How-To Guide: Birth Defects

Provided by CDC's Environmental Public Health Tracking Program

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PURPOSE AND USE OF THIS DOCUMENT

This document describes the steps for extracting and formatting the necessary data required for the Tracking Program's Nationally Consistent Data and Measures (NCDM) for birth defects.

HOW-TO GUIDE

	Description
Measures	Birth Defects
Data Source(s)	State-based birth defects registry
NCDM Data Requirements	See Data Dictionary
HTG Requirements and Cautions	For each year of data you are submitting, identify which scenario below applies to you and complete the appropriate metadata and data steps: ○ **Scenario 1:** If for the year you are submitting data, ALL counties in your state collected and reported data for ALL NCDM birth defects, then ▪ Complete standard metadata requirements and answer "Yes" to the question, "Were birth defects data collected and reported in every county within the state for all 12 birth defects?" ▪ Include all rows* of data where total live birth is > 0, even if the number of live births with the specified defect = 0. ○ **Scenario 2:** If for the year you are submitting data, ALL counties in your state collected and reported data for SOME NCDM birth defects, then ▪ Indicate in your metadata which defects were not collected by your state that year and answer "No, all counties collected and reported but not for all birth defects" to the question, "Were birth defects data collected and reported in every county within the state for all 12 birth defects?" ▪ For the defects you did collect data, include all rows* of data where total live birth is > 0, even if the number of live births with the specified defect = 0. Do not include any rows of data for defects not collected.

	Description
	○ **Scenario 3:** If for the year you are submitting data, SOME counties in your state collected and reported data for ALL NCDM birth defects, then ▪ Indicate in your metadata which defects were not collected by your state that year and answer "No, all birth defects were collected and report but not in every county" to the question, "Were birth defects data collected and reported in every county within the state for all 12 birth defects?" ▪ For each county where a defect was collected, include all rows* of data where total live birth is > 0, even if the number of cases with the specified defect = 0. Do not include any rows of data for counties and defects where data were not collected. ○ **Scenario 4:** If for the year you are submitting data, SOME counties in your state collected and reported data for SOME NCDM birth defects (i.e., one county did not collect data for one birth defect or a few counties did not collect data for a few defects, etc.) ▪ Indicate in your metadata which defects were not collected by which counties that year and answer "No, data were not collected and reported for all 12 birth defects and those that were collected were not collected in every county" to the question, "Were birth defects data collected and reported in every county within the state for all 12 birth defects?" ▪ For each county where a defect was collected, include all rows* of data where total live birth is > 0, even if the number of live births with the specified defect = 0. Do not include any rows of data for counties and defects where data were not collected. Clarifications: • By submitting all rows of data where total live birth is >0 (including those combinations where number of live births with that birth defect = 0 or number of live births, fetal deaths, and pregnancy terminations with that birth defect = 0), you will provide the correct denominator for calculating prevalence. The sum of total live births for all rows in a specified county for a specified defect or for a specified year should equal the total live births for that county in that year. • If you have defects within an unknown county, include rows using 'U' as the county FIPS code. Set variable for total live births to -999. • If you have live births within an unknown county, include rows using 'U' as the county FIPS code. Repeat these rows for each defect. This will ensure that the sum of total live births for all rows for a specified defect for a specified year should equal the total live births for your state that year. Set variables representing the number of cases to -999. • Refer to steps 5 and 6 below for further information on extracting and formatting these variables. *A row of data is a unique combination of County, Startdate, Enddate, BirthDefect, MaternalAgeGroup, MaternalRace, and MaternalEthnicity, Infantsex.

