

Original Article

Adult CHD: the ongoing need for physician counselling about heredity and contraceptive options

Camila Londono-Obregon,^{1,2} Elizabeth Goldmuntz,^{1,2} Brooke T. Davey,^{1,2} Xuemei Zhang,³
Gail B. Slap,^{2,4} Yuli Y. Kim^{1,5,6}

¹*Division of Pediatric Cardiology, The Children's Hospital of Philadelphia;* ²*Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania;* ³*Biostatistics and Data Management Core, The Children's Hospital of Philadelphia Research Institute;* ⁴*Division of Adolescent Medicine, The Children's Hospital of Philadelphia;* ⁵*Division of Cardiovascular Medicine, Hospital of the University of Pennsylvania;* ⁶*Department of Medicine, Perelman School of Medicine, University of Pennsylvania, Philadelphia, Pennsylvania, United States of America*

Abstract *Purpose:* Current guidelines recommend that patients with CHD receive age-appropriate counselling on reproduction, pregnancy, and risk of heredity. Our aim was to examine patient knowledge of reproductive health and explore the association between patient knowledge of CHD transmission risk and earlier physician counselling in adults with CHD. *Methods:* We performed a cross-sectional survey of patients with CHD aged 18 years and older in a paediatric hospital. *Results:* Of the 100 patients who completed the questionnaire, most did not report counselling on heredity (66%) or contraception (71%). Of the 54 women, 25 (46%) identified their contraceptive options correctly; 42 (78%) women were classified as being at significantly increased risk for an adverse outcome during pregnancy, and of these 20 (48%) identified this risk correctly. Of all patients surveyed, 72% did not know that having CHD placed them at increased risk for having a child with CHD. On multivariate analysis, factors associated with correct knowledge about risk of recurrence were correct identification of CHD diagnosis ($p=0.04$) and patient-reported counselling ($p=0.001$). *Conclusions:* Knowledge about heredity, pregnancy risk, and contraceptive options is inadequate among adults with CHD followed-up in a paediatric subspecialty clinic. The majority of patients did not report a history of counselling about reproductive health. There is a strong correlation between history of counselling by the patient's cardiologist and correct knowledge about recurrence risk, suggesting that effective reproductive counselling can positively impact this knowledge gap.

Keywords: reproductive health; heredity; counselling

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CONGENITAL HEART DISEASE (CHD) IS THE MOST common major birth defect in humans, affecting four to eight per 1000 live births.¹ Over the past 50 years, advances in surgical repair and medical management have improved survival such that it is estimated that 90% of children with CHD

now reach reproductive maturity.² Current guidelines recommend that age-appropriate counselling on sexual and reproductive health, contraception, and pregnancy should begin in early adolescence and continue into adulthood.^{3,4} Female adolescents with CHD should be counselled about their disease-specific contraceptive options and pregnancy risks, and both male and female adolescents with CHD should be counselled about the likelihood of CHD in their offspring.^{5–9}

Surveys of sexually active adolescent and young adult women with CHD seen in outpatient clinics in

Correspondence to: Y. Y. Kim, MD, Assistant Professor, Department of Medicine, Philadelphia Adult Congenital Heart Center, Perelman Center for Advanced Medicine, 2nd floor East Pavilion, 3400 Civic Center Boulevard, Philadelphia, PA 19104, United States of America. Tel: +1 215 615 3388; Fax: +1 215 349 5927; E-mail: Yuli.Kim@uphs.upenn.edu

Canada, Australia, and Europe reveal that one-third to one-half of patients reported no previous physician counselling about their CHD-specific contraceptive options or medical risks during pregnancy.^{5,10,11} A survey of adults with CHD in the Netherlands revealed that 56% did not know whether their offspring were at increased risk of CHD and 67% did not recall having discussed inheritance with their cardiologists.¹²

We hypothesised that adolescents and young adults with CHD may not be receiving adequate or effective counselling about their reproductive and sexual health. Our objectives were to examine patient knowledge about these issues, understand how frequently patients recall being counselled, and explore the association between patient knowledge on heredity and previous physician counselling.

Materials and methods

This study was a cross-sectional survey of patients aged 18 years and older with CHD, recruited from an outpatient clinic located in a large, free-standing children's hospital between December, 2011 and March, 2012. The study protocol and survey instrument were reviewed and approved by the hospital's Institutional Review Board before subject recruitment and data collection. Patients unable to read English or complete the questionnaire independently were excluded from the study. A study investigator approached eligible patients in the clinic waiting area, obtained informed consent from those willing to participate, and directed patients to a private space for completion of the self-administered, gender-specific questionnaire before their physician visits.

