

# Screening Newborns for Disease





# Index

- The different color titles represent the different sections in this guide:
  - General information
  - Causes of Birth Defects
  - Screening for Defects
  - Prevention of Defects

# Birth Defects and Disease

## Birth defects are common



Every 4½ minutes, a baby is born with a birth defect in the United States.

Birth defects affect

**1** in every **33**

babies born in the United States each year.

That translates into about

**120,000** babies.



- Estimates from the World Health Organization (WHO) suggest 240,000 newborns die from birth defects worldwide every year
- Birth defects range from anatomical problems, like cleft lip, to genetic problems, like sickle cell disease, which cause differences in how systems in the body function
- Many birth defects can be treated if identified early, though some require long term care



# Causes of birth defects

- One of the most preventable causes of birth defects is drinking alcohol during pregnancy, leading to Fetal Alcohol Syndrome
- Indirectly, socioeconomic factors can influence the rate of birth defects because pregnant women lack access to sufficient nutrition or have increased exposure to infectious diseases
- Environmental factors, such as infection, pollutants, and certain prescription drugs may also increase the risk of birth defects
- Some birth defects are genetic in origin

# CDC Tracker

- The CDC has a Environmental Public Health Tracking tool, where you can find information about many different birth defects
  - Click here to go to it > [National Environmental Public Health Tracking Network Data Explorer \(cdc.gov\)](https://natsnap.cdc.gov/)
- A question that could be asked is “**Is there a correlation between the prevalence of Anencephaly (an absence of the cerebral hemispheres of the brain) and acute toxic substance releases in Wisconsin?**”
  - For this, start with step 1, selecting Birth Defects, Prevalence of Anencephaly, per 10000 live births
  - For steps 2 and 3, select State by County data, for the state of Wisconsin
  - For Step 4, select all years of available date, and hit go
    - Optional filters to have gender, maternal age, and race/ethnicity could also be applied

Continued on next page

The screenshot shows the CDC Tracker Query Panel interface with the following configuration:

- STEP 1: CONTENT**
  - Search: [ ]
  - Birth Defects: [ v ]
  - Prevalence of Anencephaly: [ v ]
  - Prevalence of Anencephaly per 10,000 Live Births c: [ v ]
- STEP 2: GEOGRAPHY TYPE**
  - State By County (Data Not Smoothed): [ v ]
- STEP 3: GEOGRAPHY**
  - Missouri: [ ]
  - New Hampshire: [ ]
  - New Jersey: [ ]
  - New Mexico: [ ]
  - New York: [ ]
  - North Carolina: [ ]
  - Oregon: [ ]
  - Rhode Island: [ ]
  - South Carolina: [ ]
  - Utah: [ ]
  - Vermont: [ ]
  - Wisconsin: [ x ]
- STEP 4: TIME**
  - All Years: [ x ]
  - 2017-2021: [ x ]
  - 2016-2020: [ x ]
  - 2015-2019: [ x ]
  - 2014-2018: [ x ]
  - 2013-2017: [ x ]
  - 2012-2016: [ x ]
  - 2011-2015: [ x ]
  - 2010-2014: [ x ]
  - 2009-2013: [ x ]
  - 2008-2012: [ x ]
  - 2007-2011: [ x ]
- STEP 5: ADVANCED OPTIONS**
  - Optional: [ v ]
  - Infant Gender: [ v ]
    - Male: [ ]
    - Female: [ ]
  - Maternal Age Group: [ v ]
    - < 20: [ ]
    - 20 TO 29: [ ]
    - 30 TO 39: [ ]
    - >= 40: [ ]
  - Maternal Race Ethnicity: [ v ]
    - White, not including Hispanic: [ ]

Buttons at the bottom: Disclaimer, Clear Selections, GO →

## Question: Is there a correlation between the prevalence of Anencephaly and acute toxic substance releases in Wisconsin? (continued)

- The tracker allows for two simultaneous searches, so for the second one you can select the following settings
  - Word of caution, this data stopped being tracked in 2011

The screenshot displays the CDC Query Panel interface with the following settings:

