Clinical Genetic Services: A View from Michigan's Children's Special Health Care Services' (CSHCS) Families

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Introduction

The Michigan Birth Defects Registry (MBDR) data suggests that about 70% of individuals reported with a birth defect suspected as having a genetic etiology are not seen in genetic centers.[1] Previous research has found that genetics centers may be underutilized because health care providers lack familiarity with genetic disorders and may not be aware of all services that are available.[2] Other barriers to accessing genetics services may include lack of knowledge of genetic services, lack of awareness of risk, and location of services, among other issues.[3]

Purpose

The purpose of this study was to further explore barriers to genetic services and to determine the effects of selected variables, including type and severity of diagnoses and information given about genetics, on having been told of genetic services and having been to a genetics clinic.

Figure 1 Michigan Pediatric Genetics Centers and outreach sites through June, 2009.

Methods

• **Source of data and study design**: This is a cross-sectional study using data from the survey, "Clinical Genetic Services: A View from Michigan's Children's Special Health Care Services' (CSHCS) Families," collected from Michigan families between April 10 and June 25, 2009. The MDCH IRB reviewed the study to determine that human subjects were adequately protected.

• **Source population and subject selection**: Eligible participants were residents of Michigan, 18 years or older, and had children enrolled in Children's Special Health Care Services (CSHCS) for a birth defect(s) or congenital disorder(s) with possible genetic etiologies. Surveys were mailed to 1400 families with enrollee birth from 2004-2008 and 850 families with enrollee birth from 1994-1998. Completed surveys were returned to the Birth Defects Program and respondents were eligible to receive a gift card upon completion of the survey.

• **Data and variables**: Outcome variables were dichotomous and each diagnosis type was analyzed as a dichotomous variable. Other covariates included: syndrome type, told condition was genetic, area of residence, education, race, and age of child (born from 1994-1998 or born from 2004-2008).

• **Statistical analysis**: Logistic regression was used to estimate the crude and adjusted associations (odds ratios and 95% confidence intervals) between exposure variables and each outcome. Statistical analysis was done using SAS v. 9.1.

Results

Table 1. Study population demographics.

Variable Category	N. Subjects	% of Total
Race/Ethnicity		
White, NH	345	78.6
Black, NH	33	7.5
Other	61	13.9
Education		
<H.S.	28	6.5
H.S.	112	26.0
Some College	131	30.5
College/Grad. Degree	159	37.0
Area of Residence		
Northern/UP	39	8.6
Western	165	36.3
Central/Eastern	102	22.4
South Eastern	149	32.7
Age of child		
Born from 2004-2008	295	64.6
Born from 1994-1998	162	35.4

Table 1 and Figure 2:

• Survey responses were obtained from a total of 457 families, for a response rate of ~20%.

• About 36% of families reported being told of genetics services, and about 32% of families reported having been to a genetics clinic.

• Prevalence of **told of genetic services** was *highest* among those who were black, had not completed high school, from Northern Michigan or the Upper Peninsula, and had children born from 2004-2008.

• Prevalence of **going to a genetics clinic** was *highest* among those who were not white or black, had not completed high school, from South Eastern Michigan, and had a child born from 2004-2008.

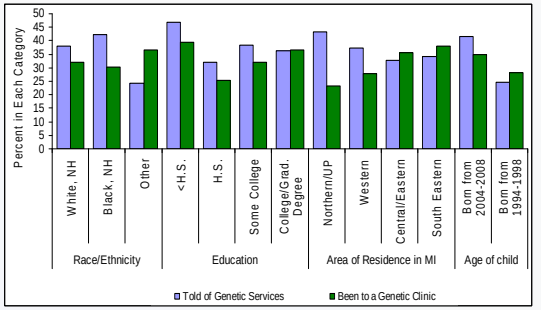

Figure 2 Frequency of having been told of genetic services and having been to a genetics clinic by category of demographic variable.

