#### Birth Defects Team



Michigan Birth Defects Registry (MBDR) - Vital Records and Health Data Development Section

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# BIRTH DEFECT

"Birth defect, congenital malformation, and congenital anomaly are synonymous terms used to describe structural, behavioral, functional, and metabolic disorders present at birth."

Langman's Medical Embryology, ed. 9

"...an abnormality of the body's structure or inherent function present at birth, whether the abnormality is detected at the time of delivery or becomes apparent at a later date." *MBDR Reporting Manual* 

### Genomics of Birth Defects Infectious disease syphilis / cytomegalovirus Chronic disease metabolic / endocrine **Birth** Defects **Genetic factors** inherited / new changes Other exposures alcohol / toxic substances



### Chondrodystrophy – 756.4

- 1/15,000-1/40,000
- Disproportionate short stature, symmetrical
- Autosomal dominant, often new mutation



#### Achondroplasia

Medical Genetics, 2<sup>nd</sup> Edition. Downloaded October 22, 2007 from the worldwide web. MBDR has 152 cases, 1992-2003, for a birth prevalence rate of ~1/10,000.



• 1/100,000

- Multiple enchondromas near growth plates, assymetrical
- Usually sporadic

#### Ollier disease

Universiteit Leiden, Medical Research Center Downloaded October 22, 2007 from the worldwide web.

#### Major Classes of Anomalies - 2003



#### Birth Defects Reporting - 2003

	Prenatal	Postnatal				
NTDs	Cases (*)	MBDR	MBDR+			
Anencephaly	10 (5)	18, <i>1.4</i>	23, <i>1.8</i>			
Spina Bifida	10 (3)	48, <i>3.7</i>	51, <i>3.9</i>			
Encephalocele	5 (4)	13, <i>1.0</i>	17, <i>1.3</i>			
Chromosomal Disorders						
Trisomy 21	25 (18)	143, <i>11.0</i>	161, <i>12.4</i>			
Trisomies 13/18	31 (22)	41, <i>3.2</i>	63, <i>4.9</i>			
Other	31 (14)	115, <i>8.9</i>	129, 10.0			

\* no. known miscarried/stillborn/VToP for 7 MI facilities

#### **Key Prevention Activities**

Promote healthy behaviors for women before and during pregnancy

January is Birth Defects Prevention Month
 Also Folic Acid Awareness Week

Collaborate to develop and deliver primary prevention messages and strategies

Listserv: <u>DCH-BDPM@LISTSERV.MICHIGAN.GOV</u>

 Multi-vitamin distribution to low income women in high risk regions: expanded to 40 counties in 2007

 Family history initiative for second degree relatives of children with NTD Folic Acid Outreach and Multivitamin Distribution in Selected Michigan Counties



Funded 2005-2007 by March of Dimes Community Awards Grant

- To distribute 30,000 bottles of free multivitamins to low-income women of childbearing age
- To instill a healthy habit that will continue after project completion
- To reinforce the critical role of the health professional in the delivery of prevention messages

## Michigan PRAMS 2003

#### Consumption of a multivitamin in the month before pregnancy by awareness of / instruction about folic acid



4-6 times per week

Daily

Patel R, Bouraoui Y, Grigorescu V, McGrath-Miller. Michigan PRAMS Report 2003: Michigan Department of Community Health. Family and Community Health, July 2006.

- Community-based programs may be most effective in reaching these at-risk women with low awareness
- These women may have limited access to information through computers, pamphlets or other specific forms of media

# Michigan Plan

- Target outreach
- Educate women
- Provide free vitamins
- Monitor distribution
- Follow-up on use

and a start			
Region	Number	Rate	
State	922	5.8	
1	274	5.2	5 7
2	93	4.9	6 2
3	74	6.0	4 3
4	92	6.1	NTDs in Michigan
5	153	6.8	Number and rate
6	49	5.6	per 10,000 live
7	48	5.3	births, MBDR
8	74	6.7	1772-2005
9	45	8.0	
10	20	5.5	



#### Family History of NTD and Folic Acid Study

1. MBDR to find index NTD cases (23)

Can we use family history to target folic acid outreach?

2. Contact index families (7) to identify eligible relatives (22) 4. Folic acid outreach
– information
mailed to female
relatives (3)

5. Follow up survey of female relatives

3. Survey female relatives (4) Not everything that can be counted counts, and not everything that counts can be counted.

Albert Einstein