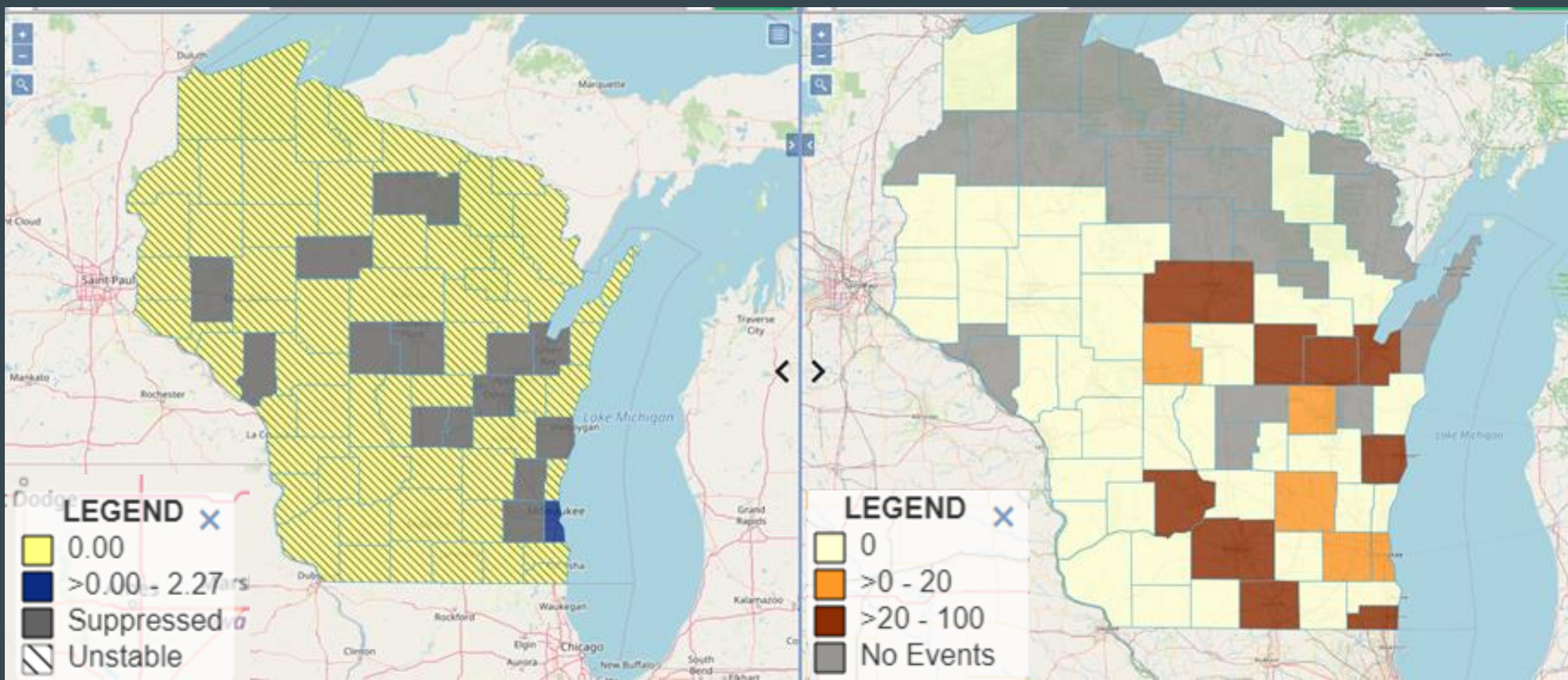
- STEP 1: CONTENT**
  - Search: Toxic Substance Releases
  - Acute Toxic Substance Releases
  - Percent of Reported Acute Toxic Substance Release
- STEP 2: GEOGRAPHY TYPE**
  - State By County
- STEP 3: GEOGRAPHY**
  - Wisconsin (checked)
- STEP 4: TIME**
  - All Years (checked)
  - 2011 (checked)
  - 2010 (checked)
  - 2009 (checked)
  - 2008 (checked)
  - 2007 (checked)
  - 2006 (checked)
  - 2005 (checked)
  - 2004 (checked)
  - 2003 (checked)
  - 2002 (checked)
  - 2001 (checked)
- STEP 5: ADVANCED OPTIONS**
  - Area Type: Industrial (unchecked)
  - Area Type: Commercial (unchecked)
  - Area Type: Residential (unchecked)
  - Area Type: Agriculture (unchecked)
  - Health Impact
    - Health Impact: With at least one injury or fatality (unchecked)
  - Incident Type
    - Incident Type: Transportation (unchecked)
    - Incident Type: Fixed-Facility (unchecked)
  - Primary Contributing Factor
    - Primary Contributing Factor: Equipment Failure (unchecked)

Buttons at the bottom: Disclaimer, Clear Selections, GO →

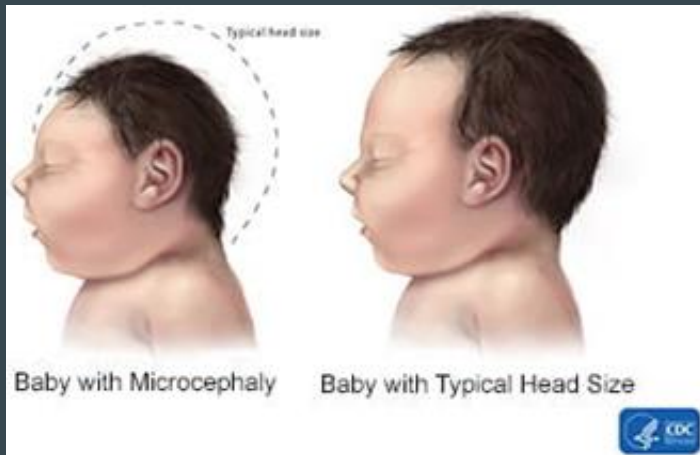
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## Question: Is there a correlation between the prevalence of Anencephaly and acute toxic substance releases in Wisconsin? (continued)

- As seen below, the settings pull up maps for the requested data, and puts them side by side for comparison
- Click here to learn more about the individual statistics tracked > [National Environmental Public Health Tracking Network | Indicators and Data \(cdc.gov\)](#)



# Infectious Influence: “Zika Virus”



- Zika virus is a mosquito-borne virus that is found mainly around the Equator
- Zika has occasional outbreaks, with one of the most recent occurring in India in 2021
- A causal link was established between Zika virus and birth defects like microcephaly, an abnormally small head
- Zika is usually mild, but due to the fact it can cause birth defects, travel restrictions are put in place if an outbreak occurs
- Currently, no vaccine for Zika virus is approved, so the best way to avoid it is to restrict travel to areas where Zika is common

