The Michigan Monitor

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MBDR MICHIGAN BIRTH DEFECTS REGISTRY

CONGENITAL DIAPHRAGMATIC HERNIA

Background

Congenital diaphragmatic hernia (CDH) is a defect in the diaphragm that a person is born with due to abnormal development.[1,2] The diaphragm is the main muscle used to breathe. It separates the lungs and heart in the chest from the stomach and other organs in the abdomen. The diaphragm develops early in pregnancy, during weeks 5 to 7 after conception.

CDH can range in severity from a thinned area or a hole in a part of the diaphragm to its complete absence.[2] If organs in the abdomen, such as intestines, stomach, or liver, push through the hole, it can prevent normal lung development. Newborns with CDH may have life-threatening breathing difficulties.[3]

The diagnosis of CDH often happens prior to birth. If not diagnosed during pregnancy, it may be diagnosed soon after birth if the baby has trouble breathing.[3] Treatment depends on the type and severity of the CDH; it typically includes surgery.[4]

CDH may occur as part of a syndrome and may be caused by a genetic mutation or by a chromosome abnormality. CDH may occur with other birth defects, or as an isolated defect with no other abnormalities.[2] CDH is rarely inherited.[1] Often, the cause is unknown.

Environmental factors that are known to influence fetal development may increase the risk of CDH. More research is needed.[3,5]

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Cases of CDH are reported to the Michigan Birth Defects Registry (MBDR) by hospitals and other reporting entities. This issue presents CDH trends for babies born in Michigan during 2008-2016.

Points of Interest

- In the United States, CDH affects around 950 – 1,000 newborns each year.
- CDH may occur alone, may be associated with a syndrome, and can occur with other birth defects.
- Treatment of CDH typically involves surgery; lung and heart health are key.

Etiology

The cause of CDH in most babies is unknown. Possible causes include:

- Chromosome abnormalities and other genetic factors
- Rare genetic syndromes
- Environmental factors

Congenital Diaphragmatic Hernia per MBDR Reporting, 2008-2016

Occurrence

In the United States, CDH affects about 1 in 2,500 to 1 in 4,000 babies each year.[1,6]

In Michigan, a total of 354 infants born between 2008 and 2016 were reported to the MBDR with a CDH diagnosis. The prevalence of CDH in Michigan remained stable over this time period (Figure 1). The overall CDH prevalence rate for the entire 2008-2016 time period was 3.4 cases per 10,000 live births.

Demographics

The prevalence of CDH in Michigan between 2008 and 2016 was assessed by selected demographic factors (Table 1). The overall prevalence was highest among infants born to mothers under 20 years of age (5.1 per 10,000 live births) and infants born preterm (9.4 per 10,000 live births). The highest rate of CDH by maternal race was reported for infants born to mothers of other races (4.9 per 10,000 live births) and infants born to Arabic mothers (4.3 per 10,000 live births). There was a small difference in the prevalence of CDH by sex of the infant (male infants: 3.8 per 10,000 live births versus female infants: 3.1 per 10,000 live births).

Infant Mortality and Fatality

CDH is a serious and often life-threatening condition. Between 2008 and 2016, the infant mortality rate (IMR) due to CDH in Michigan was 1 death of a baby with CDH per 10,000 live births. The case fatality rate (CFR) was 282.5 deaths per 1,000 infants born with CDH.

About 3 out of every 4 babies with CDH who died during their first year of life were born with CDH plus other birth defects. Studies find that babies born with CDH as part of a genetic syndrome or with major structural birth defects, especially heart defects, are at highest risk.[7]

Figure 1: Three year moving prevalence rate of congenital diaphragmatic hernia: MBDR, 2008-2016

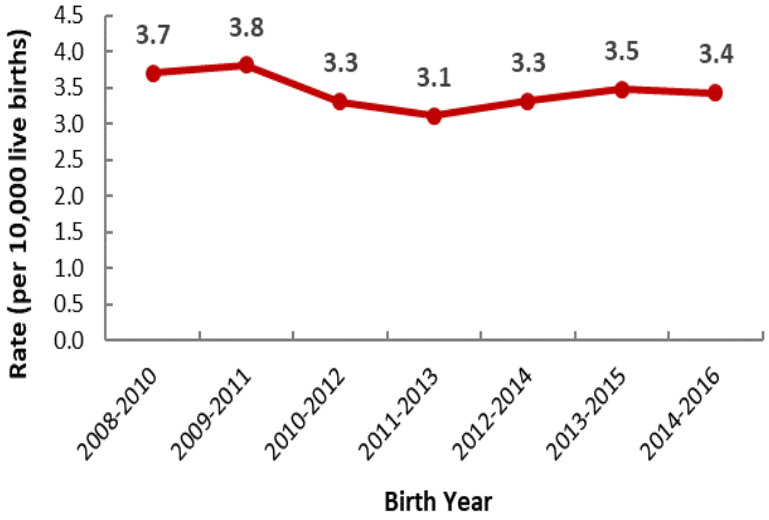


Table 1: Prevalence rate of congenital diaphragmatic hernia (CDH) stratified by selected demographic variables: MBDR, 2008-2016

Demographic	Number of CDH Cases	Prevalence Rate[1,2]
Total[3]	**354**	3.4
Maternal Age*		
<20	41	5.1
20-24	72	2.9
25-29	103	3.0
30-34	98	3.8
35+	40	2.9
Maternal Race		
White	260	3.4
Black	59	3.0
Other[4]	34	4.9
Gestational Age at Birth*		
Preterm (<37 Weeks)	98	9.4
Not Preterm (37+ Weeks)	256	2.8
Maternal Ethnicity*		
Hispanic	27	3.8
Arabic	17	4.3
Sex of Infant*		
Male	200	3.8
Female	154	3.1

Footnotes to Table 1.

