Parents' perceptions of genetics services for congenital heart disease: the role of demographic, clinical, and psychological factors in determining service attendance

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Purpose: We sought to identify the demographic, clinical, and psychological factors associated with parents' attendance at clinical genetics services for congenital heart disease.

Methods: A survey assessing access to cardiac genetics services and a range of other variables was sent to the families of 213 children diagnosed with congenital heart disease between the years 2000 and 2009 at the Sydney Children's Hospital, Australia.

Results: Of the 114 respondents, 22% had accessed cardiac genetics services. Variables strongly associated with service attendance included presence of a syndrome associated with congenital heart disease (odds ratio = 17.93; $P < 0.001$) and antenatal diagnosis of congenital heart disease (odds ratio = 4.13; $P = 0.02$). Most participants (87%) perceived genetic factors as "quite" or "extremely important" in the development of congenital heart disease, and many (73%)

believed that receiving information about congenital heart disease and genetics was "quite" or "extremely important"; however, only 36% of participants could recall receiving information of this nature. Forty-two percent of parents reported current concerns about their child's health, and a substantial subset reported levels of depression (26%), anxiety (27%), and stress (32%) warranting clinical attention.

Conclusion: There is a strong desire among parents of children with congenital heart disease for greater information about the role of genetic factors; however, most families do not access cardiac genetics services and report limited recall of information gathered from other sources.

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Key Words: causal attributions; congenital heart disease; genetic counseling; genetics services; health services; psychological stress

INTRODUCTION

Congenital heart disease (CHD) is a spectrum of structural disorders affecting the heart. CHD affects 6–8 babies in every 1,000 live births, and about one-third of these infants require cardiac surgery in the first year of life. The burden of CHD is significant—it accounts for 20% of perinatal deaths and contributes more than 9,000 disability-adjusted life years to the total burden of disease in Australia. This makes CHD one of the top 10 contributors to burden of disease in children aged 0–14 years.¹

When a child is diagnosed with heart disease, almost all parents ask "Why has this happened to us?" Questions concerning causation are at the forefront of parents' minds, and the process of adapting to a child's diagnosis is psychologically difficult and complex.^{2,3} Although several risk factors associated with CHD have been well described, the complete etiological picture remains unclear, and the majority of CHD cases are considered sporadic in nature. In recent years, substantial progress has been

made into the underlying genetic mechanisms of abnormal heart formation.⁴ Molecular evaluations of genetic syndromes in which cardiac defects are a cardinal feature provide insights into genes associated with structural CHD and the pathways to abnormal heart development. For example, microdeletions in chromosome 22q11 which cause velocardiofacial syndrome⁵ are also associated with more than 50% of cases of interrupted aortic arch type B, 35% of cases of persistent truncus arteriosus, and 15% of cases of tetralogy of Fallot, as well as other cardiac lesions.⁴ Other genes in which mutations cause CHD include *NKX2-5*, *NKX2-6*, *MYH6*, and *TBX20* (e.g., refs. 6,7).

The identification of rare families with multiple affected members is another source of insight into genetic contributions to structural abnormalities of the heart. Most CHD is, however, not familial or syndromic but the result of multiple genetic mutations, or an interaction between single or multiple genetic mutations and the fetal environment.⁴ Population studies suggest that the genetic component of CHD is high, with

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heritability estimates ranging from 0.6 to 0.7^{8,9} and the averaged risk for siblings of affected children ranging between 2 and 5%. Risk for offspring is estimated at 3–5%¹⁰ but may be as high as 10–15% for certain lesions,¹¹ with the risk to offspring of affected females greater than that for affected males.¹² This relatively modest risk of recurrence within families is consistent with non-syndromic (or “common”) CHD.

As the molecular basis of heart formation comes into clearer focus, cardiac genetics services are likely to play an increasingly important clinical role. At present, genetics consultations purely for CHD are relatively uncommon. In day-to-day pediatric cardiology, generally only children with multiple congenital anomalies, a strong family history of CHD, abnormal karyotypes, or velocardiofacial syndrome proven by fluorescence in situ hybridization analysis are routinely referred for consultation.⁷ Outside these groups, the diagnosis of sporadic CHD does not usually trigger a genetics referral. Coinciding with this is a lack of research examining parents' beliefs about the role of genetic factors in CHD, as well as parents' interest in or desire for cardiac genetics services. Even less is known about the factors likely to contribute to genetics service attendance for CHD. Studies examining parents' use of genetics services for congenital abnormalities in general report relatively low but increasing utilization rates,^{13,14} with low utilization attributed to a range of factors, including: limited awareness of the availability of genetics services, a perceived lack of need for such services, negative views regarding pregnancy termination, and fears regarding the emotional consequences of genetics consultation, such as the arousal of guilt or blame.¹⁴ It is well established, however, that the presence of multiple anomalies or a syndrome in one's child, and death of a fetus or infant, increases the likelihood of referral to genetics services for congenital malformations in general.^{14–16}