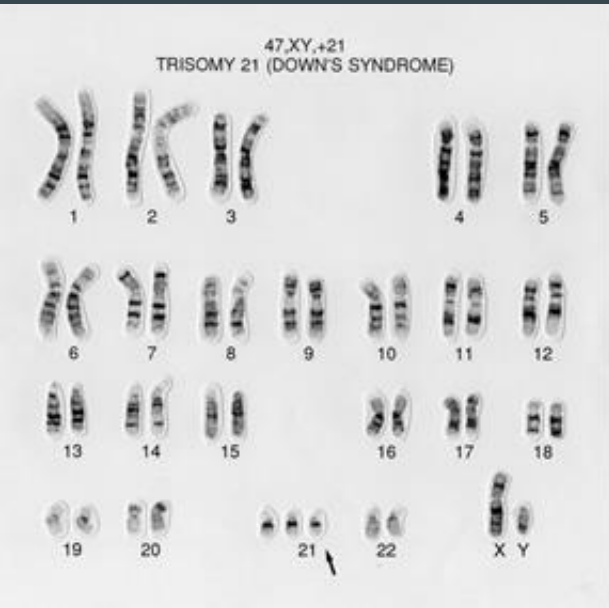


# Prescription Drug: “Thalidomide”

- Thalidomide was a prescription drug formerly used as a sedative, and to treat morning sickness in pregnant women
- However, thalidomid caused severe birth defects, such as short or missing limbs, malformation of hands and skeleton, and damage to organs
- It took around 5 years from the first identified case until the connection to these birth defects was clearly established
- Newborns who survived into adulthood suffer from additional effects from their disability, such as pain, reduced mobility, and numbness
- Pregnant people need to avoid medications that cause birth defects. Other such medications are Accutane, Roaccutane, and Warfarin



# Genetic Influence: “Trisomy 21”



- Trisomy 21, more commonly referred to as Down’s Syndrome, is a genetic defect
- Typically, a baby will receive half of the DNA from both parents, which get grouped into Chromosome pairs
  - See image to the left
- Trisomy 21 is a defect where the baby inherits three copies of chromosome 21, causing an imbalance that affects their development
- It is a lifelong condition, but early services can help improve the child’s life
- Trisomy 21 is just one of the many genetic causes of birth defects

# Environmental Contaminants: “Love Canal Incident”

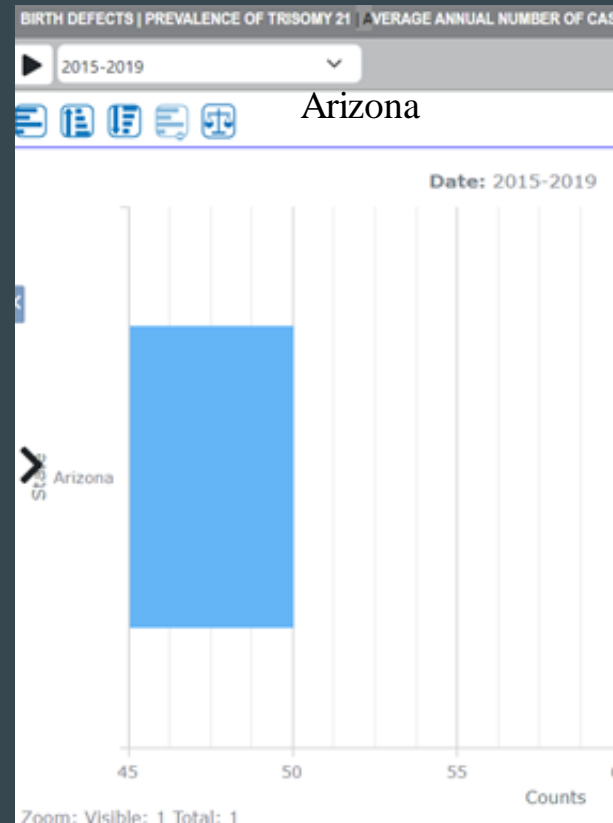
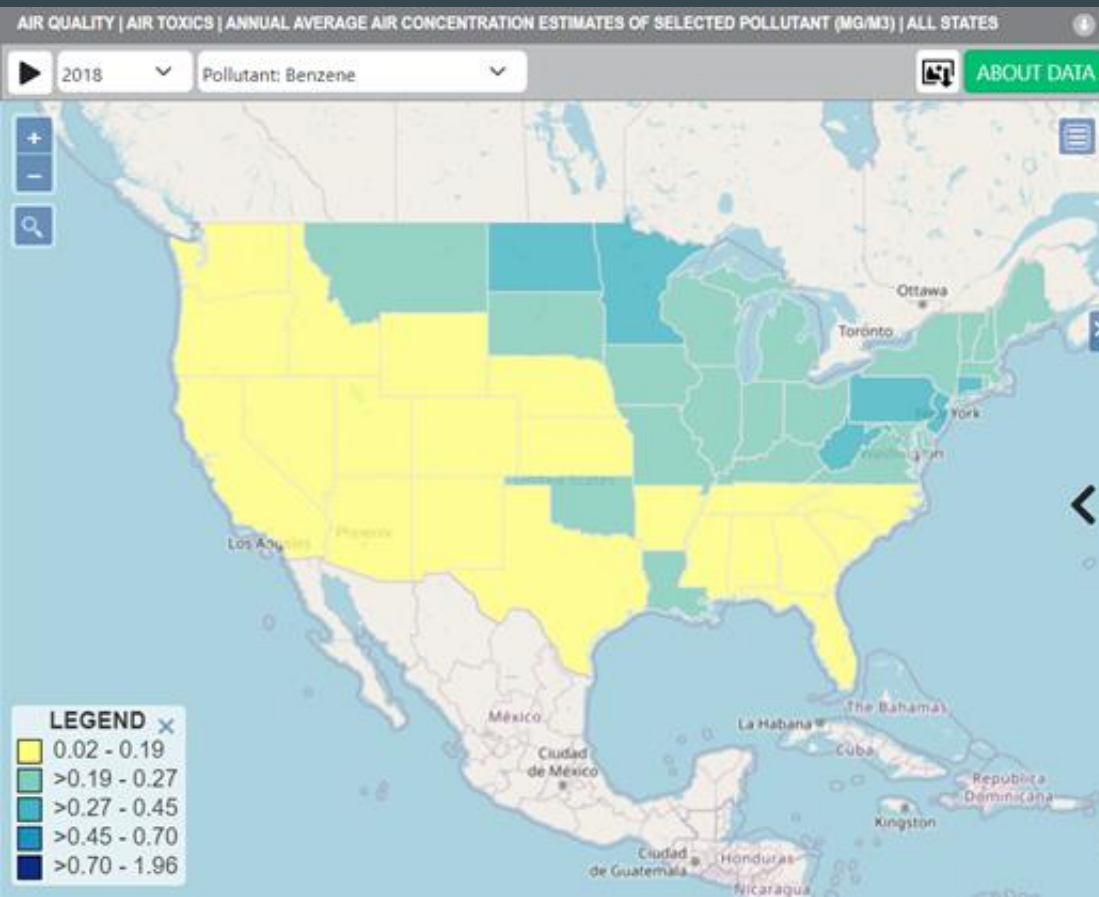


