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Public Perception of Birth Defects Terminology

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Abstract

BACKGROUND—‘Birth defect’ is a common phrase, yet concerns have been expressed that the word ‘defect’ carries a negative connotation. Our objective was to examine public perceptions of terms used to refer to birth defects.

METHODS—Four questions about terminology of birth defects were included in the U.S. nationally representative 2007 *HealthStyles* survey. Respondents answered questions about whether they or a family member were affected by birth defects (condition status), and which terms used to refer to birth defects they found preferable and which offensive. We further examined whether condition status, race/ethnicity, gender, income, geographical region, and education level impacted respondents’ term selection. Chi-square tests and multinomial logistic regression were performed using SAS 9.1.

RESULTS—‘Birth defects’ was most frequently selected as the first choice preferred term (35.4%), followed by 21.9% who selected ‘children with special needs.’ For respondents who said they themselves or a family member were affected by birth defects (11.5%), their responses differed statistically ($p = 0.0001$) from nonaffected respondents, but the leading choices were still ‘birth defects’ (28.5%) and ‘children with special needs’ (27.2%). Condition status, race/ethnicity, gender, income, and education level were all significant predictors of the respondents’ choice of a preferred term. When asked which phrases might be offensive, the top choices were ‘none of the phrases listed’ (37.0%), ‘adverse pregnancy outcomes’ (23.1%), and ‘birth defects’ (21.4%).

CONCLUSIONS—‘Birth defect’ was the preferred term; however, survey participants affected by birth defects responded less positively to the term. Continued dialogue about accepted and appropriate terminology is necessary.

Keywords

Birth defect; congenital anomaly; name; terminology

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INTRODUCTION

'Birth defect' is a common phrase, yet people with birth defects and their family members have voiced concerns that the word 'defect' might connote a negative perception and labeling. It is unclear how widespread this attitude is, and a better term (i.e., one less likely to carry any negative connotation) has yet to surface. Currently, no consistent terminology exists across state surveillance systems. A publicly accepted term will help unify efforts in birth defects surveillance and prevention, facilitate positive engagement with families of affected children, and promote more effective communication with the public.

Although birth defects terminology is discussed with parents of children with birth defects and within the professional community (National Institutes of Health, 2012), no previously published studies evaluating this issue have been conducted. However, similar evaluations of terminology have occurred in other fields, such as the introduction of the expression 'intellectual disability' to replace 'mental retardation' and 'physical disability' to replace 'handicapped' or 'crippled' (Danforth, 2002; Finlay and Lyons, 2005; Schalock et al., 2007). Our objective was to evaluate public perceptions of the connotation of terms used to signify birth defects. To avoid any confusion that the wide variety of terms used to describe these conditions (e.g., 'congenital anomaly,' 'birth disorder,' and 'congenital malformations') might engender, we have selected what we take to be the most widely recognized expression, namely 'birth defect,' for this study.

METHODS

This study used data from the 2007 *HealthStyles* survey, which is part of the *Styles 2007* consumer panel surveys conducted by Synovate, Inc. The panel consisted of 380,000 potential adult respondents in the United States who were eligible for the multistage *Styles 2007* surveys. In the first stage, the *ConsumerStyles* survey was sent to a stratified random sample of 20,000 potential adult respondents from the panel during May through June 2007, and responses were received from 11,758 individuals (response rate of 58.8%). A U.S. nationally representative sample was created by balancing the respondent pool by region, household income, population density, age, and household size. In the second stage conducted from July through August in 2007, half of the mail panel households who returned a *ConsumerStyles* survey were then invited to participate in the *HealthStyles* survey. A total of 6600 surveys were mailed, and responses were received from 4398 participants (response rate of 66.6%).

The Centers for Disease Control and Prevention (CDC) licensed the results of the *HealthStyles* mail panel surveys from Porter Novelli, and analysis of these results was exempt from institutional review board approval as personal identifiers were not included in the data provided to the CDC. The mail panel survey methodology has been shown to have very close agreement in level of response, trends over time, and pattern of demographic correlates to national probability sampling interview surveys (Pollard, 2002).

Four questions about personal or family history of birth defects and perception of birth defects terminology were included in the 2007 *HealthStyles* survey. Questions were

prefaced with: “The next few questions ask about problems present at birth that affect a baby and can result in physical or mental disabilities – even death. Examples might include cleft lip, club foot, Down syndrome, or spina bifida.” To assess their family history of birth defects, respondents were asked, “Were you or a family member born with a problem like the ones described above?” To assess preference of birth defect terminology, respondents were first asked, “Which of the following phrases would be a *good* way for describing the types of problems listed above?,” and then asked, “Which one of the following phrases would be your *first choice* for describing the types of problems listed above?” To assess negative perception of birth defect terminology, respondents were asked, “Which of the following phrases do you feel may be offensive for describing the types of problems listed above?” Nine phrases were presented as terminology choices for these questions (Table 1).