The aims of this study were twofold: (i) to describe the beliefs and perceptions of parents of a child with CHD in regards to the role of genetic factors in CHD, as well as access to and utilization of cardiac genetics services and (ii) to identify the demographic, clinical, and psychological factors associated with attendance at genetics services for CHD. Based on the studies of parents' utilization of clinical genetics services for congenital abnormalities in general, it was hypothesized that attendance at cardiac genetics services would be low^{13,14} and would be associated with the presence of multiple anomalies or a syndrome in the child,^{14–16} increased maternal age,¹³ maternity care at a tertiary-level hospital,¹³ a family history of CHD, and greater perceived importance of the role of genetic factors in CHD. We also sought to examine the relationship between risk-related information-seeking style and genetics service attendance, hypothesizing that genetics service attendance would be associated with a general tendency in some parents to seek out or monitor for risk-related information.¹⁷

MATERIALS AND METHODS

Participants

Parents or guardians of a living child diagnosed with a congenital cardiac malformation between the years 2000 and

2009 and who had undergone cardiac surgery were identified for study participation via the databases of the Department of Cardiology at the Sydney Children's Hospital, Australia. Fully consented and contactable individuals were eligible for participation if they were aged older than 18 years and could complete the study questionnaire in English. To limit the burden on families, only one parent per family was invited to participate. The choice regarding who took part was left up to the individual families.

Procedure

The appropriate Human Research Ethics Committee gave approval for the study, and informed consent was obtained for all participants (SESIHHS approval number: 08/202). A study package comprising an invitation letter from the child's treating pediatric cardiologist, participant information sheet, questionnaire, and reply-paid envelope was mailed to all eligible families. Reminder letters and telephone calls were made, as appropriate, to parents who did not return the questionnaire within 1 month. Due to stipulation from the Human Research Ethics Committee, no further attempts were made to contact families after one telephone conversation and a second mail out. Study recruitment commenced in October 2009 and ceased in February 2010.

Measures

Based on a systematic review of the literature and expert consultation, a survey instrument was developed specifically for the study, comprised of both validated and purposely designed scales. Prior to administration, the questionnaire was pilot tested with a convenience sample of five parents of children with CHD, with modifications made according to parents' feedback. The self-administered questionnaire assessed sociodemographic and clinical characteristics, as well as a range of psychological variables. In addition, the questionnaire featured a discrete choice experiment to assess parents' preferences for various attributes of a hypothetical cardiac genetics consultation. Findings relating to this aspect of the study will be reported elsewhere.

1. *Demographic characteristics* (10 items): parental age, marital status, education, gross annual family income, residential location, nationality, language primarily spoken at home, number and age of children, and relationship to the child with CHD (e.g., mother, legal guardian). Income was categorized as above or below the National average based on the Australian Survey of Income and Housing 2007–2008.¹⁸
2. *Clinical characteristics* (13 items): Participants were asked to indicate the name of their child's cardiac abnormality; year and time (antenatal or postnatal) of cardiac diagnosis; presence of a chromosomal abnormality, syndrome, or other associated abnormality; number of cardiac surgical procedures; previous pregnancy termination or fetal death due to CHD; personal and family history of

CHD; and death of a child due to CHD. Participants were also asked to indicate current concerns they held in relation to any aspect of their child's health and development. Data were collected on the child's place of birth (public or private hospital) and the use of assisted reproductive technologies (e.g., in vitro fertilization). All self-reported cardiac and extra-cardiac diagnoses and cardiac treatments were verified via medical records, and surgical risk was categorized using the six-level Risk Adjustment for Congenital Heart Surgery system,¹⁹ with level 1 indicating the lowest and level 6 the highest surgical risk.

3. *Cardiac genetics service attendance* (2 items): Participants were asked to indicate whether they had ever been offered an appointment with a clinical geneticist to discuss their child's cardiac abnormality. Participants then indicated whether they or another family member had attended this appointment. Response options for both items were "yes", "no", and "unsure", with space provided to indicate reason(s) for nonattendance.
4. *Previously accessed or received information about genetics and CHD* (4 items): Participants indicated whether they had ever accessed or received information regarding possible genetic factors associated with CHD from sources other than a clinical geneticist or genetic counselor (see [Figure 2](#) for the full list of options). Participants were also asked to indicate the timing of information receipt (i.e., during pregnancy or postpartum) and how satisfied they were with the quality and quantity of information provided, with options ranging from 1 ("not at all satisfied") to 5 ("extremely satisfied"). Finally, participants rated the perceived importance of receiving information about genetics and CHD on a 5-point Likert scale ranging from 1 ("not at all important") to 5 ("extremely important").
5. *Causal attributions for CHD* were assessed via 14 items based on our previous qualitative research.² Participants rated the perceived importance of each item ([Figure 1](#)) as a cause of CHD on a 5-point Likert scale ranging from 1 ("not at all important") to 5 ("extremely important"). Several strategies were employed in an effort to minimize potential bias toward a genetic model of CHD: (i) the study title and questionnaire cover did not mention the researchers' interest in genetics; (ii) causal attributions were elicited before items about attendance at genetics services or receipt of genetics-related information; and (iii) "genetic factors" were listed ninth in the list of possible factors associated with CHD. Participants were also offered an opportunity to indicate and rate additional possible causes they identified spontaneously.
6. *Depression, anxiety, and stress* (21 items): The Depression, Anxiety and Stress Scale (DASS-21) is a widely used and validated measure of depression, anxiety, and stress experienced in the past 7 days, for which normative data in nonclinical Australian adult samples are available.²⁰ Response options range from 0 ("did not apply to me at all") to 3 ("applied to me very much or most of the time"),

and clinical cutoff scores are available for each of the three subscales.²¹ In the present sample, the internal consistency was 0.87 for each of the depression, anxiety, and stress subscales.