- Love Canal was a town built in in the late 1950’s on-top of a chemical disposal site
- There were many low-income apartments and single family homes built at Love Canal
- After a few decades, the chemicals began leaking into the environment and the homes
- From 1974 to 1978 an estimated 56% of children were born with birth defects
- The kinds of chemicals found at Love Canal included, but were not limited to, Benzene, Chloroform, Dioxin, and Toluene
- This example provides an example of one way to prevent birth defects: avoiding environmental contaminants like toxic chemicals, heavy metals, and pollution
- A question that could be asked using the CDC National Environmental Public Health Tracking Network is “**What correlation exists between Trisomy 21 and air pollution caused by benzene?**”

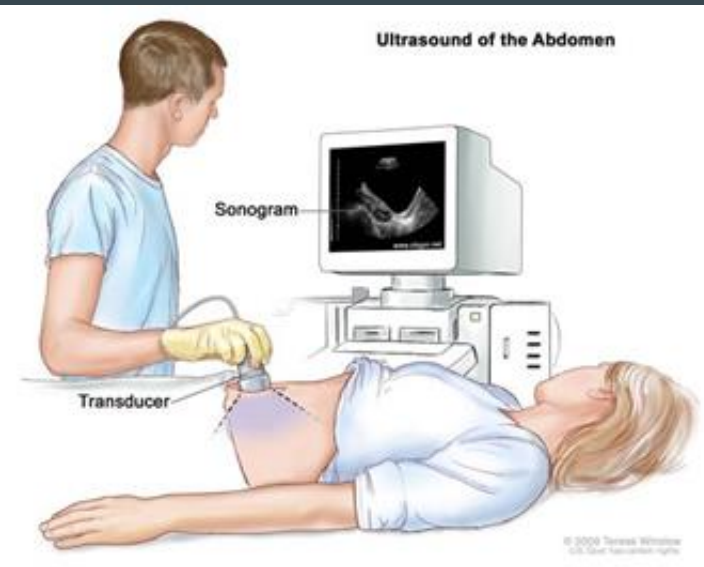
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## Question: What correlation exists between Trisomy 21 and air pollution of benzene?

- Utilize the CDC tracker once again, using the air pollution
- Utilize single state (no map) setting, to get graphical count
- Find that in Arizona, the concentration of Benzene in the air in 2018 was 0.06 with only 50 reported births of Trisomy 21, vs New York with a concentration of 0.25 and 126 births of Trisomy 21

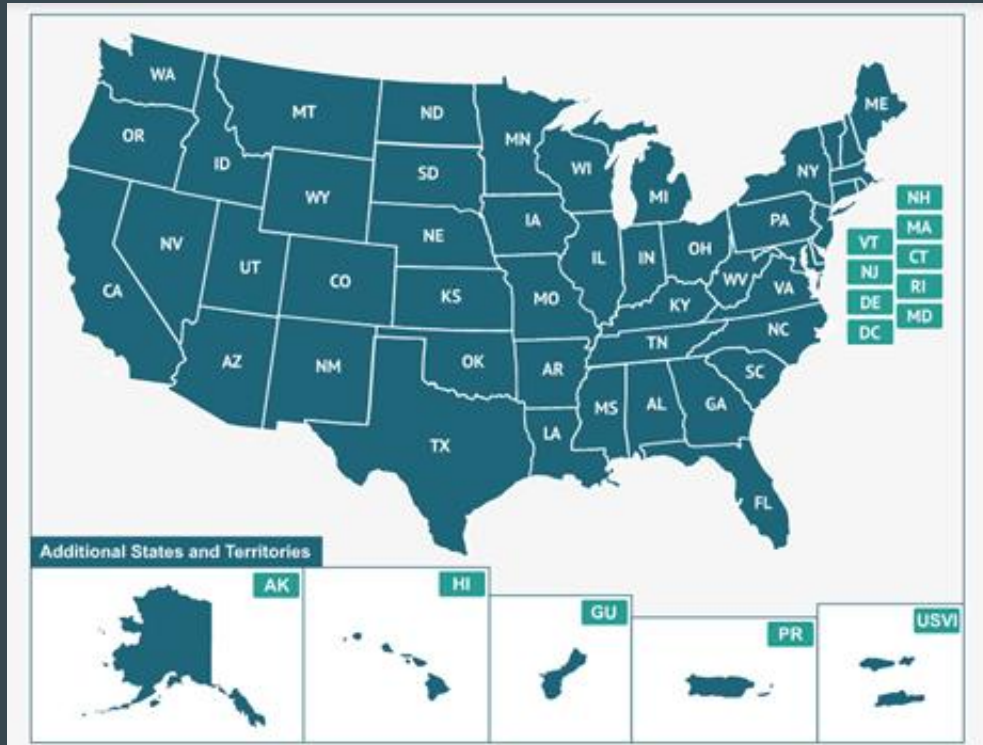


# Valuable Screening



- Screenings are valuable tests help detect birth defects early
- There are First Trimester, Second Trimester, and Newborn Screenings
- The screenings include blood tests, ultrasounds, fetal echocardiograms, to find any structural abnormalities or unusual hormone levels in the body
- If screening is positive, diagnostic tests such as High Resolution Ultrasound or Amniocentesis are used to confirm problems
- Amniocentesis is a test where some of the fluid around the fetus is taken and analyzed for genetic or chemical markers of birth defects
- To find out more, click here > [Diagnosis of Birth Defects | CDC](#)