The survey asked for demographic information including age, gender, race/ethnicity, income level, education, and geographic region. We examined the first choice preferred phrase for describing birth defects by whether the respondents were affected or had a family member affected by a birth defect. We also examined the first choice preferred phrase for describing birth defects for demographic covariates, including respondent race/ethnicity (non-Hispanic white, non-Hispanic black, Hispanic, other), gender (male, female), annual family income (low <\$25,000, medium \$25,000–\$60,000, high >\$60,000), education (high school graduate or less, 1–3 years of college, college graduate or more), and geographic region (New England, Middle Atlantic, East North Central, West North Central, South Atlantic, East South Central, West South Central, Mountain, Pacific). Factors associated with term preference with p values < 0.05 in bivariate analyses were included in a multivariable model that used a multilevel outcome variable with the term ‘birth defects’ as the reference compared to preference for each of the other eight choices.

The *HealthStyles* survey data were post-stratified and weighted to the U.S. Census data on five demographic variables: age, gender, race/ethnicity, household size, and household income, thus creating a nationally representative sample accounting for nonresponse bias among participants. This weighting adjusted for overrepresentation or underrepresentation of categories within these demographic variables. All data analyses were conducted using SAS 9.1 (SAS Institute, Cary, NC).

RESULTS

A total of 4398 adults responded to the 2007 *Health-Styles* survey. Approximately 12% ($n = 507$ respondents) indicated that they themselves or a family member were born with a birth defect. There were more female than male respondents (52% vs 48%), and approximately 26% reported low income, 39% reported medium income, and 34% reported high income. Nearly 69% were white, 12% were black, 13% were Hispanic, and 7% were of another race. Almost 32% of respondents had a high school education or less, 36% completed 1 to 3 years of college, and 31% had an undergraduate or graduate degree.

The top three choices chosen by respondents (multiple selections allowed) as a good way to describe “problems present at birth that affect a baby and can result in physical or mental disabilities – even death” were ‘birth defects’ (49.6%), ‘children with special needs’

(46.1%), and ‘birth disorders’ (25.2%). When responders were only allowed to select their *first* choice preferred term for describing the types of problems listed, 35.4% of the respondents indicated ‘birth defects’ as their first choice, followed by 21.9% who selected ‘children with special needs,’ and almost 15% of respondents did not specify their preference (Table 1). For respondents who said they themselves or a family member were affected by birth defects (condition status), their *first* choice term selection differed statistically ($p = 0.0001$) from nonaffected respondents, but the leading choices were still ‘birth defects’ (28.5%) and ‘children with special needs’ (27.2%). Condition status significantly affected the first choice selection of preferred phrases ‘birth defects’ (p value = 0.005), ‘birth conditions’ (p value = 0.006), ‘congenital disorders’ (p value = 0.028), and ‘children with special needs’ (p value = 0.002).

Respondents indicated that their top choices for offensive phrases (multiple selections allowed) were ‘none of the phrases listed’ (37.0%), ‘adverse pregnancy outcomes’ (23.1%), and ‘birth defects’ (21.4%) (Table 2). When data were stratified by condition status, the leading choices were still ‘none of preferred phrases’ (38.1%) and ‘adverse pregnancy outcomes’ (24.9%).

In the multivariable multinomial (more than two-level outcome) logistic regression model, we included all factors that were significantly associated with a preferred terminology and used the preferred phrase ‘birth defects’ as a reference (Table 3). Controlling for the condition status, race/ethnicity, gender, family income, and education level attenuated the significance of geographic region (data not shown). Our final multivariable analysis revealed that the condition status, race/ethnicity, gender, income, and education level were significant predictors for the respondents’ choice in selecting a preferred terminology.

DISCUSSION

In this study, ‘birth defects’ was the term most frequently chosen by respondents; however, it was also a term considered offensive by over 20% of respondents. Additionally, respondents affected by birth defects were significantly less likely to choose the term ‘birth defect’ as a preferred term, but there was no statistical difference in potential for offense. Perceptions of terms were affected by respondents’ backgrounds, with preferred terminology differing depending on education level, race/ethnicity, gender, and family income. This may help us to understand the challenges in establishing a term with wide acceptance and adaptation.