7. *Posttraumatic stress symptoms* (22 items) associated with CHD diagnosis and treatment were assessed using the Impact of Events Scale-Revised (IES-R).²² Participants rated the frequency and severity of posttraumatic stress symptoms in the preceding week along three dimensions (intrusion, avoidance, and hyperarousal) using a 5-point frequency scale. Internal consistency for the IES-R total score was 0.96. Elevated and probable posttraumatic stress levels were defined as IES-R total scores above 24 and 32, respectively.^{23,24}
8. *Information-seeking style* (32 items): According to the monitoring process model, the way people cope under situations of threat can be classified as either cognitive confrontation ("high monitoring") or cognitive avoidance ("low monitoring").²⁵ Individuals are characterized as either "high monitors" or "low monitors" depending on how they select, encode, and manage risk-related information, as well as how they respond to this information. Under conditions of threat, people with a tendency toward high monitoring typically prefer detailed information, tend to actively seek out and scan for threatening cues, and experience increased anxiety in relation to threats.²⁶ High monitors also typically report feeling dissatisfied with the information provided in routine medical care,²⁶ and for these people, monitoring is motivated by the need to minimize uncertainty and gain control over the threat. By contrast, low monitors tend to avoid detailed threat-related information in order to

Figure 1 Mean perceived importance scores for each of the 14 factors presented to participants as possibly, or possibly not, associated with the development of congenital heart disease.

minimize anxiety. In this study, information-seeking style was assessed using the Miller Behavioral Style Scale.²⁵ Respondents were asked to imagine four hypothetical stress-invoking scenarios of a largely uncontrollable nature. Each scenario was followed by eight responses indicative of either *high* or *low* information-seeking (“monitoring”) style. For the analysis, participants were categorized as high or low monitors on the basis of how they anticipated their responses to these threat-related cues, using a median split.²⁶

Data analysis

Data were analyzed using the IBM Statistical Package for the Social Sciences (SPSS) 20.0 (IBM, Armonk, NY). Descriptive statistics were used to assess the demographic and clinical characteristics of the sample, as well as genetics-related attitudes, beliefs, and behaviors. Spearman's correlations were used to investigate associations between psychological distress scores and diagnostic variables (i.e., time since cardiac diagnosis and surgical risk). For all bivariate analyses, the outcome variable (cardiac genetics service attendance) was treated as a dichotomous variable, as no participants reported being “unsure” about attendance. Fisher's exact test was used to examine the associations between categorical independent variables with two levels and cardiac genetics service attendance.²⁷ Independent samples *t*-tests or Mann–Whitney *U* tests were used, as appropriate, for continuous predictor variables.

Binary logistic regression analysis was used to examine the relationship between cardiac genetics service attendance and independent predictor variables. Two approaches to this analysis were taken. First, due to the limited number of participants who had attended cardiac genetics services, a forward modeling strategy was used, starting with the predictor variable with the lowest *P* value at the bivariate level and adding each variable sequentially, based on its *P* value. Models were limited to two predictor variables to avoid the problematic results of logistic models with fewer than 10 events (i.e., participants) per variable.²⁸ Second, a progressive, backward elimination modeling strategy, which included all predictor variables significant at the bivariate level, was also tested. Each approach resulted in the same final model. To assess multicollinearity, the variance inflation factor for each predictor variable in the model was checked, and no problems were detected.

RESULTS

Response rates

Based on the eligibility criteria for the study, 257 families were identified and approached for study participation. Of these, 44 families were not contactable (due to incorrect address or disconnected telephone line), 21 families declined participation, and 78 families did not return the study questionnaire, resulting in 114 completed questionnaires and yielding a participation rate of 53.5% among eligible, contactable families (114/213).

Demographic, clinical, and psychological characteristics of the sample (*N* = 114)

Overall, the majority of the sample were mothers (79.8%), the mean age of the sample was 36.7 years (*SD* = 5.7), the median time since cardiac diagnosis was 3 years, and less than a quarter of parents (21.9%) had received their child's diagnosis in the antenatal period. Participants reported a range of current concerns about their child's health and development, including concerns about feeding and weight gain (*n* = 16, 14%), global developmental delay (*n* = 10, 9%), emotional and social development (*n* = 8, 7%), language and speech development (*n* = 8, 7%), ongoing medical issues (*n* = 8, 7%), fine and gross motor skills (*n* = 7, 6%), intellectual functioning (*n* = 6, 5%), and future quality of life (*n* = 6, 5%). Three parents (2.6%) had a child who had died due to CHD, and no participants reported previous pregnancy termination or fetal death due to CHD (Table 1).