# Valuable Screening: Newborn Screening



- Newborn screening is a public health service whose goal is to identify disorders in newborn children that need early recognition/treatment
- Each state has their own panel of screens, differing in scope
- Three parts, heel stick to gather a small blood sample, measure oxygen levels, and a hearing screening
- While the majority of the conditions are rare, screening detects disease in over 5000 newborns a year
- One useful tool for finding State Specific Newborn Screening information is [Baby's First Test | Newborn Screening | Baby Health \(babysfirsttest.org\)](https://babysfirsttest.org)
  - Click here to go to the website for individual state details > [Conditions Screened by State | Baby's First Test | Newborn Screening | Baby Health \(babysfirsttest.org\)](https://babysfirsttest.org)

# Screening Method: Congenital Heart Defects

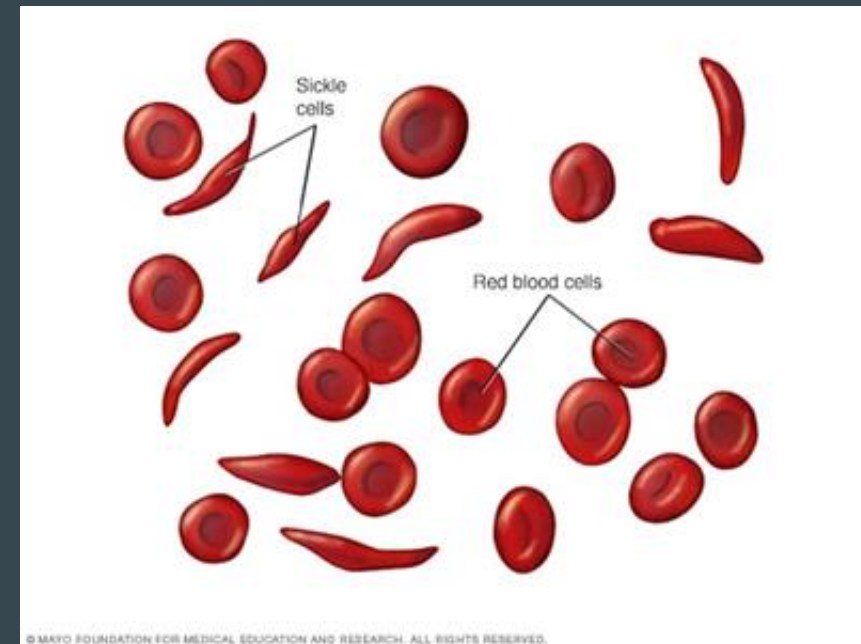
- Congenital heart defects (CHD) are some of the most common kinds of birth defects
- The kinds of heart defect can vary, with 1 in 4 children born with a CHD are “critical” and require surgery and procedures within the first year of life
- Ultrasounds can screen for CHD, but definitive diagnosis requires an echocardiogram, a specialized ultrasound that examines the function of the child's heart
- One tool used to screen newborns for CHD is a pulse oximetry, a device that measures level of oxygen in the blood as seen in the image (CHD can lead to poor circulation and so adequate oxygen is not circulated to all the infant's tissues)



# Commonly Screened disease: Sickle Cell Disease

- Sickle Cell Disease (SCD) is a disease that affects around 100,000 Americans, with a significantly higher proportion of African Americans affected, estimated to be around 1 in 500
- As seen below (picture on right), SCD leads to a change in the shape of blood cells that can damage or block blood vessels which leads to pain and organ damage. Cells will degrade more quickly, decreasing amount of oxygen in the blood
- Unlike Trisomy 21, SCD is a recessive disease, so one must inherit a mutant gene (little “a” in the chart on the left) from both parents
- A question could be “**Is the number of screened Sickle cell Births related to population density of African Americans in Alabama?**”