‘Birth defects’ is the most commonly used phrase, but its use is not geographically or historically consistent. Population-based birth defects surveillance programs use various terms to describe themselves. Thirty-three of 47 state programs have the term ‘birth defect’ in their program titles, whereas the remaining programs use ‘children with special needs,’ ‘birth outcomes,’ ‘birth information,’ ‘birth conditions,’ ‘congenital defects,’ ‘adverse pregnancy outcomes,’ ‘congenital malformations,’ ‘congenital and inherited disorders,’ and ‘congenital anomalies’ (National Birth Defects Prevention Network, 2011). Likewise, International Classification of Diseases (ICD) codes have listed different terms as headings; ICD-10 (World Health Organization, 1994) uses the term ‘congenital malformations,

deformations, and chromosomal abnormalities’ whereas ICD-9 (National Center for Health Statistics, 1987) uses the term ‘congenital anomalies.’ The World Health Organization report on birth defects in 2010 uses the term ‘birth defects’ as its title and uses ‘congenital disorders’ interchangeably with ‘birth defects’ throughout the report (World Health Organization, 2010). Internationally, terms used for research organizations, such as International Clearinghouse for Birth Defects Surveillance and Research and European Registration of Congenital Anomalies, are diverse, including ‘birth defects,’ ‘congenital malformations,’ and ‘congenital anomalies.’ The March of Dimes, a national nonprofit organization, uses the expression ‘birth defects’ in its mission statement (marchforbabies.org); this term may be most recognized by the public.

There are no previously published studies regarding the public preferences for birth defects terminology. However, debate over and evolution in name change has occurred around terminology for intellectual disability (Schalock et al., 2007). Similar to those with intellectual disabilities, people with birth defects are a heterogeneous group with a variety of etiologies and broad variation in mortality, morbidity, and management. Investigations of the preferred terminology for intellectual disability were conducted among United Kingdom college students demonstrated that almost all terms tested were seen to have negative connotations (except ‘challenging behavior’ and ‘exceptional’); however, the labels that carried a negative connotation did so to varying degrees, with ‘mental retardation’ being highly negative whereas newer expressions like ‘learning difficulty’ and ‘learning disability’ were viewed as less negative (Hastings and Remington, 1993; Hastings et al., 1994).

Reasons for and against terminology change in the field of intellectual disability have previously been published and may be similar to reasons at play in the field of birth defects. Proponents for change in the terminology argued that stigma and negative connotations were connected with the expression ‘mental retardation.’ It was noted that previously accepted medical terms, such as ‘imbecile,’ ‘idiot,’ and ‘subnormal’ now seem utterly offensive but had to undergo a change to transition from acceptable to unacceptable (Danforth, 2002; Gelb, 2002). The label of ‘mental retardation’ was being increasingly rejected by persons with intellectual disability and their families (Luckasson and Reeve, 2001; Finlay and Lyons, 2005), and leaders in the field felt a necessity to find the most appropriate term possible (Luckasson and Reeve, 2001). However, others, including some parents of children affected by mental retardation and advocates, argued against a change in the terminology because they were concerned that the negative connotation would follow the new term, as this negative connotation was a byproduct of society, and a change in terminology would be nothing save another step in the “perpetual labeling cycle,” unless it were accompanied by a societal change (Hastings et al., 1993; Devlieger, 2003). Ultimately, the term ‘mental retardation’ was seen to have such a negative connotation that the terminology was changed to ensure individuals with intellectual disability obtain the necessary services and support to manage successfully in society.

If such a change in terminology was seen to be necessary in the field of birth defects, Luckasson and Reeve (2001) suggested the following questions to minimize negative impacts when introducing a novel terminology:

1. Does this term name this and nothing else?
2. Does this term provide consistent nomenclature?
3. Does this term facilitate communication?
4. Does this term incorporate current knowledge and is it likely to incorporate future knowledge?
5. Does this term meet the purposes for which it is being proposed?
6. Does this term contribute positively to the portrayal of people with [condition]?

Applying these questions in our examination of terminology in the field of birth defects, terms such as ‘children with special needs’ and ‘adverse pregnancy outcomes’ may indeed describe children with birth defects; however, they may also describe other conditions such as intellectual disabilities or prematurity, thus, these expressions are not specific in naming birth defects, which might lead to confusion. As we have stated above, terminology used across surveillance systems and organizations is inconsistent. Expressions must also be understood by a variety of stakeholders, including but not limited to patients, families, clinicians, researchers, and policymakers. Another possible consideration is to determine how well the term might be communicated and translated across languages and cultures. To address the positive portrayal of people with a disability, several authors (Hastings, 1994; Snell and Voorhees, 2006) emphasized the importance of involving those individuals with intellectual disability and their families in determining the preferred term. Historically, people with intellectual disabilities have not had any power to influence the labeling of their various disorders (Danforth, 2002; Luckasson, 2003). However, other minority groups have had success in proposing labels that went on to gain widespread recognition, such as renaming minority studies African American studies, Gay and Lesbian studies, and Women’s studies (Goode, 2002).