Mean scores for depression, anxiety, and stress were higher than norms derived from the general Australian adult population (Table 2).²⁰ Depression, anxiety, and stress scores potentially warranting clinical intervention were reported by 26, 27, and 32% of the sample, respectively. In terms of posttraumatic stress symptoms, 49 participants (43%) indicated that they were “extremely” or “quite a bit” affected by one or more symptoms in the past week. Items with the highest mean scores were “any reminder brought back feelings about it” (mean = 1.60; *SD* = 1.26), “pictures about it popped into my mind” (mean = 0.97; *SD* = 1.21), and “I had waves of strong feelings about it” (mean = 0.89; *SD* = 1.21). Elevated posttraumatic stress was identified in 16.7% of parents, and 14% of parents reported posttraumatic stress symptoms consistent with the need for referral to clinical psychology services. Both general ($r_s = -0.20$; $P = 0.04$) and traumatic ($r_s = -0.20$; $P = 0.04$) stress scores were negatively correlated with time since diagnosis; thus, the greater the time since diagnosis, the lower the stress reported by parents. No other associations between psychological distress and time since diagnosis or surgical risk were found.

Causal attributions for CHD

Overall, participants perceived genetics to be the most important factor associated with the development of CHD, with 97 participants (86.7%) rating genetic factors as “quite” or “extremely important” and only 2 participants (1.8%) rating genetics as “not at all important” as a cause of CHD (Figure 1).

Access to, and attitudes toward, information about genetics and CHD

Participants were asked to indicate the importance of receiving information about CHD and possible associated genetic factors. The mean importance score for the sample was 4.0 (*SD* = 1.1), indicating that on average, participants perceived information on CHD and genetics as “quite important”. In terms of the distribution of scores, 4 participants (3.5%; all mothers) perceived genetics-related information as “not at all” important, 8 participants (7%) as “slightly” important, 19 participants (16.7%) as “moderately” important, 36 participants

Table 1 Demographic and clinical characteristics of the sample (*N* = 114)

Variable	Level	<i>n</i>	Percentage
Demographic variables			
Participant relationship to child	Mother	91	79.8
	Father	20	17.5
	Other (aunt)	1	0.9
Age (years)	18–19	0	0
	20–29	12	10.5
	30–39	68	59.6
	40–49	33	28.9
	>50	1	0.9
	Mean age (SD)	36.7	(5.7)
Marital status	Married	98	86.0
	Unmarried	16	14.0
Residence at time of childbirth	Metropolitan New South Wales	80	70.2
	Regional or rural New South Wales	31	27.2
	Overseas	3	2.6
Education	University degree	55	48.2
	No university degree	58	50.9
Gross family income ^a	Below national average	48	42.1
	Above national average	50	43.9
Country of birth	Australia	83	72.8
	Other	31	27.2
Language most frequently spoken at home	English	101	88.6
	Other	13	11.4
Number of children	One	27	23.7
	Two	45	39.5
	Three	28	24.6
	Four or more	14	12.3
Clinical variables			
Time since cardiac diagnosis	Within last 12 months	8	7.0
	1–2 years	21	18.4
	2–3 years	30	26.3
	3–4 years	20	17.5
	>4 years	35	30.7
Time of cardiac diagnosis	Antenatal	25	21.9
	Postnatal	89	78.1

Table 1 Continued

Variable	Level	<i>n</i>	Percentage
Cardiac surgical intervention	Mean number of surgeries per child (SD)	1.8	(1.4)
Cardiac surgical risk score (RACHS)	1–2	66	57.9
	3–4	42	36.8
	5–6	2	1.8
Presence of chromosomal abnormality or syndrome ^b	Yes	24	21.1
	No	74	64.9
	Unsure	15	13.2
Current concerns about child's development	Yes	47	41.2
	No	66	57.9
Type of maternity hospital	Public	82	71.9
	Private	32	28.1
Use of IVF or assisted reproductive therapy	Yes	9	7.9
	No	103	90.4
Self-reported personal history of CHD ^c	Yes	4	3.5
	No	110	96.5
Self-reported family history of CHD ^c	Yes	17	14.9
	No	90	78.9
	Unsure	7	6.1
Other living or deceased children with CHD ^c	Yes	7	6.1
	No	106	93.0

CHD, congenital heart disease; IVF, in vitro fertilization; RACHS, Risk Adjustment for Congenital Heart Surgery.

^a16 participants (14%) did not respond to this item. ^bSelf-reported data.

^cParticipants were also asked to provide the name of the cardiac abnormality. These were examined by the research team and cardiac conditions that were not congenital were recorded accordingly.

(31.6%) as “quite” important, and 47 participants (41.2%) as “extremely” important. Forty-one participants (36.3%) indicated that they had previously accessed or received information about CHD and genetic factors. For these participants, the most common sources of information about genetics were their pediatric cardiologist (*n* = 30, 73.2%), the Internet (*n* = 23, 56.1%), and paper-based materials such as books or pamphlets (*n* = 14, 34.1%; **Figure 2**). Twelve participants had accessed or received information about CHD and genetics during pregnancy (29.3%). The mean satisfaction score for those participants who had accessed genetics-related information was 3.2 (SD = 1.2), indicating that on average, participants were “fairly satisfied” with the information received. Of those who had accessed information,

Table 2 Psychological characteristics of the study sample, as compared with Australian normative data where available
Depression, Anxiety, and Stress Scales (DASS-21)^a

