MED-ECHO data within first year of life.	ICD-9 and ICD-10	Methods	6.8%
Eltonsy 2015 [20]	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Singleton live births	At birth or within first year of life	≥1 hospitalization with a primary diagnosis or admission diagnosis related a malformation that was recorded in the MED-ECHO database	ICD-9 and ICD-10	Methods and Appendix Table 1	7.1%
Richard-Tremblay 2013 [21]	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Live births	At birth or within first year of life	≥2 codes (on two different occasions) for the same organ or system affected	ICD-9 and ICD-10-CA codes	Methods	3.7%
Blais 2013 [22]*	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Not specified, live births for validation study	At birth or within first year of life	To identify all malformations and select one per infant, ≥1 code in a single group. For those with ≥2 diagnostic codes for the same group of malformations, the code that was recorded during a hospitalization or at an outpatient clinic affiliated to a hospital at the time closest to infant's first birthday was preferentially selected. To identify infants without a congenital	ICD-9	Methods, Supplemental Material	Not calculated

						malformation, exactly 0 codes with requirement of hospitalization or outpatient clinic claims ≥2 times in the first year of life.			
Berard 2012 [23]	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Live births	At birth or within first year of life	≥1 code	ICD-9	Methods	7.9%
Kulaga 2011 [24]	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Live births	At birth or within first year of life	≥1 code	ICD-9	Methods	Not provided, no unexposed population available
Kulaga 2010 [25]*	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Singleton live births	At birth or within first year of life	≥1 code	ICD-9	Methods	Not calculated, no denominator
Blais 2010 [26]	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Live births or stillbirth	At birth or within first year of life	≥1 code	ICD-9	Methods and Appendix 1	4.4%
Nakhai-Pour 2010 [27]	Canada	RAMQ and MED-ECHO	Claims and hospital discharge data	Liveborn singletons	At birth or within first year of life	≥1 code	ICD-9	Methods	Not calculated, case-control study
Newton 2024 [28]	USA	The Surveillance for Emerging Threats to Pregnant People and Infants Network (SET-NET)	EMR and vital records	Births	Not specified	≥1 code	ICD-10-CM	Methods	Not provided
Kharbanda, 2021 [29]*	USA	Vaccine Safety Datalink	Claims and EHR	Live born infants	Birth through 1 year of life	Not reported	ICD-10-CM	Methods, Tables 1 and 3, Supplemental Table 1	1.8%
Kharbanda, 2017 [30]*	USA	Vaccine Safety Datalink	Claims and EHR	Live born infants	Birth through 1 year of life	Number of required diagnoses and setting varied by organ system	ICD-9-CM	Methods, Table 1, Table 3	4.4%

Abbreviations: EFEMERIS: Evaluation chez la Femme Enceinte des Médicaments et de leurs RISques; RAMQ: Régie de l'assurance maladie du Québec (administrative claims); MED-ECHO: Maintenance and Exploitation of Data for the Study of Hospital Clientele (hospital discharge data)
*Denotes a validation study, also described in Table 2.

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