	Description

Additional metadata requirements	In the metadata record, please answer the following questions and provide any additional information:
	• What was the surveillance method used to ascertain birth defect cases?
	  ○ Active
	  ○ Passive with follow-up
	  ○ Passive
	  ○ Other
	• Were maternal race and ethnicity collected and reported separately by the primary data steward?
	  ○ Yes
	  ○ No
	  ○ Race and ethnicity not collected
	  ○ Other
	• What medical coding standard was used to classify birth defects?
	  ○ ICD-9 CM
	  ○ ICD-9, CDC coding based on British Pediatric Association
	  ○ ICD-10 CM
	• Were birth defects data collected and reported in every county within the state for all 12 birth defects? (See scenarios above for guidance on choosing the correct answer)
	  ○ Yes
	  ○ No, all counties collected and reported but not for all birth defects
	  ○ No, all birth defects were collected and reported but not in every county
	  ○ No, data were not collected and reported for all 12 birth defects and those that were collected were not collected in every county
	• What pregnancy outcomes were included in ascertaining cases?
	  ○ Live births only
	  ○ Live births and fetal deaths
	  ○ Live births and pregnancy terminations
	  ○ Live births, fetal deaths, and pregnancy terminations

Step #1	Identify cases of selected birth defects among babies born in the year (or years) of data for submission. See Appendix A for listing of birth defects, relevant ICD9-CM, CDC/BPA, and ICD10-CM codes. • Anencephaly • Cleft lip with cleft palate • Cleft lip w/o cleft palate • Cleft palate w/o cleft lip • Gastroschisis • Hypoplastic left heart syndrome • Hypospadias • Limb deficiencies combined • Spina bifida (w/o anencephaly) • Tetralogy of Fallot • Transposition of the great arteries (vessels) • Trisomy 21

<table>
<tr><th></th><th>Description</th></tr>
<tr><td>Step #2</td><td>Retain the following elements:
Maternal county of residence
Year of birth
Birth defect category
Maternal age group
Maternal state of residence
Maternal ethnicity
Maternal race
Infant sex</td></tr>
<tr><td>Step #3</td><td>Use the data elements retained to create the following variables as described in the data dictionary:
County
StartDate
EndDate
BirthDefect
MaternalAgeGroup
MaternalEthnicity
MaternalRace
InfantSex</td></tr>
<tr><td>Step #4a</td><td>StartDate and Enddate will most often be YYYY0101 and YYYY1231 as these represent the start and end date of year YYYY over which you are aggregating cases of birth defects. If a birth defect (either in a county or across the state) was not collected the entire year, then your StartDate variable should reflect the day collection for that defect began that year or EndDate variable should reflect the day collection stopped that year.</td></tr>
<tr><td>Step #4b</td><td>Maternal Ethnicity and Race can be reported in two ways depending on how your system collects this information (Appendix B). Please see the data dictionary for explanations of the abbreviations used below.

1 – Ethnicity and Race specified
<table>
<tr><th>MaternalEthnicity</th><th>MaternalRace</th></tr>
<tr><td>H</td><td>W, B, O, U</td></tr>
<tr><td>NH</td><td>W, B, O, U</td></tr>
<tr><td>U</td><td>W, B, O, U</td></tr>
</table>
2 - Race not specified when ethnicity is Hispanic
<table>
<tr><th>MaternalEthnicity</th><th>MaternalRace</th></tr>
<tr><td>H</td><td>NS*</td></tr>
<tr><td>NH</td><td>W, B, O, U</td></tr>
<tr><td>U</td><td>W, B, O, U</td></tr>
</table></td></tr>
</table>

	Description
	*NS = not specified. Invalid entry: Ethnicity Unknown (U) and Race Not Specified (NS) Ethnicity Not Hispanic (NH) and Race Not Specified (NS)
Step #5	Create the denominator variable 'TLB' as the total number of live births which correspond to your numerator in step 5, i.e., summarized over the same combinations of infant sex, maternal age group, maternal ethnicity, maternal race, start date, end date, and county or state. Refer to the "Clarifications" section under "How-To Guide Requirements and Cautions" for further information.
Step #6	Create variable 'LBWBD' for the number of live births with the specified birth defects. Aggregate the dataset by summarizing the number of live births with each birth defect by the variables described in the data dictionary for the dataset you are creating. Refer to the "Clarifications" section under "How-To Guide Requirements and Cautions" for further information.
Step #6a	Optional: Create the variable 'LBFDTWD' for number of live births, fetal deaths, or pregnancy terminations with the specified birth defects. This is an optional variable. Only include this variable if your state collects these birth outcomes. Refer to the "Clarifications" section under "How-To Guide Requirements and Cautions" for further information.
Step #7	Order the variables according to the schema: • County • StartDate • EndDate • BirthDefect • MaternalAgeGroup • MaternalEthnicity • MaternalRace • InfantSex • TLB • LBWBD • LBFDTWD
Step #8	Create a variable called "RowIdentifier". RowIdentifier should be a sequence of numbers from one to the number of rows in your dataset.
Step #9	Convert file to XML according to the corresponding schema.