Subscale	Study sample	Australian norms ^b	Clinical classifications	<i>n</i>	Percentage
	Mean (SD)	Mean (SD)			
Depression	5.40 (7.21)	2.21 (3.60)	Normal	87	76.3
			Mild	13	11.4
			Moderate	6	5.3
			Severe	3	2.6
			Extremely severe	4	3.5
Anxiety	4.85 (7.74)	1.48 (2.60)	Normal	86	75.4
			Mild	6	5.3
			Moderate	10	8.8
			Severe	3	2.6
			Extremely severe	8	7.0
Stress	11.01 (9.13)	3.79 (4.10)	Normal	81	71.1
			Mild	9	7.9
			Moderate	14	12.3
			Severe	8	7.0
			Extremely severe	1	0.9

Impact of Events Scale–Revised (IES-R)^a

Post-traumatic stress	Mean (SD)	Range	Clinical indication for IES-R total score ^c		
				<i>n</i>	Percentage
Intrusion	6.57 (6.80)	0–28	Elevated traumatic stress	19	16.7
Avoidance	4.92 (6.64)	0–26	Probable traumatic stress	16	14.0
Hyperarousal	2.66 (4.60)	0–20			
IES-R total score	14.15 (16.93)	0–69			

Miller Behavioral Style Scale (MBSS)

Scale	Mean (SD)	Median	Category	<i>n</i>	Percentage
Information-seeking style	8.58 (3.34)	8.00	High monitoring	56	49.1
			Low monitoring	58	50.9

^a*N* = 113 due to missing data for one participant. ^bNorms for the general Australian adult population sample aged 25–90 years. ²¹ ^cElevated post-traumatic stress was defined as an IES-R total score above 24, and probable post-traumatic stress was defined as an IES-R total score above 32.

4 participants (9.8%; 3 mothers and 1 father) were “not at all” satisfied, 9 participants (22%) were “somewhat” satisfied, 9 participants (22%) were “fairly” satisfied, 15 participants (36.6%) were “quite” satisfied, and 4 participants (9.8%) were “extremely” satisfied.

Attendance at clinical genetics services for CHD

Only 28 participants (24.6%) could recall being offered a genetics consultation, and of these, 25 participants (89%) had attended the consultation. Thus, in this cross-sectional study of Australian parents of children with CHD requiring surgical intervention, the overall attendance rate at genetics services was 22% (25/114). Among those who were referred to a clinical geneticist, reasons for nonattendance included feeling “too scared about what it might reveal” and having to wait for an available appointment. Variables associated with genetics service attendance at the bivariate level are presented in [Table 3](#). In the final logistic regression model, presence of a syndromal condition increased the odds of uptake of genetics services by 18 times (odds ratio = 17.93; 95% confidence interval: 5.73–56.07; *P* < 0.001), whereas antenatal diagnosis of CHD increased the odds of genetics service attendance by over four times (odds

ratio = 4.13; 95% confidence interval: 1.23–13.91; *P* = 0.02). Thus, both factors were very strong predictors of genetics service attendance in this sample.

DISCUSSION

Very little is known about parents' interest in or desire for genetics services for CHD, and even less is known about the factors likely to contribute to cardiac genetics service attendance. In this study, we found that despite high levels of interest in clinical genetics services and a strong desire for more information about the role of genetic factors in CHD, only 25% of parents reported awareness of the availability of cardiac genetics services, and overall, only 22% reported service attendance. Moreover, only 36% of the total sample could recall accessing or receiving information about CHD and genetic factors from other sources, such as their child's cardiologist. In comparison with previously reported data on Australian parents' use of genetics services after the diagnosis of a congenital abnormality in their child, where rates of uptake of genetic counseling are as high as 60%,¹⁴ awareness of and engagement with genetics services for CHD seems low. This is particularly interesting in light of data that suggest that the timing of

Table 3 Bivariate analysis of demographic, medical, and psychological variables associated with parents' attendance at genetics services for CHD (*N* = 114)

Variable	Genetics attendees	Nonattendees	Test statistics	
	(<i>n</i> = 25) Percentage	(<i>n</i> = 89) Percentage		
Education	54% university degree	47	1	0.54
Gross family income	63% above average	48	1	0.24
Marital status	76% married	89	1	0.11
Type of maternity hospital	76% public	71	1	0.61
Time of CHD diagnosis	48% antenatal diagnosis	15	1	<0.001
Syndrome identified or suspected ^a	76% with syndrome	18	1	<0.001
Concerns about child's development	68% had concerns	34	1	0.003
Contemplating another pregnancy	44% in contemplation	51	1	0.56
Personal or family history of CHD	12% with CHD history	16	1	0.76
Other children with CHD ^b	8% affected children	6	1	0.65
Previously accessed genetics information ^c	60% accessed information	30	1	0.009
	Mean (SD)	Mean (SD)	<i>t</i>	<i>df</i>
Parental age	37.68 (4.86)	36.39 (5.93)	−1.00	112
Time since cardiac diagnosis	3.20 (2.29)	2.84 (2.04)	−0.75	112
	Mean (SD)	Mean (SD)	<i>Z</i>	<i>P</i>
Belief in genetic model of CHD ^d	4.32 (0.90)	4.34 (0.94)	−0.27	0.79
Perceived importance of genetics information ^d	4.28 (0.79)	3.92 (1.15)	1.16	0.25
Post-traumatic stress response ^d	18.04 (20.18)	13.05 (15.85)	1.38	0.17
Information-seeking style ^d	8.64 (3.32)	8.56 (3.36)	0.12	0.90

CHD, congenital heart disease.