There are several limitations to our study. Previous studies have used a scale to quantify the amount of negative connotation associated with each term, which may be helpful in future research. By contrast, our study asked respondents to indicate whether term(s) were preferred or deemed offensive. Although efforts were undertaken to make the study generalizable to the national population, the mail panel survey sample might have underrepresented minorities and transitory households because completion of the survey required literacy in English and a mailing address. Likely, the participants were more interested in the topic of the survey (i.e., health behaviors) and have more free time than the nonparticipants. Additionally, we must consider those who did not specify a terminology preference; participants who indicated they were not affected or did not have a family member affected were significantly more likely to select “not specified” as an answer ($p = 0.004$), which suggests that this issue is of greater interest to those affected by birth defects.

A strength of this study lies in our ability to stratify the preferred terminology by whether the respondents themselves were affected or had a family member affected by a birth defect. Although we found a preference for ‘birth defect’ over other expressions used to refer to such conditions described, those with a personal or family history of birth defects favored the term less than the unaffected. Continued review of public perceptions of the terms used

to describe birth defects, with a particular interest in those affected, will allow us to examine changes in perceptions over time. If a change in terminology is deemed necessary, guidelines from the field of intellectual disability may prove helpful. Further dialogue about the most appropriate term is necessary to best serve people affected by birth defects.

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Table 1

First Choice Terminology Preference By Condition Status, *HealthStyles* 2007

Preferred term (first choice) ^a	Condition status ^b						
	Present		Absent		Total		
	No.	%	No.	%	No.	%	
Birth defects	144	28.5	1293	36.4	0.005	1437	35.4
Birth conditions	35	7.0	150	4.2	0.006	185	4.6
Birth disorders	39	7.6	286	8.1	0.746	325	8.0
Birth anomalies	26	5.2	155	4.4	0.412	181	4.5
Congenital anomalies	22	4.3	104	2.9	0.090	125	3.1
Congenital disorders	33	6.5	154	4.3	0.028	187	4.6
Congenital malformations	5	0.9	66	1.9	0.139	71	1.7
Adverse pregnancy outcomes	9	1.8	28	0.8	0.022	38	0.9
Children with special needs	138	27.2	750	21.1	0.002	888	21.9
No specified term	56	11.0	567	16.0	0.004	623	15.3
Total	507	12.5	3553	87.5	0.0001	4060 ^c	100.0

^a Responders were only allowed to select one choice.

^b Condition status refers to whether respondents were affected or had a family member affected by a birth defect.

^c Missing excluded from analysis.

Table 2

Terms That Might be Offensive by Condition Status, *HealthStyles* 2007

Offensive term ^a	Condition status ^b						
	Present		Absent		Total		
	No.	%	No.	%	No.	%	
Birth defects	121	23.9	748	21.1	0.153	869	21.4
Birth conditions	37	7.3	190	5.4	0.072	227	5.6
Birth disorders	67	13.2	424	11.9	0.431	491	12.1
Birth anomalies	64	12.6	373	10.5	0.144	437	10.8
Congenital anomalies	54	10.6	295	8.3	0.078	349	8.6
Congenital disorders	43	8.5	305	8.6	0.945	348	8.6
Congenital malformations	82	16.1	623	17.5	0.439	705	17.4
Adverse pregnancy outcomes	126	24.9	812	22.9	0.307	938	23.1
Children with special needs	61	12.1	297	8.4	0.006	358	8.8
None of these	193	38.1	1309	36.8	0.579	1502	37.0
Not specified ^c	15	3.0	323	9.1	0.0001	338	8.3

^a Responders were allowed to select all terms that apply.

^b Condition status refers to whether respondents were affected or had a family member affected by a birth defect.

^c Not specified included all missing responses.