[1]Prevalence rates are based on births to mothers living in Michigan at the time of delivery. Data are current through December 2016.

[2]Prevalence rate expressed as cases per 10,000 live births

[3]Number of cases in specific categories may not add up to the total number of cases due to missing variables

[4]Includes women who do not define themselves as Black or White and includes Native American and Asian/Pacific Islander

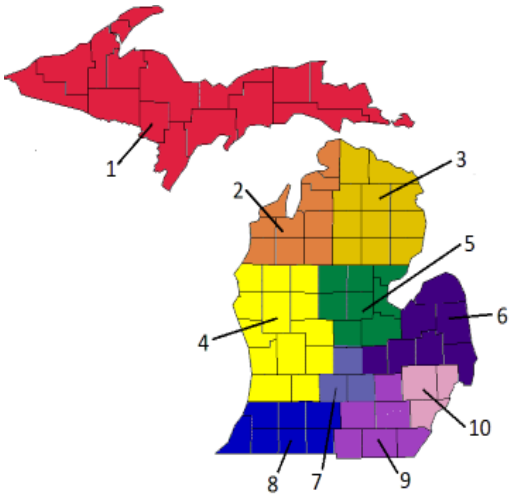
***Significantly different at p ≤ 0.05**

Maternal Residence

Michigan is organized into 10 prosperity regions. Table 2 displays the prevalence of CDH by the mother's resident prosperity region at the time of the birth. There were infants born with CDH throughout the state. The prosperity region with the highest prevalence rate was region 7 with 5.7 cases per 10,000 live births. Prosperity region 8 had the lowest prevalence rate with 2.3 cases per 10,000 live births.

Table 2: Prevalence of CDH by prosperity region of maternal residence: MBDR, 2008-2016

Prosperity Region	Number of Cases	Prevalence Rate[1,2]
1	*	*
2	14	5.2
3	*	*
4	81	4.5
5	15	2.7
6	23	2.7
7	27	5.7
8	20	2.3
9	32	3.4
10	134	3.2
Total	354	3.4



Footnotes to Table 2.

[1]Prevalence rates are based on births to mothers living in Michigan at the time of delivery. Data are current through December 2016.

[2]Prevalence rate expressed as cases per 10,000 live births.

***Data not shown due to small sample size.**

Unlocking the Mystery

The exact cause of CDH in a baby is rarely known. Studies suggest a role for both genetic and environmental factors. A specific **genetic** cause may be found in about 10-30% of cases. Most babies born with CDH will be the only affected person in their family.

Studies show that the development of the heart, lungs and diaphragm are closely related. New genetic technologies have helped find more genes that contribute to the normal development of the diaphragm. Some current research is focused on the part that these genes play, how they work together, and how gene variants (also called mutations) affect development. Twin studies and other research shows that a genetic variant can sometimes be new and unique (also called *de novo*) in a baby born with CDH. New genetic variants happen by chance.

CDH may be linked to chromosomal conditions. This includes, for example, Trisomy 21 (Down syndrome), as well as rare chromosome anomalies. Most of these occur by chance and are not inherited.

CDH is also known to be associated with inherited genetic syndromes. Babies with CDH are more likely to be born with other birth defects, such as heart defects. A genetic evaluation may be very helpful for families who learn that their baby has CDH during pregnancy or after birth.

A variety of **environmental** factors appear to be linked to a higher chance of having a baby with CDH. Some examples are:

- Maternal health factors such as malnutrition in pregnancy (with very low Vitamin A levels) and pregestational diabetes type 1 and type 2,
- Maternal health behaviors (like smoking and alcohol use), and
- Use of certain medications during pregnancy, like mycophenolate (helps prevent rejection of a donor organ).[8]

These observations provide clues to the complex factors behind CDH and other serious birth defects.

Good News

The prevalence rate for CDH in Michigan appears steady over time. The outlook for people with CDH keeps improving. Research continues to improve surgery and other life-saving treatment options. Regular screening and family support help infants and children with CDH receive needed therapy and other services promptly.

Birth Defects Education and Outreach Program,
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The Michigan Monitor is online:
URL: www.michigan.gov/mchepi
MBDR reporting:
URL: www.michigan.gov/mbdr

Give us your feedback!

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MDBR Data Limitations and Cautions

- Data are based on passive reporting which means MBDR relies on facilities to identify and report cases of birth defects. Not all facilities report cases as completely and timely as would be the ideal.
- Some facilities may report children with a birth defect that is later "ruled out" resulting in an overcount of the actual number of cases.
- Children diagnosed in other states may be missed which will affect the completeness of data for Michigan's border counties.

Meet our New Staff Member!

Amy Bohner is the new Birth Defects/Family Planning Epidemiologist. She received her undergraduate degree in public health from Calvin College (now called Calvin University) and received a master's degree in epidemiology from the University of Iowa. Before coming to the MDBR, Amy worked in injury epidemiology and is excited to now be working in a field more aligned with her interests. You can contact Amy via email at BohnerA2@Michigan.gov.

Resources for Health Professionals and Families

Michigan

- Children's Special Health Care Services (CSHCS) Program URL: **www.michigan.gov/cshcs**
- Family to Family Information Center URL: **f2fmichigan.org**
- Michigan Association of Genetic Counselors URL: **MAGCinc.org**

National

- Centers for Disease Control and Prevention, National Center on Birth Defects and Disabilities URL: **www.CDC.gov/NCBDDD/birthdefects**
- March of Dimes URL: **http://www.marchofdimes.org**
- National Birth Defects Prevention Network (NBDPN) URL: **www.nbdpn.org**
- Congenital Diaphragmatic Hernia (CDH) Support URL: **cdhi.org**

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