	A	a
A	AA	Aa
a	Aa	aa





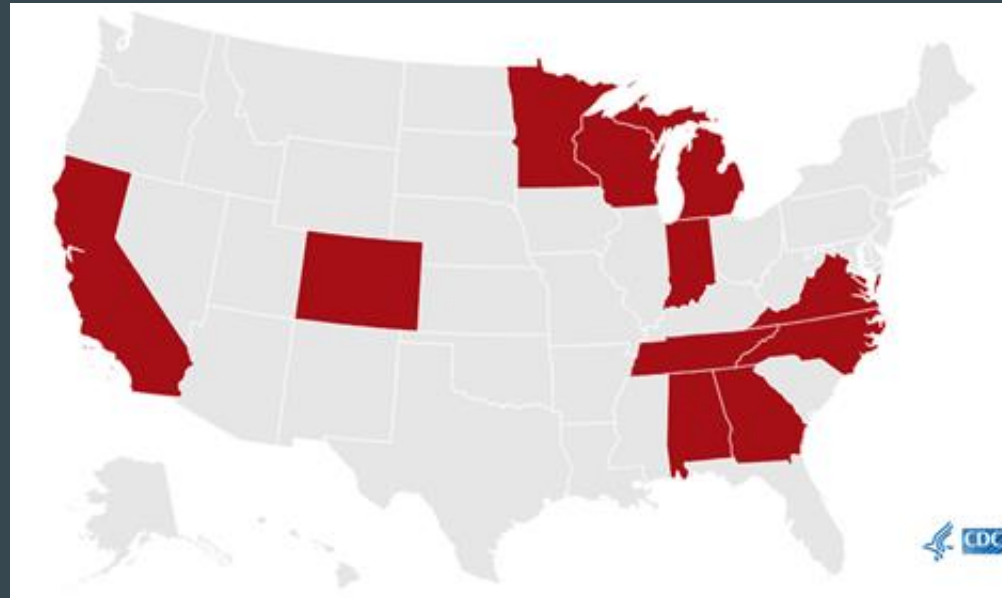
# Census Data Maps

- To answer the question “Is the number of screened Sickle cell Births related to population density of African Americans in Alabama?”, United States Census Bureau’s Interactive Maps is helpful
  - To go to the website, click here > [Interactive Maps \(census.gov\)](https://www.census.gov/interactive-maps/)
- Click on the 2020 Census Demographic Data Viewer
- Scroll down again to find the live application link on the webpage (See below)
- Zooming into Alabama, you get the graphical view of each county.
  - Clicking on individual counties will bring up the exact percentage, along with total population of just African American vs total county population.
- Continued on next slide



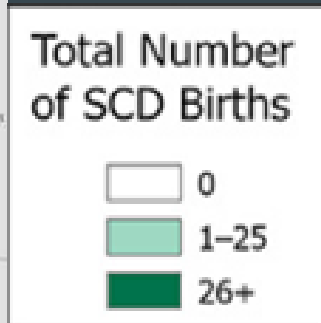
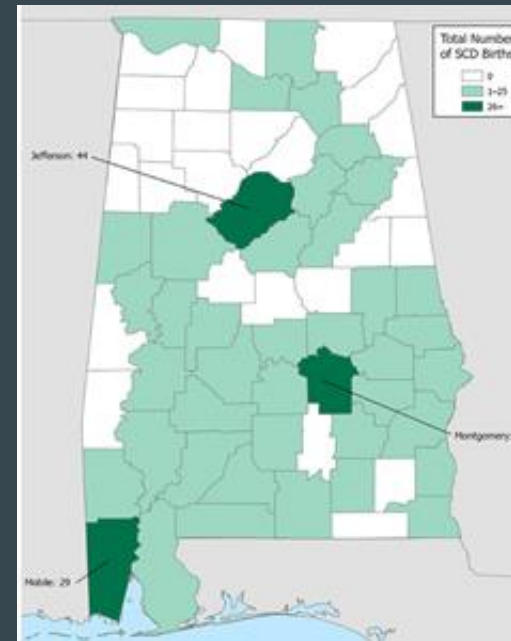
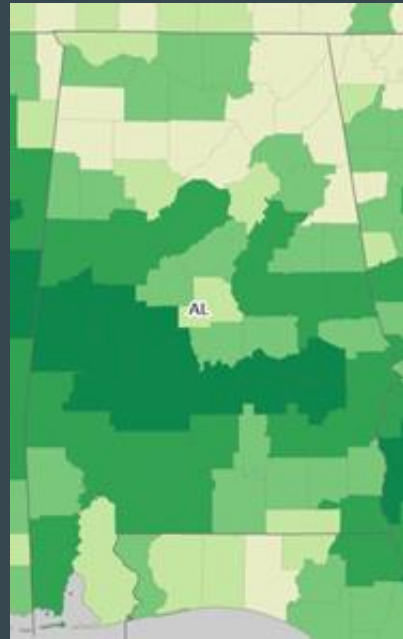
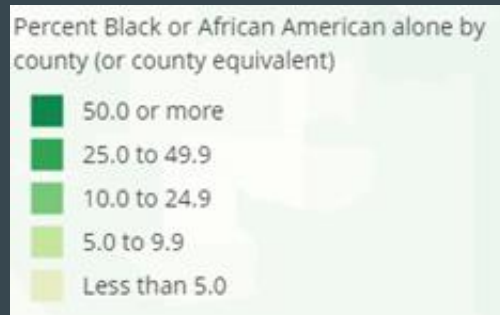
# Sickle Cell Disease Research Newborn Screening Data (continued)

- **Question: Is the number of screened Sickle cell Births related to population density of African Americans in Alabama?**
- To help answer this question with the Census Data, it needs to be compared with Data for SCD
- The Center for Disease Control has a dataset for SCD that covers 11 states > [Newborn Screening \(NBS\) Data | CDC](#)
- Click on a state, and it will provide graphical data for number of SCD births by county, annual number of SCD births, the number of total births by sex, and type of SCD



# Question: Is the number of screened Sickle cell Births related to population density of African Americans in Alabama? (continued)

- The left two images were taken from the Census Data Maps, while the both right images were from the NBS Data for Alabama
- We find that most of the reported births of SCD are in three counties, of which one was predominantly African American.
- Overall, there appears to be some overlap between counties with SCD births and a higher density of African American population, but it doesn't perfectly correlate





# Importance of Early Screening

- Early Screening is very important
- Early screening leads to early identification of a problem and therefor early treatment
  - For birth defects, it allows for earlier interventions for correction or preparation
  - For newborn screening, it can help mitigate diseases with specialized treatment
- Without this early detection, many children would face long term disability when all that is needed to prevent this is as simple as avoiding certain foods.