Table 3

Predictors for Responders' First Choice in Selecting a Terminology Preference, *HealthStyles* 2007

Predictor	First Choice Terminology Preference																	
	Birth conditions		Birth disorders		Birth anomalies		Congenital anomalies		Congenital disorders		Congenital malformations		Adverse pregnancy outcomes		Children with special needs		Birth defects	
	n (%)	OR (95% CI)	n (%)	OR (95% CI)	n (%)	OR (95% CI)	n (%)	OR (95% CI)	n (%)	OR (95% CI)	n (%)	OR (95% CI)	n (%)	OR (95% CI)	n (%)	OR (95% CI)	n (%)	OR (95% CI)
Affected by birth defect (Condition Status) ^d	35 (19.0)	2.2 (1.4-3.3)	39 (11.9)	1.2 (0.8-1.8)	26 (14.5)	1.6 (1.0-2.6)	22 (17.4)	1.9 (1.2-3.2)	33 (17.7)	2.0 (1.3-3.0)	5 (6.7)	0.6 (0.3-1.7)	9 (24.8)	3.2 (1.5-7.0)	138 (15.5)	1.6 (1.3-2.1)	144 (10.1)	1.0
Non-Hispanic Black ^b	23 (11.6)	1.3 (0.9-1.9)	44 (12.7)	1.3 (0.9-1.8)	33 (16.5)	1.4 (1.0-2.1)	5 (3.4)	0.4 (0.2-0.9)	21 (10.1)	1.1 (0.7-1.7)	4 (4.7)	0.4 (0.2-1.0)	9 (19.1)	1.6 (0.8-3.2)	107 (11.1)	0.9 (0.8-1.2)	169 (11.3)	1.0
Hispanic ^b	21 (10.5)	1.0 (0.7-1.6)	58 (17.0)	1.6 (1.2-2.2)	19 (9.5)	0.9 (0.6-1.4)	13 (10.0)	1.2 (0.7-2.0)	24 (11.5)	1.1 (0.7-1.6)	8 (11.1)	1.2 (0.6-2.3)	10 (21.6)	1.9 (1.0-3.5)	124 (12.9)	1.1 (0.9-1.3)	167 (11.3)	1.0
Male ^c	93 (45.9)	0.8 (0.7-1.0)	169 (49.4)	0.9 (0.8-1.1)	81 (41.0)	0.7 (0.6-0.8)	60 (45.8)	0.9 (0.7-1.1)	110 (53.0)	1.0 (0.8-1.1)	34 (47.0)	0.9 (0.7-1.1)	21 (47.2)	0.8 (0.6-1.1)	413 (43.0)	0.8 (0.7-0.9)	722 (48.2)	1.0
Annual family income <\$25,000 ^d	42 (20.9)	0.8 (0.6-1.0)	63 (18.4)	0.6 (0.5-0.8)	36 (18.2)	0.8 (0.6-1.1)	26 (20.2)	1.1 (0.8-1.6)	24 (11.4)	0.5 (0.4-0.7)	9 (12.8)	0.7 (0.4-1.1)	11.3 (25.3)	0.6 (0.4-1.2)	290 (30.2)	1.1 (0.9-1.2)	359 (24.0)	1.0
Annual family income >\$60,000 ^d	74 (37.0)	1.0 (0.8-1.2)	144 (42.1)	1.2 (1.0-1.4)	69 (34.8)	0.9 (0.7-1.1)	65 (49.9)	1.0 (0.8-1.3)	113 (54.7)	1.5 (1.2-2.0)	35 (48.6)	1.1 (0.8-1.6)	22 (48.2)	1.6 (1.0-2.6)	348 (36.2)	0.9 (0.8-1.0)	652 (43.5)	1.0
High school education or less ^e	64 (32.2)	1.0 (0.8-1.3)	127 (37.8)	1.3 (1.1-1.5)	57 (29.6)	0.8 (0.6-1.1)	20 (15.3)	0.6 (0.4-0.8)	35 (17.3)	0.7 (0.6-1.0)	13 (18.8)	0.7 (0.5-1.1)	16 (35.1)	1.1 (0.7-1.8)	310 (32.6)	1.0 (0.9-1.1)	445 (30.1)	1.0
1-3 Years of college ^e	78 (40.0)	1.0 (0.8-1.2)	116 (34.5)	0.9 (0.8-1.1)	61 (31.6)	0.9 (0.7-1.1)	41 (31.3)	0.9 (0.7-1.2)	87 (43.0)	1.1 (0.9-1.4)	23 (32.8)	0.9 (0.6-1.3)	10 (22.9)	0.7 (0.4-1.1)	348 (36.7)	0.9 (0.8-1.1)	557 (37.8)	1.0

^aReference = Individual or family member not affected by birth defects.

^bReference = Non-Hispanic white.

^cReference = Female.

^dReference = \$25,000-\$60,000.

^eReference = College Graduate or more.

OR, odds ratio; CI, confidence interval.