The male and female versions of the study questionnaire examined patients' knowledge of their cardiac diagnoses and risk of transmission of CHD to their offspring, personal history of sexual activity, and contraceptive use, as well as previous reproductive counselling by their cardiologists. In our institution, there is no protocol for reproductive health counselling, and history of counselling was recorded from the questionnaire. The questionnaire for female patients included items related to their knowledge of contraceptive options and pregnancy risks associated with their specific cardiac defects and personal history of contraceptive use, pregnancy, and childbearing. The questionnaire for male patients included a question about paternity. Items pertaining to reproductive health knowledge, history, and behaviour were adapted from the national Youth Risk Behavior Survey administered annually by the United States Centers for Disease Control and Prevention.¹³ The electronic medical record of each study participant was reviewed for the following

information: demographics, presence or absence of health insurance, insurance type, and cardiac diagnosis.

The primary CHD diagnosis abstracted from the electronic medical record was categorised as simple, moderate, or severe according to the American College of Cardiology/American Heart Association Guidelines for the Management of Adults with Congenital Heart Disease.⁴ Diagnoses not included in the guidelines were categorised as follows: hypertrophic cardiomyopathy (moderate); Marfan syndrome without aortopathy (moderate); Marfan syndrome with aortopathy (severe); and pulmonary hypertension on medical therapy (severe). Aortopathy was defined as history of aortic root repair or aortic root diameter over 4 cm. Women were classified as being at significantly increased risk for maternal morbidity or mortality in pregnancy according to the modified World Health Organization classification – that is, World Health Organization class 3 or 4.⁷

Patient response to the question about diagnosis – *what is the name of your heart disease?* – was classified as correct if it agreed with the cardiac lesion of primary importance noted on the medical record. All other responses were classified as incorrect; three cardiologists reviewed all answers and reached consensus on the classification of each response. Patient response to the question about heredity – *what is the risk that a biologic child of yours might be born with heart disease?* – was classified as correct if it indicated that the risk is higher than that of the general population. Patient responses to the questions about the health risks of contraception – *does your heart disease limit your choices for birth control?* – and pregnancy – *is your health at risk if you get pregnant?* – were classified as correct or incorrect according to the American College of Cardiology/American Heart Association Guidelines for the Management of Adults with Congenital Heart Disease⁴ and the European Society of Cardiology Guidelines on the Management of Cardiovascular Diseases during Pregnancy.¹⁴

Data were stored in REDCap¹⁵ and exported to SPSS, version 19, for statistical analysis. For comparison of discrete variables between two groups, the χ^2 or Fisher's exact test was used. Statistical significance was set at $p < 0.05$. The full sample ($n = 100$) was used to explore the potential association of patient response to the question about heredity with gender, age, race, ethnicity, disease complexity, health insurance, college attendance, patient description of his/her heart defect, and patient report of previous counselling about heredity by his/her cardiologist. The study was powered at 0.80 with an α of 0.05 to detect a 0.30 difference in the rates of incorrect response to the question about heredity between patients who did and did not report previous

counselling by their physicians about the heredity of their cardiac defects. Logistic regression analysis was performed on the full sample to assess the adjusted contributions of those variables demonstrating associations with patient response to the question about heredity. The adjusted odds ratios (AOR) and 95% confidence intervals (95% CI) are reported for each variable comprising the final model.

Results

Of the 120 patients invited to participate in the study, 54 females and 46 males completed the study questionnaire and comprised the study sample (Table 1). Correct descriptions of their heart defects were provided by 50% of patients with simple CHD, 54% of patients with moderate CHD, and 63% of patients with severe CHD ($p = 0.578$).

In all, 78 (78%) patients reported a history of sexual intercourse with 70% of females and 86% of males having been sexually active at some point in time. Of these, first intercourse occurred at age 17 years or older in 50 (64%) patients, ages 15–16 years in 25 (32%) patients, and ages 12–14 years in three (4%) patients; 16 (29%) of the 54 female patients reported previous pregnancies, and eight (17%) of the 46 male patients reported that they had fathered children.

Patient knowledge of pregnancy risk and contraception

Of the 54 female patients, 42 (78%) women were classified as being at significantly increased risk for adverse outcome during pregnancy, and of these 20 (48%) identified this risk correctly. Regarding contraception, only 25 of all women (46%) were able to correctly answer whether their heart disease limits their contraceptive options. Moreover, 21 women (39%) had CHD diagnoses that limited their contraceptive options, and of these nine (43%) failed to identify this limitation.

Just over half of the female sample (54%) reported previous counselling on contraception by their cardiologists. Patients reported other sources of information on birth control, including primary-care doctors (26%), gynaecologist (32%), friends (25%), family members (46%), and teachers at school (30%).