APPENDIX A – Birth Defect Case Definitions

Additional information and guidance for these birth defects can be found in Appendix 3.1 of the National Birth Defects Prevention Network's "Guidelines for Conducting Birth Defects Surveillance (updated March 2021)".

https://www.nbdpn.org/docs/Appendix_3_1_BirthDefectsDescriptions_2021MAR12_Rev.pdf

	Birth Defect	ICD-9-CM	CDC/BPA	ICD-10-CM
21	Anencephaly	740.0-740.1	740.00-740.10	Q00.0 - Q00.1
22	Cleft Lip with Cleft Palate	749.20 - 749.25	749.20-749.29	Q37.0-Q37.9
23	Cleft Lip without Cleft Palate	749.1	749.10-749.19	Q36.0-Q36.9
24	Cleft Palate without Cleft Lip	749.0	749.00-749.09	Q35.1 – Q35.9
25	Gastroschisis (prior to 10/01/2009)	756.79 (This is a shared code with omphalocele. If your state uses ICD-9 codes, please indicate in your metadata if you have another method to distinguish these two conditions. If not, you do not have to report the data.)	756.71	NA
	Gastroschisis (starting on 10/01/2009)	756.73	756.71	Q79.3
26	Hypoplastic Left Heart Syndrome	746.7	746.70	Q23.4
27	Hypospadias	752.61	752.60 – 752.62 (excluding 752.61 and 752.621)	Q54.0 – Q54.9 (excluding Q54.4)
28	Limb Deficiencies	755.2 – 755.4	755.20 – 755.49	Q71.0 – Q71.9, Q72.0 – Q72.9, Q73.0 – Q73.8
29	Spina Bifida (w/out anencephaly)	741.0, 741.9 without 740.0-740.1	741.00-741.99 without 740.00-740.10	Q05.0 - Q05.9, Q07.01, Q07.03 without Q00.0 - Q00.1
30	Tetralogy of Fallot	745.2	745.20 – 745.21, 747.31	Q21.3
31	Transposition of the Great Arteries	745.10, 745.12, 745.19	745.10 – 745.12, 745.18 – 745.19	Q20.3, Q20.5
32	Trisomy 21	758.0	758.00-758.09	Q90.0 – Q90.9

APPENDIX B – Maternal Ethnicity and Race can be reported in two ways depending on how your system collects this information

Option 1:

Race	Ethnicity	Portal Display: Maternal Race Ethnicity
W	H	Hispanic
B	H	Hispanic
O	H	Hispanic
U	H	Hispanic
W	NH	White, Non-Hispanic
B	NH	Black, Non-Hispanic
O	NH	Other, Non-Hispanic
U	NH	(used only in unstratified calculations)
W	U	(used only in unstratified calculations)
B	U	(used only in unstratified calculations)
O	U	(used only in unstratified calculations)
U	U	(used only in unstratified calculations)

Option 2: The code 'NS' is intended to allow the maternal race field to be selectively collapsed, for example,

Race	Ethnicity	Portal Display: Maternal Race Ethnicity
W	NH	White, Non-Hispanic
B	NH	Black, Non-Hispanic
O	NH	Other, Non-Hispanic
U	NH	(used only in unstratified calculations)
NS*	H	Hispanic
W	U	(used only in unstratified calculations)
B	U	(used only in unstratified calculations)
O	U	(used only in unstratified calculations)
U	U	(used only in unstratified calculations)

*NS = not specified.

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