^aSelf-reported data, with responses examined by a clinical geneticist (E.K.). ^bVariable combines other living or deceased children affected by CHD. ^cFrom sources other than genetics professionals. ^dMann–Whitney *U* test was used. *df* denotes degrees of freedom. **Figure 2** Sources, other than the clinical genetics team, from which participants reported accessing or receiving information about genetic factors associated with congenital heart disease.

genetics consultations may bear importance, with parents perceiving access to genetics services as most useful 6 or more months after initial diagnosis.²⁹ All parents in this study were surveyed at least 12 months after initial cardiac diagnosis, and all children were diagnosed at a tertiary pediatric hospital with an established genetics service.

As hypothesized, the presence of a chromosomal abnormality or syndrome in one's child was the strongest predictor of attendance at cardiac genetics services.^{14,16} Although this appears to indicate a largely "common sense" view of cardiac genetics service utilization among parents, 39% of parents of a child with multiple physical anomalies or a syndrome had *not* attended a genetics service. These data flag a need to understand, in both practical and psychological terms, the potential "gaps" between parents' beliefs about their child's medical condition, their awareness of available services, and actual health behaviors. The lack of parental awareness of genetics services observed in this study is not uncommon,^{14,29} however, the specific barriers to service use are unclear. Parental factors that may potentially account for low service use include misconceptions about the purpose of genetics services and fears regarding the emotional consequences of genetics consultation.¹⁴ It is possible that practical (e.g., availability, cost)³⁰ and clinician-related factors (e.g., limited doctor–patient communication about genetics services)¹⁴ may also play a role. Prospective cohort studies are a means of better understanding the dynamics of genetics service use in this setting.

Antenatal diagnosis of CHD was also strongly associated with cardiac genetics service attendance, with 48% of participants who had received an antenatal diagnosis attending a genetics consultation. Unlike most other major organ abnormalities, almost half of all infants requiring cardiac surgery during infancy are diagnosed antenatally.³¹ One of the major advantages of antenatal diagnosis is that potentially unstable newborns with CHD can have planned delivery close to specialized pediatric cardiac services, reducing morbidity and, in some lesions, measurably improving the chances of survival.^{32,33} Antenatal diagnosis also presents an opportunity to provide counseling to families regarding the nature of their baby's abnormality, the expected medical course and prognosis, and often leads to multiple visits with health-care providers before birth, including pediatric cardiologists, cardiothoracic surgeons, and clinical geneticists. Recent research by our group, however, shows that a substantial subset of mothers and fathers who receive their baby's cardiac diagnosis antenatally experience persistent symptoms of severe traumatic stress.²

In terms of psychological functioning, a substantial subset of parents in the present sample reported psychological stress, including anxiety and posttraumatic stress, indicative of the need for referral to clinical psychology services. Indeed, mean scores for depression and stress were *more than double* documented Australian norms, and the mean anxiety score was *more than three times* that expected in the general Australian adult population.³⁴ These results are striking and signal the need for greater attention to be directed toward the prevention and treatment of severe emotional distress in families of children with CHD, particularly given the potential for some parents to experience persistent symptoms over many years.

Genetic factors played a role in almost all participants' causal attributions for CHD. What this means in terms of parents' *understanding* of the role of genetic factors in CHD requires further inquiry. In the general community, concepts such as genetics and heritability are often perceived as equivalent,³⁵ and this can lead to misconceptions about disease onset, inheritance, and risk of recurrence, as well as intense emotional reactions such as "transmission guilt" among parents and "survivor guilt" among siblings. Previous research suggests that people generally tend to perceive the causes of many diseases as multifactorial, due to genetic factors in combination with a variety of environmental, behavioral, and psychosocial factors.³⁶ Learning more about the potential role of a range of factors, including genetics, may serve to alleviate powerful feelings of guilt or blame experienced by some parents.³⁵

Conversely, some parents may hold causal beliefs suggestive of an "all or nothing" or binary principle of causality. The discounting principle by Kelley³⁷ states that, if people perceive a number of different causes as sufficient to produce an effect, when evidence for the presence of one cause is given, the role of other plausible causes will be discounted. An alternative explanation is that attributing CHD entirely to genetic factors may serve to reduce or remove a sense of randomness about the development of heart disease in one's child.³⁸ It is also possible,

however, that parents may misperceive, misunderstand, or distort genetics information, thus altering its intended meaning and its emotional consequences. Qualitative methodologies have the capacity to provide rich data on these phenomena, and studies of this nature may be helpful in elucidating the diversity and meaning of causal beliefs held by children with CHD, their parents, and other family members.