We examined potential factors associated with correct knowledge of contraceptive options and found that over half of women with moderate (53%) and severe (56%) disease complexity were able to correctly answer whether their heart disease limits their contraceptive options, whereas 90% of women with mild disease did not ($p = 0.04$). Women with mild forms of CHD may incorrectly believe that their contraceptive options are limited when in fact they are not. Significantly more patients with moderate

Table 1. Patient characteristics and reproductive health history, knowledge, and counselling.

	Females (n = 54) (n (%))	Males (n = 46) (n (%))	All (n = 100) (n (%))
Age (years)			
18–26	36 (67)	25 (54)	61 (61)
27–45	16 (30)	19 (41)	35 (35)
>45	2 (4)	2 (4)	4 (4)
Race			
White	40 (74)	33 (72)	73 (73)
Black	13 (24)	7 (15)	20 (20)
Other	1 (2)	6 (13)	7 (7)
Ethnicity			
Hispanic	5 (9)	3 (7)	8 (8)
Non-Hispanic	49 (91)	43 (93)	92 (92)
Health insurance			
Private	38 (70)	39 (85)	77 (77)
Government-sponsored	16 (30)	6 (13)	22 (22)
None	0 (0)	1 (2)	1 (1)
Employment			
Employed	27 (50)	28 (61)	55 (55)
Unemployed	27 (50)	18 (39)	45 (45)
College attendance			
Yes	31 (57)	27 (59)	58 (58)
No	23 (43)	19 (41)	42 (42)
CHD complexity			
Simple	10 (19)	6 (13)	16 (16)
Moderate	19 (35)	22 (48)	41 (41)
Severe	25 (46)	18 (39)	43 (43)
What is the name of your heart disease?			
Correct answer	32 (59)	25 (54)	57 (57)
Incorrect answer	22 (41)	21 (46)	43 (43)
History of pregnancy or paternity			
Yes	16 (30)	8 (17)	24 (24)
No	38 (70)	38 (83)	76 (76)
Does your heart disease limit your choices for birth control?			
Correct answer	25 (46)	NA	
Incorrect answer	29 (54)	NA	
Is your health at risk if you get pregnant?			
Correct answer	26 (48)	NA	
Incorrect answer	28 (52)	NA	
What is the risk that a child of yours might be born with heart disease?			
Correct answer	18 (33)	10 (22)	28 (28)
Incorrect answer	36 (67)	36 (78)	72 (72)
Has your cardiologist ever talked to you about birth control?			
Yes	25 (46)	4 (9)	29 (29)
No or unsure	29 (54)	42 (91)	71 (71)
Has your cardiologist ever talked to you about the chance of having a child born with heart disease?			
Yes	26 (48)	8 (17)	34 (34)
No or unsure	28 (52)	38 (83)	66 (66)

and severe CHD reported being counselled by a cardiologist on birth control than those with simple CHD (33 versus 6%, $p = 0.034$), and correct identification of contraceptive options was associated with previous contraceptive counselling by a cardiologist ($p = 0.03$).

Patient knowledge of heredity

A minority of patients (28%) correctly reported that they had an increased risk of vertical transmission of CHD compared with the general population, and 34% of patients reported previous counselling about heredity from their cardiologist. Female patients were more likely than male patients to report that their cardiologists had counselled them about CHD-related heredity (48 versus 17%, $p = 0.001$). There was no significant difference in the proportions of female and male patients who answered correctly that their biological children were at increased risk of having CHD (33 versus 22%, $p = 0.265$). The variables that comprised the logistic regression model for correct patient response to the question about heredity are summarised in Table 2 and include patient report of previous heredity counselling by his/her cardiologist (AOR 5.1, 95% CI 1.9–14.0; $p = 0.001$) and correct patient description of his/her heart defect (AOR 3.2, 95% CI 1.1–9.6; $p = 0.04$).

Discussion

Less than half of all adult patients with CHD who are followed-up in a free-standing children's hospital recalled counselling by their cardiologists on birth control and CHD-related heredity, and knowledge about such issues is incomplete in this patient population. Our results are consistent with previous studies highlighting the knowledge gap in CHD patients regarding sexual health and reproduction and raise the question of why these patients lack this information. Specifically, patients may not be receiving the correct information they need, the counselling could be ineffective, or patients may not be retaining what they hear.^{16–18}

We show that there is an association between patient reproductive knowledge and patient-reported

history of previous counselling by the cardiologist. A higher proportion of women who correctly identified their contraceptive options reported previous counselling on birth control compared with those who did not correctly identify their contraceptive options. We found that patients with moderate or severe CHD were more likely to have been counselled by their cardiologists on contraception, which may explain why 90% of women with simple disease incorrectly identified their contraceptive options. Cardiologists may be biased towards counselling those with more complex disease, and therefore leaving those with milder forms of CHD with inadequate education.