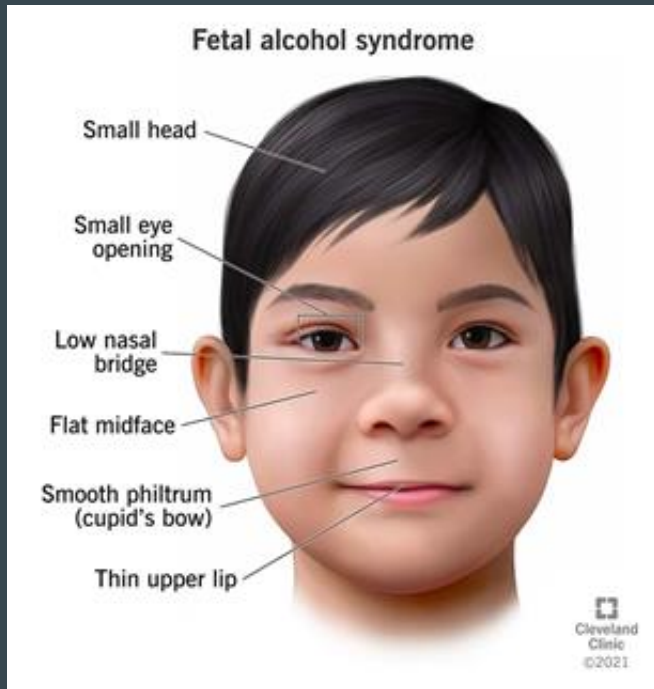


# Prevention of Birth Defects

- Even though many birth defects and newborn illness can be screened for, with some being treatable, not all of them can be prevented
- Avoiding external risk factors, such as the previously mentioned Zika virus, is one way to reduce the risk of birth defects
- There are, however, some birth defects that are easily preventable

# Prevention: “Fetal Alcohol Spectrum Disorders”

- This spectrum of disorders occurs when a fetus is exposed to alcohol prior to birth
- There is no safe amount of alcohol to drink when pregnant, and all kinds of alcohol are harmful
- There are numerous symptoms FAS, but specific birth defects include abnormal facial features, smaller head size, vision/hearing problems, and problems with the heart, kidneys, or bones.
- It is also important to note that tobacco use can increase risks for birth defects, so avoiding recreational drugs like alcohol and tobacco while pregnant is strongly recommended
- A possible research question could be “**Is binge drinking more common in rural or urban locations?**”
  - Continued on next slide



## Question: Is binge drinking more common in rural or urban locations? (continued)



- To answer this question, the United States Census Bureau's Interactive Maps is helpful
  - To go to the website, click here > [Interactive Maps \(census.gov\)](https://www.census.gov/interactive-maps/)
- Scroll down to click on "Rural America: A Story Map"
- From there Click on the "Did you Know these Rural Facts" and scroll down to the "Geographic Areas and Rural Data selection".
- Continued on next slide

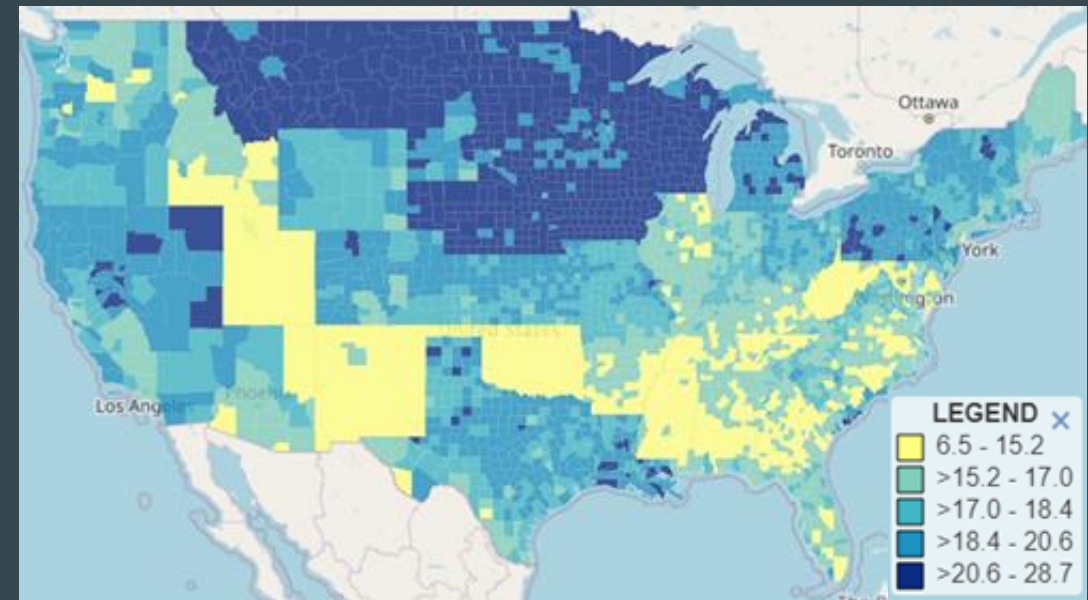
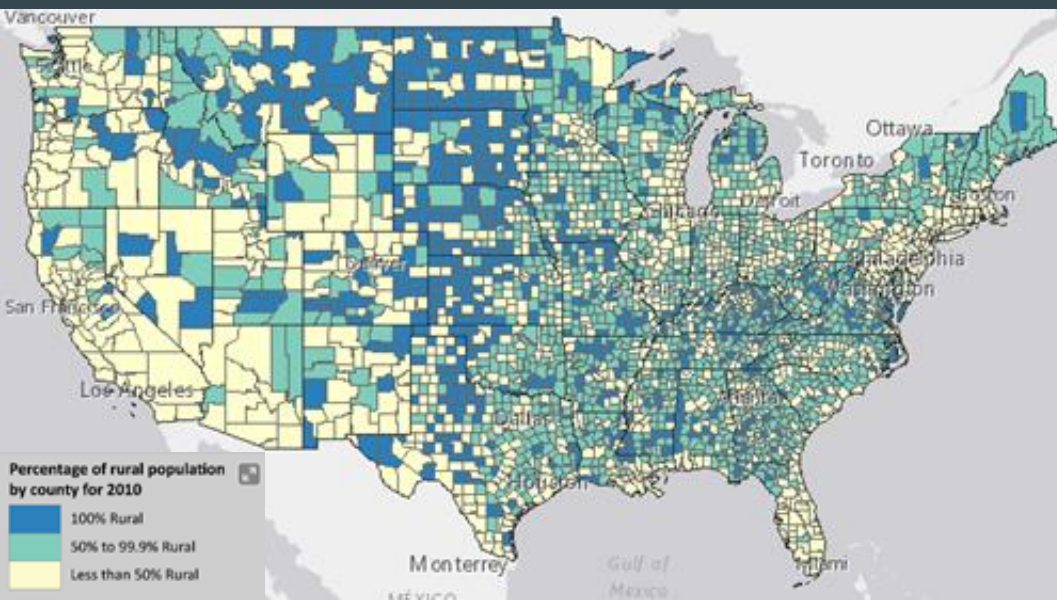
How Does the Census Bureau Define Rural?

