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 or involving genetic centers. 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. Other barriers to accessing genetic services may include lack of knowledge of genetic services, lack of awareness of risk, and location of services, among other issues.

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 on genetics, on having been told of genetic services and having been to a genetics clinic.

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 from birth date 2004-2008 and 850 families with enrollee 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 diagnoses type was analyzed as a dichotomous variable. Other covariates included: syndrome type, told condition was genetic, area of residence, education, age of child, and age of child (born from 1994-1998 or born from 2004-2008).

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

Results

Table 1: Survey responses were obtained from a total of 457 families, for a response rate of ~20%.

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 told condition was genetic:

• Parents of children with a known syndrome were more likely than parents of children with a single anomaly to be told of services (OR=2.3, 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 told about the condition to be told of genetic services (OR=2.9, 95% CI: 1.4, 4.0).

Table 3: Having been to Clinical Genetic Services

By diagnosis:

• Parents of children with a chromosomal or ear/face/neck condition were more likely than those parents with a genetic condition to have been to genetic clinic (OR=5.2, 95% CI: 2.3, 11.6 and OR=2.9, 95% CI: 1.4, 5.8, respectively). parents with a heart condition were less likely 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 those with a single anomaly to have been to a genetics clinic (OR=2.4, 95% CI: 1.3, 4.4 and OR=19, 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).

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. Nonetheless, we found that a small percentage of families had been told about genetic services or gone to genetic clinics, despite their child’s diagnosis.

Possible explanations:

• Patients 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.

• Patients may be less likely to go to genetic 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 their child with the same condition.

Limitations:

• The categories of diagnoses are not exclusive – children could have more than one defect may be in 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 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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References

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