Interpretation of the present findings must take into account a number of limitations. Due to the cross-sectional study design, the causal direction of associations is unclear. Prospective studies are needed to better understand the potential changes in parents' motivations for cardiac genetics service attendance over time. Self-report is subject to the possibility of response and recall bias, and the relatively small sample may not have yielded sufficient power to detect important effects. Moreover, given the high proportion of mothers and Australian-born participants with a tertiary education, the sample may not be representative of all parents of a child with CHD. Despite these limitations, the findings highlight a range of clinically relevant issues.

RECOMMENDATIONS

There is strong interest among parents of children with CHD for greater information about the role of causal factors, including genetics. Most families, however, do not access cardiac genetics services and report limited recall of, or satisfaction with, information about etiological factors. Given the central role that pediatric cardiologists and general practitioners play in communicating with families about the potential causes of CHD, as well as guiding referrals to genetics services, it seems imperative that we develop a better understanding of the ways in which clinicians view cardiac genetics services and parents' desire for genetics-related information. Little is known about doctor-parent communication about the causes of CHD in general, nor the provision of information directly relevant to genetics. The results of this study support the incorporation of genetics-related information in cardiology consultations as a means of introducing this area to parents and facilitating referral to genetics services, if indicated and desired. Although current knowledge does not suggest that a referral to cardiac genetics services should be standard for all families, well-established factors that prompt referral include the presence of multiple congenital anomalies, family history of CHD, abnormal karyotypes, or velocardiofacial syndrome. Other relevant factors may include the nature and severity of the cardiac abnormality, physician or parental perceived likelihood of recurrence, and parental desire for more information. Given that parents may experience difficulties recalling this information, particularly if it is provided at the time of diagnosis or medical intervention,² it may be beneficial to address questions relating to CHD causation more than once, as parents' emotional receptiveness evolves over time. It may also be beneficial to provide etiological information in multiple formats, including print- and web-based resources (e.g., <http://www.heartcentreforchildren.com.au/how-did-this-happen-.html>).

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DISCLOSURE

The authors declare no conflict of interest.