Where Can I Find Data for Rural Areas?

Did You Know These Rural Facts?

## Question: Is binge drinking more common in rural or urban locations? (continued)

- In the Geographic Areas section, it splits and categorizes the counties in America based on percentage of rural population, shown by the map in the bottom left
- For comparison, utilizing the CDC tracker once more, we can view prevalence of binge drinking by county across all of the United States
- While there appears to be some overlap between rural population and binge drinking, there are also urban locations that have a high prevalence of binge drinking







# Additional Alcohol related databases

- The CDC has a web page that shows prevalence of alcohol use for specifically women, as it relates to prevention of fetal alcohol syndrome, it shows reported alcohol use % by state.
  - Click here to learn more > [State-level Estimates of Alcohol Use Among Women – 2019 | CDC](#)
- The CDC also has a web page for statistics relating between Alcohol use and Public Health outcomes with numeric values for each state
  - To learn more, click here > [Alcohol-Related Disease Impact | CDC](#)



# Prevention: Spina Bifida

- Spina bifida is a birth defect where the spine does not develop correctly, with some of the spinal cord exposed on the back
- Spina Bifida can be detected via an ultrasound
- The best way to help prevent this birth defect is providing sufficient folic acid (a vitamin that helps prevent this condition) to the mother during pregnancy
- Folic acid is found in various foods like peas, oranges, broccoli, and spinach, though additional supplemental vitamin pills are recommended
- In general, properly balanced diet with proper micronutrients like Iron, Iodine, Zinc, Vitamin A and Vitamin D are necessary for a healthy pregnancy
- One question could be **“Is there a correlation between Spina Bifida birth rates and high poverty areas in Missouri?”**

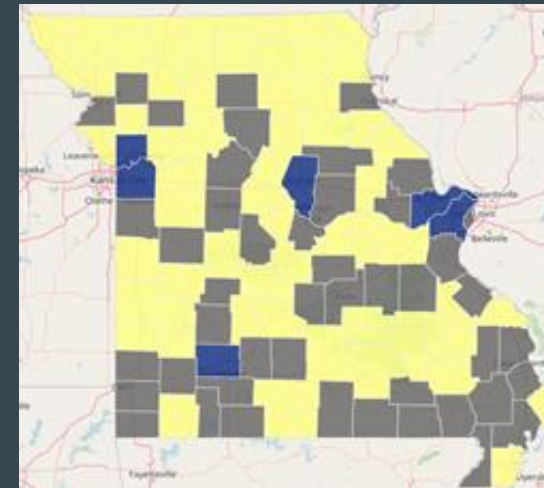
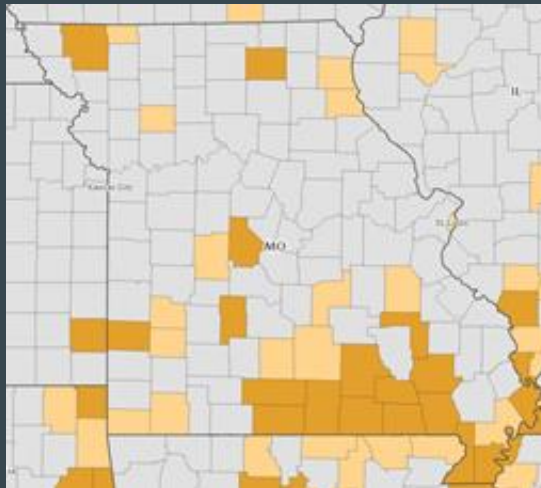
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next slide

## Question: Is there a correlation between Spina Bifida birth rates and high poverty areas in Missouri?

- Once again, using the [Interactive Maps \(census.gov\)](https://data.census.gov/tables//geographies/states/missouri/interactive-maps), scroll down to the EDA section, it will help provide data on poverty levels by county.
- Zooming in on Missouri, the map is divided into the individual counties
  - The provided legend shows which counties are considered to have high poverty status
- To compare, the [National Environmental Public Health Tracking Network Data Explorer \(cdc.gov\)](https://nndex.cdc.gov/) will provide a comparison map
  - In this example, the selected metric was “average annual number of cases among live births over a 4 year period”
- The interactive map and legend shows that only a few counties reported live births of Spina Bifida, and there is no apparent overlap between the rate of Spina Bifida and poverty levels

### High Poverty Status

-  **High poverty area**  
20% of the population or more are below the poverty level
-  **Possible high poverty area**  
The confidence interval includes “20% of the population or more are below the poverty level,” but the point estimate is lower than that threshold
-  **Not a high poverty area**  
Areas that do not meet either of these criteria
-  **Data not available**  
Data is not shown for these areas



# Additional Helpful Resources

- [Data & Statistics on Birth Defects | CDC](#)



- [Birth defects and your baby | March of Dimes](#)