We specifically wanted to explore the relationship between previous counselling and patient knowledge on heredity and found that previous counselling by the cardiologist about heredity was associated with a five-fold increased adjusted odds for correct knowledge of CHD transmission. A recent study by van Engelen et al¹² surveyed 332 adults with CHD and found that one-third received information about inheritance of CHD from their cardiologist and only 44% correctly estimated recurrence risk in their offspring. Contrary to our study, however, they did not find that self-report of having received information from a cardiologist was associated with higher knowledge on recurrence, and the authors surmise that patients do not receive or recall the provided information correctly; van Engelen et al did identify higher educational level as an independent predictor of correct knowledge on heredity. Our multivariate model showed a trend towards improved knowledge on heredity with higher education level but it did not reach statistical significance.

Of great concern is that only half of the patients surveyed knew the name of their CHD. Poor knowledge and understanding of their heart conditions have been identified in adolescents with CHD as well,¹⁹ highlighting the fact that self-knowledge and effective education is lacking across the age spectrum. We found that correct identification of the CHD lesion was an independent factor associated with good understanding of heredity. Lack of knowledge about their own disease may place patients at risk for poor understanding of transmission risk and underscores the importance of appropriate genetic counselling in order to make informed decisions regarding family planning.

Current guidelines recommend that patients with CHD receive age-appropriate counselling on reproduction, pregnancy, and risk of heredity and that this counselling should ideally begin in adolescence.³ Our study shows that over one-third of adult patients have their first sexual intercourse before the age of 17, supporting the need for transition clinics dedicated to

Table 2. Multivariate logistic regression analysis of variables associated with patient knowledge on risk of recurrence ($n = 100$).

	AOR	95% CI	p-value
Reported having received recurrence risk counselling	5.1	1.9–14.0	0.001
Knew CHD diagnosis	3.2	1.1–9.6	0.04
Reported college education	3.0	1.0–8.9	0.06

AOR = adjusted odds ratio; CI = confidence interval.

the education and counselling of reproductive issues. Protocols stipulating how, when, and who is to do this counselling are variable in the cardiac community.²⁰ In our institution, at present, there is no standardised care pathway for counselling or anticipatory guidance about these issues for CHD patients, and the onus is on the cardiovascular provider to independently meet this need. Cardiologists highly skilled in the management of CHD may face barriers of time, training, and comfort when caring for a patient in need of reproductive health counselling and care. Contraceptive and reproductive counselling for patients with CHD is further complicated by the variability across cardiac defects in heredity, oestrogen-associated risk, and pregnancy outcome and needs to be tailored to the individual patient. For these reasons, we are in the midst of planning and implementing a formal cardiac transition clinic targeting those aged 16 years and above. Collaboration with a contraception clinic or gynaecologic practitioner, adolescent medicine provider, as well as genetics specialists in a transition clinic can ensure the patient receives accurate, timely information.^{21,22} Although these care models may vary, our study highlights the important role the cardiovascular care provider plays in patient education on reproductive issues. Physician knowledge gaps and quality of physician counselling may be worthy of studying, and based on this reproductive health counselling protocols could be incorporated into this team approach.

Although there are guidelines on reproductive counselling for CHD patients, there are no universally accepted protocols on how to effectively deliver this information. Our study shows that patients obtain reproductive information from other sources outside of medical providers. We found that women and patients with complex disease were more likely to have received counselling, suggesting that cardiologists may be biased towards educating only certain subpopulations. Our study supports the development of standardised counselling protocols to ensure that all CHD patients receive balanced, consistent information.

A limitation of this study, aside from those associated with surveys, is recall bias. It is possible that a number of patients simply did not recall being counselled. We posit that recollection of counselling may serve as a proxy for the effectiveness of reproductive counselling on the part of the cardiologist. Finally, the study population was obtained from a cardiology clinic in a free-standing children's hospital, and our results may not be generalisable to the wider adult CHD population. That being said, the knowledge gap we demonstrate in our adult patients in a paediatric setting is similar to that of patients followed-up in an adult CHD clinic located in an adult hospital setting.¹⁸

In summary, clinician counselling and patient knowledge about CHD-related heredity, pregnancy

risks, and contraceptive options are inadequate among adults with CHD followed-up in a children's hospital CHD clinic; however, when recalled, clinician counselling was positively associated with correct reproductive knowledge on heredity. Our study emphasises the important role of the cardiovascular provider in the effective education of patients with CHD and highlights areas of improvement in the delivery of care for this complex growing population.

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Conflicts of Interest

None.

Ethical Standards

The authors assert that all procedures contributing to this study comply with the ethical standards of relevant national guidelines on human experimentation and with the Helsinki Declaration of 1975, as revised in 2008, and has been approved by the Institutional Review Board at the Children's Hospital of Philadelphia.

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