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Genetic counselling in parents of children with congenital heart disease significantly improves knowledge about causation and enhances psychosocial functioning

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ABSTRACT

Background: One of the key questions asked by parents of children with congenital heart disease (CHD) is 'why' and 'how did this happen?'. Receiving more information in response to these questions is therefore important to parents. This study sought to assess the efficacy of individualised genetic counselling sessions in improving knowledge of causation and psychosocial functioning in parents of children with CHD.
Methods: Parents of children undergoing surgery for CHD were offered individualised genetic counselling during their child's hospital admission. Assessments occurred at three time-points (immediately pre-, immediately post-, and two months post-session) using questionnaires comprising a purpose-designed knowledge measure, as well as validated psychological measures.
Results: Of the 94 participants approached, 57 attended the genetic counselling session (participation rate = 60.6%). Knowledge scores for the participants who completed all three questionnaires improved significantly from pre- ($\bar{x}$ = 7.38/16, SD = 3.53) to post-session ($\bar{x}$ = 13.33/16, SD = 2.82) ($p < 0.001$). Participants retained this knowledge over time, with no changes in scores at two-month follow-up ($p = 0.11$). Perceived personal control also increased post-session, while reported guilt, shame, depression, anxiety and stress decreased. Overall satisfaction was high, with 96.4% of participants indicating they would recommend this service to other parents of children with CHD.
Conclusion: Individualised genetic counselling sessions were highly beneficial to parents of children with CHD in regards to improving knowledge about the causes of CHD and enhancing psychosocial functioning, and should be considered as part of 'best care' practices.

1. Introduction

Congenital heart disease (CHD) affects approximately 6–8 per 1000 live born infants [1]. It places a significant burden on families with an affected child at the time of cardiac surgery and throughout the years of ongoing treatment. Very little clinically applicable information is known about the aetiology of CHD. Segregation of CHD within families has long suggested a heritable component and current understanding implicates both genetic and environmental contributions to disease development [2]. A number of genes have been associated with both syndromic and non-syndromic forms of CHD; however, the cause(s) of the majority of cases, which occur as sporadic events, remains unknown [3]. Advances in genetic technologies, such as whole exome sequencing, are beginning to unravel the complex genetic architecture of abnormal heart development [4]; however, this information can be difficult for families to access and understand.

In the past, the majority of cardiovascular genetic counselling has focused on arrhythmias and cardiomyopathies, with very little focus on structural CHD. This is primarily due to established causes and genetic testing options available for these groups of patients [5,6]. A key question asked by parents when their child is diagnosed with CHD is 'why' and 'how did this happen?'. Receiving more information on possible causes, proposed models of inheritance, and individualised recurrence risks is therefore important to families. A recent study conducted by our group found that 73% of parents were of the opinion that receiving information about the genetic causes of CHD was important; however,

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only 36% could recall ever having received this information [7]. Another study found that 41% of patients with CHD desired more information on the inheritance of their structural heart defect [8].

Clearly, there is a need for improved patient education on the genetic aspects of CHD and this is not only from the patient or parent's perspective. Receiving accurate information regarding inheritance and recurrence risk estimates also has implications for health services. Patients with CHD who reach reproductive age should be made aware of the increased recurrence risks for future family planning, even if this risk is still relatively low for sporadic forms of disease [9–11]. Numerous studies have found a substantial lack of knowledge about CHD inheritance and recurrence risks amongst patients and their families. A study conducted by van Engelen et al. found that 56% of adults with CHD had incorrect information on transmitting CHD to their future children [8]. Another study found that 37% of women with CHD did not think their future children would be at an increased risk of having CHD [12], and a more recent study involving adolescents with CHD reported that 80% of participants had significant gaps in knowledge pertaining to hereditary aspects of CHD [13].

Aside from information provision, genetic counselling also offers psychosocial support to families. It is well documented that parents of children with CHD suffer from increased levels of hopelessness and distress, including depression, anxiety and somatisation [14–16]. Lawoko and Soares suggest that increased levels of parental distress and hopelessness may be due to prolonged feelings of guilt in regards to their child's heart condition [15]. Parental guilt can be associated with reduced self-esteem, self-blame and feelings of worthlessness which in turn can manifest as symptoms of depression or anxiety [17]. The supportive aspects of genetic counselling may assist in reducing levels of anxiety and guilt, particularly those associated with, or related to, the inheritance of CHD.

In this study we assessed the efficacy of an individualised genetic counselling session developed specifically for parents of a child with CHD. We provided information about the causes and inheritance of CHD and supported parents in understanding and adjusting to this information, according to their personal circumstances. The specific aims of this study were therefore to: (1) assess parental knowledge of the hereditary aspects of CHD pre- and post-session; (2) assess psychosocial functioning, hypothesizing that the individualised genetic counselling sessions, would assist in alleviating anxiety and guilt, particularly in relation to inheritance; and (3) obtain feedback in regards to the format and content of the genetic counselling session for the potential future implementation of the intervention.