Table 2. Estimated crude and adjusted effects of selected variables on being told of genetic services: Clinical Genetic Services Survey, Michigan, 2009.

Predictor Category	Number of Subjects	% of Total	Crude OR	Adjusted† OR	Adjusted† 95% Confidence Interval
Diagnosis*					
CNS/Spina bifida	57	8.8	**2.0**	1.9	(0.93, 3.7)
Chromosomal	45	6.9	**2.4**	**2.3**	**(1.1, 4.9)**
Cleft lip/palate	136	21.0	1.3	1.5	(0.90, 2.5)
Ear, Face, Neck	39	6.0	1.4	1.4	(0.61, 3.3)
Endocrine/Metabolic	8	1.2	*Insufficient Data*		
Eye	24	3.7	1.2	1.1	(0.35, 3.6)
Hearing Loss	47	7.3	1.4	1.4	(0.64, 3.0)
Heart	93	14.4	**0.51**	**0.49**	**(0.25, 0.95)**
Limb	29	4.5	1.1	1.0	(0.40, 2.6)
Urogenital/Renal	46	7.1	1.7	1.7	(0.77, 3.8)
Other	124	19.1	**2.0**	**2.3**	**(1.3, 4.0)**
Total**	648				
Syndrome					
Single Anomaly	222	52.1	1	1	*reference*
Multiple Anomalies	93	21.8	**1.8**	1.8	(0.94, 3.4)
Known Syndrome	111	26.1	**3.0**	**3.2**	**(1.7, 5.8)**
Total**	426				
Told Condition was Genetic					
No	215	48.5	1	1	*reference*
Yes	228	51.5	**2.3**	**2.4**	**(1.4, 4.0)**
Total¥	443				

* The reference category for each diagnosis category is those who do not have that specific dianosis, but have any other diagnosis.
** Total is more than 457 (the total sample size) because subjects may have more than one type of diagnosis and could be counted more than once.
¥ Totals are less than 457 (the total sample size) because of missing data.
† Adjusted for area of residence, education, race, and age of child.

Table 2: Being Told of Clinical Genetic Services

By diagnosis:

• Parents of children with a **chromosomal** condition were more likely to be told of services than children with any other condition (OR=2.3, 95% CI: 1.1, 4.9).

• Parents of children with a **heart** condition were less likely to be told of services than children with any other condition (OR=0.49, 95% CI: 0.25, 0.95).

By syndrome:

• Parents of children with a **known syndrome** were more likely than parents of children with a **single anomaly** to be told of services (OR=3.2, 95% CI: 1.7, 5.8).

By told condition was genetic:

• Parents who were **told their child's condition was genetic** were more likely than those not told about the condition to be told of genetic services (OR=2.4, 95% CI: 1.4, 4.0).

Table 3. Estimated crude and adjusted effects of selected variables on having been to a genetics clinic: Clinical Genetic Services Survey, Michigan, 2009.

Predictor Category	Number of Subjects	% of Total	Crude OR	Adjusted† OR	Adjusted† 95% Confidence Interval
Diagnosis*					
CNS/Spina bifida	57	8.8	1.3	1.3	(0.72, 2.5)
Chromosomal	45	6.9	**5.2**	**5.8**	**(2.9, 11.6)**
Cleft lip/palate	136	21.0	0.72	0.72	(0.45, 1.2)
Ear, Face, Neck	39	6.0	**2.8**	**2.9**	**(1.4, 5.8)**
Endocrine/Metabolic	8	1.2	*Insufficient Data*		
Eye	24	3.7	2.3	2.3	(0.93, 5.4)
Hearing Loss	47	7.3	1.5	1.2	(0.60, 2.4)
Heart	93	14.4	0.66	0.65	(0.38, 1.1)
Limb	29	4.5	1.1	1.2	(0.52, 2.6)
Urogenital/Renal	46	7.1	1.6	1.6	(0.82, 3.0)
Other	124	19.1	**2.5**	**2.5**	**(1.6, 4.0)**
Total**	648				
Syndrome					
Single Anomaly	222	52.1	1	1	*reference*
Multiple Anomalies	93	21.8	**2.3**	**2.4**	**(1.3, 4.4)**
Known Syndrome	111	26.1	**9.1**	**9.9**	**(5.6, 17.4)**
Total**	426				
Told Condition was Genetic					
No	215	48.5	1	1	*reference*
Yes	228	51.5	**3.1**	**3.2**	**(2.1, 5.1)**
Total¥	443				