REFERENCES

- Begg S, Vos T, Barker B, Stevenson C, Stanley L, Lopez A. *The Burden of Disease and Injury in Australia 2003*. AIHM: Canberra, 2007.
- Kasparian N. When hearts and minds meet: a psychological perspective on fetal cardiac diagnosis. *Infant Mental Health J* 2010;32:104.
- Majnemer A, Limperopoulos C, Shevell M, Rohlicek C, Rosenblatt B, Tchervenkov C. Health and well-being of children with congenital cardiac malformations, and their families, following open-heart surgery. *Cardiol Young* 2006;16:157–164.
- Sohr-Preston SL, Scaramella LV. Implications of timing of maternal depressive symptoms for early cognitive and language development. *Clin Child Fam Psychol Rev* 2006;9:65–83.
- Yagi H, Furutani Y, Hamada H, et al. Role of TBX1 in human del22q11.2 syndrome. *Lancet* 2003;362:1366–1373.
- Kirk EP, Sunde M, Costa MW, et al. Mutations in cardiac T-box factor gene TBX20 are associated with diverse cardiac pathologies, including defects of septation and valvulogenesis and cardiomyopathy. *Am J Hum Genet* 2007;81:280–291.
- Pierpont ME, Basson CT, Benson DW Jr, et al.; American Heart Association Congenital Cardiac Defects Committee, Council on Cardiovascular Disease in the Young. Genetic basis for congenital heart defects: current knowledge: a scientific statement from the American Heart Association Congenital Cardiac Defects Committee, Council on Cardiovascular Disease in the Young: endorsed by the American Academy of Pediatrics. *Circulation* 2007;115:3015–3038.
- Burn J, Goodship J. Congenital heart disease. In: Rimoin DL, Connor JM, Pyeritz RE, Korf BR (eds). *Emery and Rimoin's Principles and Practice of Medical Genetics*, 4th edn. Churchill Livingstone: London, 2002:1239–1326.
- Williamson EM. A family study of atrial septal defect. *J Med Genet* 1969;6:255–265.
- Burn J, Brennan P, Little J, et al. Recurrence risks in offspring of adults with major heart defects: results from first cohort of British collaborative study. *Lancet* 1998;351:311–316.
- McBride KL, Pignatelli R, Lewin M, et al. Inheritance analysis of congenital left ventricular outflow tract obstruction malformations: segregation, multiplex relative risk, and heritability. *Am J Med Genet A* 2005;134A:180–186.
- Nora JJ, Nora AH. Maternal transmission of congenital heart diseases: new recurrence risk figures and the questions of cytoplasmic inheritance and vulnerability to teratogens. *Am J Cardiol* 1987;59:459–463.
- Glynn A, Collins V, Halliday J. Utilization of genetic counseling after diagnosis of a birth defect—trends over time and variables associated with utilization. *Genet Med* 2009;11:287–293.
- Glynn A, Saya S, Halliday J. Use and non-use of genetic counseling after diagnosis of a birth defect. *Am J Med Genet A* 2012;158A:559–566.
- Sikkens EH, de Waale HE, Reefhuis J, van Tintelen JP, van Essen AJ. Referral for genetic counseling after the birth of a child with a congenital anomaly in the Northern Netherlands. *Am J Med Genet* 2002;112:133–137.
- Forrester MB, Merz RD. Genetic counseling utilization by families with offspring affected by birth defects, Hawaii, 1986–2003. *Am J Med Genet* 2007;143A:1045–1052.
- Shiloh S, Ben-Sinai R, Keinan G. Effects of controllability, predictability, and information-seeking style on interest in predictive genetic testing. *Pers Soc Psychol Bull* 1999;25:1187–1195.
- Australian Bureau of Statistics. Survey of Income and Housing. 2007–08 <http://www.abs.gov.au/ausstats/abs@nsf/1020492cfcd63696ca2568a1002477b5/5f4bb49c975c64c9ca256d6b00827adb!OpenDocument>. Accessed 12 February 2010.
- Jenkins KJ, Gauvreau K, Newburger JW, Spray TL, Moller JH, Iezzoni LI. Consensus-based method for risk adjustment for surgery for congenital heart disease. *J Thorac Cardiovasc Surg* 2002;123:110–118.
- Crawford J, Cayley C, Lovibond PF, Wilson PH, Hartley C. Percentile norms and accompanying interval estimates from an Australian general adult population sample for self-report mood scales (BAI, BDI, CRS-D, CES-D, DASS, DASS-21, STAI-X, STAI-Y, SRDS, and SRAS). *Aust Psychol* 2011;46:3–14.
- Lovibond PF, Lovibond SH. The structure of negative emotional states: comparison of the Depression Anxiety Stress Scales (DASS) with the Beck Depression and Anxiety Inventories. *Behav Res Ther* 1995;33:335–343.
- Weiss D, Marmar C. The impact of event scale-revised. *Assessing Psychological Trauma and PTSD*. 1997:399–411.
- Boer KR, van Ruler O, van Emmerik AA, et al.; Dutch Peritonitis Study Group. Factors associated with posttraumatic stress symptoms in a prospective cohort of patients after abdominal sepsis: a nomogram. *Intensive Care Med* 2008;34:664–674.
- Creamer M, Bell R, Failla S. Psychometric properties of the Impact of Event Scale - Revised. *Behav Res Ther* 2003;41:1489–1496.
- Miller SM. Monitoring and blunting: validation of a questionnaire to assess styles of information seeking under threat. *J Pers Soc Psychol* 1987;52:345–353.
- Miller SM. Monitoring and blunting of threatening information: cognitive interference and facilitation in the coping process. In: Sarason I, Pierce G, Sarason B (eds). *Cognitive Interference: Theories, Methods, and Findings*. Lawrence Erlbaum Associates: Hillsdale, NJ, England, 1996:175–190.
- Howell DC. *Statistical Methods for Psychology*. Duxbury Press: Belmont, CA, 1987.
- Peduzzi P, Concato J, Kemper E, Holford TR, Feinstein AR. A simulation study of the number of events per variable in logistic regression analysis. *J Clin Epidemiol* 1996;49:1373–1379.
- Collins V, Halliday J, Kahler S, Williamson R. Parents' experiences with genetic counseling after the birth of a baby with a genetic disorder: an exploratory study. *J Genet Couns* 2001;10:53–72.
- Kasparian NA, Meiser B, Butow PN, Simpson JM, Mann GJ. Genetic testing for melanoma risk: a prospective cohort study of uptake and outcomes among Australian families. *Genet Med* 2009;11:265–278.
- Sholler GF, Kasparian NA, Pye VE, Cole AD, Winlaw DS. Fetal and post-natal diagnosis of major congenital heart disease: implications for medical and psychological care in the current era. *J Paediatr Child Health* 2011;47:717–722.
- Sivarajan V, Penny DJ, Filan P, Brizard C, Shekerdemian LS. Impact of antenatal diagnosis of hypoplastic left heart syndrome on the clinical presentation and surgical outcomes: the Australian experience. *J Paediatr Child Health* 2009;45:112–117.
- Blyth M, Howe D, Gnanapragasam J, Wellesley D. The hidden mortality of transposition of the great arteries and survival advantage provided by prenatal diagnosis. *BJOG* 2008;115:1096–1100.
- Lovibond SH, Lovibond PF. *Manual for the Depression Anxiety Stress Scales*. Psychology Foundation: Sydney, 1995.
- Edhborg M, Seimyr L, Lundh V, Widström AM. Fussy child-difficult parenthood? Comparisons between families with a 'depressed' mother and non-depressed mother 2 months postpartum. *J Reprod Infant Psychol* 2000;18:225–238.
- Marteau T, Senior V. Illness representations after the human genome project: the perceived role of genes in causing illness. In: Petrie K, Weinman J (eds). *Perceptions of Illness and Treatment: Current Psychological Research and Implications*. Ilarwood Academic Press: Amsterdam, 1997:241–264.
- Kelley H. Causal schemata and the attribution process. In: Jones E, Kanouse D, Kelley H, Nisbett R, Valins S, Weiner B (eds). *Attribution: Perceiving the Causes of Behaviour*. General Learning Press: Morristown, NJ, 1971.
- Senior V, Marteau TM, Weinman J. Impact of genetic testing on causal models of heart disease and arthritis: an analogue study. *Psychol Health* 2000;14:1077–1088.