2. Materials and methods

2.1. Participants

Participants included parents attending preadmission clinic prior to their child's elective cardiac surgery. Patients undergoing complex neonatal or emergency surgeries do not attend preadmission clinic and were therefore excluded from this study. Parents of children with a diagnosed genetic syndrome, or those who had previously been seen by a genetics professional were excluded as familiarity with genetic information could affect the analyses. Parents who did not have sufficient English language skills to take part without the aid of an interpreter were also ineligible.

2.2. Procedure

Ethics approval (LNR/12/SCHN/191) and site specific approval (LNR/12/SCHN/365) was granted by the Sydney Children's Hospital Network Human Research Ethics Committee. For participation ease, and to ensure maximum recruitment potential, the study was aligned to the routine surgical process for patients undergoing elective cardiac surgery at The Children's Hospital at Westmead. Participants were approached about the study during preadmission clinic prior to their child's cardiothoracic surgery, at which time consent was obtained. Following surgery completion and discharge from the intensive care unit, a suitable time for the genetic counselling session was arranged a day or two prior to discharge from the hospital. Participants were asked to complete three questionnaires; one immediately prior to the session, immediately after the session, and approximately two months post-session. The timing of the follow-up questionnaire coincided with the patients' clinical or surgical follow-up appointments, at which point the participants were approached and handed the final questionnaire.

2.3. Genetic counselling session design

The genetic counselling session was delivered by the genetic counsellor on an individual family basis in which either one or both parents attended. One hour was allocated per session. The content of the sessions was developed using current evidence from the literature in conjunction with expert consultation. The format was semi-structured, covering a set of key issues and tailoring information such as recurrence risk estimates, as well as the emotional support, to individual participants' needs. Key areas covered in the session included:

- An overview of CHD incidence
- A detailed family history
- Information on chromosome, genes, DNA and mutations
- Possible genetic causes of CHD (including syndromic, multifactorial, familial)
- Environmental and teratogenic contributions
- Heterogeneity, variable expression, reduced penetrance
- Individualised recurrence risk estimates
- Genetic testing and research options.

A detailed outline of the genetic counselling session, including the resources used, is presented in 'Supplementary Materials and Methods' (Table S1).

2.4. Measures

The survey was designed specifically to evaluate the effect of, and parents' response to, the genetic counselling session and was comprised of both purposely-designed and validated measures, as described below.

2.4.1. Demographic characteristics (11 items)

Included items are about the participants as well as their families. Items also included clinical information about the child with CHD such as the STS-EACTS mortality score (which ranges from 0.1–5) associated with their child's surgical procedure.

2.4.2. Sources of genetics information (8 items)

Participants were asked about the sources of information relating to the causes and inheritance aspects of CHD by rating statements as 'Yes', 'No' or 'Unsure'.

2.4.3. Emotional aspects of having a child with CHD (7 items)

To explore parents' feelings associated with having a child with CHD, participants were asked whether or not they agreed with a series of statements, rating 'Yes', 'No' or 'Unsure' on a range of issues.

2.4.4. Knowledge (16 items)

To date there is no validated measure to assess parental knowledge on genetic factors and inheritance of CHD. For this reason a purposely-designed knowledge measure was created for this study by reviewing questionnaires created for similar studies [8,18]. It also enabled direct assessment of knowledge in regards to the information presented in the genetic counselling session. The final version of this measure consisted of 14 statements on genetic aspects of CHD which participants rated as either 'True', 'False' or 'Unsure', as well as two multiple choice items asking participants to indicate perceived recurrence risks. Higher scores were indicative of greater knowledge. Statements included 'Most cases of CHD occur without a family history' and 'After having a child with CHD, the chance of having another child with CHD is higher'. Internal consistency of the individual items was high (Cronbach's $\alpha = 0.78$). The knowledge measure was included in all three surveys.

2.4.5. Perceived Personal Control — PPC (9 items)

The PPC measure has been used to evaluate the benefit of genetic counselling in a number of published studies [19,20]. The PPC measure is designed for families with genetic conditions with a known cause and testing options; however, in the majority of CHD the cause is unknown and there are limited genetic testing options. For this reason, the PPC measure used in this study was modified slightly, but care was taken to ensure the cognitive, behavioural and decisional constructs were maintained. The PPC measure was included in all three surveys.

2.4.6. Personal Feelings Questionnaire — PFQ-2 (22 items)

The PFQ-2 measures the degree of chronic guilt and shame experienced by an individual with six items relating to 'guilt', 10 to 'shame' and six additional 'filler' items. The PFQ-2 was included in the pre-session and follow-up surveys.

2.4.7. Depression, Anxiety and Stress Scale — DASS-21 (21 items)

The DASS-21 is a measure of distress along three symptom clusters; depression, anxiety, and stress. Normative data, as well as clinical cut-offs, are available for this measure, increasing the value of this tool. The DASS-21 was included in the pre-session and follow-up surveys. Cronbach's α for baseline DASS-21, PFQ-2 as well as PPC scores in this sample were 0.87, 0.87 and 0.85 respectively.

2.4.8. Genetic Counselling Satisfaction Scale — GCSS (6 items)

The GCSS is used to measure client satisfaction with the genetic counselling process. It is broadly applicable and has been used to assess satisfaction with genetic counselling in