* The reference category for each diagnosis category is those who do not have that specific dianosis, but have any other diagnosis.
** Total is more than 457 (the total sample size) because subjects may have more than one type of diagnosis and could be counted more than once.
¥ Totals are less than 457 (the total sample size) because of missing data.
† Adjusted for area of residence, education, race, and age of child.

Table 3: Having Been to Clinical Genetic Services

By diagnosis:

• Parents of children with a **chromosomal** or **ear/face/neck** condition were more likely to have been to a genetics clinic than children with any other condition (OR=5.8, 95% CI: 2.9, 11.6 and OR=2.9, 95% CI: 1.4, 5.8, respectively).

• Parents of children with a **heart** condition were less likely to have been to a genetics clinic than children with any other type of condition (OR=0.65, 95% CI: 0.38, 1.1).

By Syndrome:

• Parents of children with **multiple anomalies** or a **known syndrome** were more likely than children with a single anomaly to have been to a genetics clinic (OR=2.4, 95% CI: 1.3, 4.4 and OR=9.9, 95% CI: 5.6, 17.4, respectively).

By told condition was genetic:

• Parents who were **told their child's condition was genetic** were more likely than those who had not been told to have been to a genetics clinic (OR=3.2, 95% CI: 2.1, 5.1).

Figure 3 Frequency of having been told of genetic services and having been to a genetics clinic by category of exposure variable.

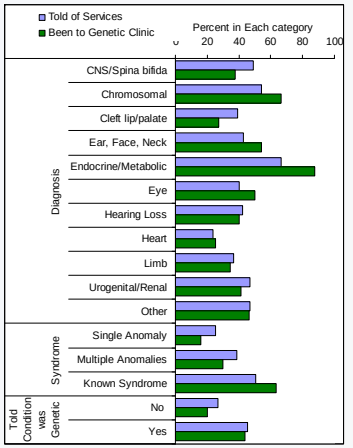

Figure 3:
Prevalence of being told of genetic services and having been to a genetics clinic was *highest* among those who:

- had an endocrine/metabolic diagnosis
- had a chromosomal diagnosis
- had a known syndrome
- were told the condition was genetic.

Discussion

Guidelines to help health care providers determine when to refer a family for genetic consultation have been published; referral is suggested for nearly any and all birth defects.[4] Nonetheless, we found that a small percentage of families had been told about services or gone to genetic clinics, despite their child's diagnosis.

Possible explanations:

- Families may be less likely to be told or go to services when conditions are less life-threatening, less obvious and/or amenable to surgical repair or other treatments.
- Families may be less likely to go to genetics clinics if the condition is more severe because of competing medical needs.

Regardless of the severity of the condition, parents may learn valuable information about their child's condition from clinical genetic services. Health care providers should be aware of genetic services and referral guidelines so that they can refer families to genetic consultations. Genetic clinics are an important place for genetic testing and counseling parents on inheritance, recurrence risk, and the chance for their child to have children with the same condition.

Limitations

• The categories of diagnoses are not exclusive – children with more than one defect may be in more than one category. We attempted to resolve this issue by additionally analyzing single, multiple anomalies, and syndromes. Diagnoses are self reported and we did not have information beyond diagnosis to assess or compare severity of conditions.

• These results are specific to the CSHCS population. We may not be able to generalize our findings. CSHCS eligibility is based on diagnosis, severity, and chronicity. Enrollment is voluntary and families may have a payment agreement for CSHCS coverage.

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