

Parental preferences for *CDKN2A/p16* testing of minors

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Purpose: Genetic testing of minors is controversial, as ethical considerations depend on multiple aspects of the particular disease and familial context. For melanoma, there is a well-established and avoidable environmental influence and a documented benefit of early detection. **Methods:** We surveyed 61 *CDKN2A/p16* mutation-tested adults from two kindreds about their attitudes toward genetic testing of minors immediately posttesting and 2 years later. **Results:** Overall, 86.9% expressed support of melanoma genetic testing of minors, with the importance of risk awareness (77.4%) and the likelihood of improved prevention and screening behaviors (69.8%) as the most frequently cited potential benefits. Among mutation carriers, 82.6% wanted genetic testing for their own children. These preferences remained stable over a 2-year period. Most respondents (62.3%) favored complete involvement of their children in genetic counseling and test reporting; 19.7% suggested that children be tested but not informed of the results. Concerns about inducing psychological distress or compromising children's decision autonomy were infrequently cited. Testing preferences did not vary by respondent age, gender, or melanoma history. **Conclusion:** Respondents strongly supported melanoma genetic testing of minors, with most citing improved health behavior as a likely outcome. We discuss options for melanoma genetic counseling and testing of minors. *Genet Med* 2010;12(12):823–838.

Key Words: *CDKN2A*, familial melanoma, genetic counseling, children, prevention

Researchers and medical professionals have debated the merits and harms of genetic test reporting for minors at length,^{1,2} but these issues have yet to be applied to melanoma genetic testing. The purpose of this article is to review ethical aspects of melanoma genetic testing of minors and to provide detailed quantitative and qualitative survey data regarding the preferences of *CDKN2A/p16* (*p16*) mutation-tested adults for and against such testing of minors. We then discuss the implication of these survey results for the development of guidelines for melanoma genetic testing of minors.

Melanoma penetrance estimates for *CDKN2A/p16* mutation carriers vary significantly by country and participant selection criteria. Among US residents in high-risk pedigrees, a pathogenic *CDKN2A/p16* mutation confers a 76% lifetime risk for melanoma, the most aggressive and deadly form of skin cancer.³ Across all high-risk populations, the penetrance of the *CDKN2A/p16* mutation is lower—67% by age 80 years (95%

confidence interval [CI] = 0.31–0.96), which is due in part to geographic and ethnic variance. Additionally, population-based studies have indicated a lower, but substantial, risk of 28%.⁴ *CDKN2A/p16* genetic counseling and test reporting have recently entered clinical use for adults in high-risk melanoma families. International consensus guidelines governing both counseling and test reporting of adults have been recently disseminated^{5,6}; however, no one has established formal, data-based guidelines for testing individuals younger than 18 years.

Predictive genetic testing of minors is advocated only when there is a clear medical benefit.⁷ Genetic testing of children with cancer predisposition syndromes such as familial adenomatous polyposis (FAP) and multiple endocrine neoplasia type 2 is generally supported because children who are found to have a mutation are recommended to have increased screening and often prophylactic removal of the at-risk organ to avoid cancer development.^{8,9} On the other hand, medical professionals and genetic counselors do not recommend that minors undergo *BRCA1/2* predictive genetic testing for breast cancer because only individuals aged 25 years or older are advised to engage in clinical surveillance behaviors, and prophylactic surgery and chemoprevention are the only possible prevention behaviors.¹⁰

We believe that convincing arguments can be made both in favor of and against melanoma genetic testing of minors. We will present a summary of these arguments, thus providing context for the data on parental beliefs and preferences that we will report. Although there are presently no data regarding minors' behavioral or psychological outcomes after receipt of *CDKN2A/p16* genetic testing, we will review relevant behavioral and psychological outcomes that may result from genetic testing of minors.

Childhood sun exposure is a significant risk factor for melanoma,¹¹ and minors are apt to receive sunburns, use tanning beds, and be unlikely to protect their skin consistently.^{12–15} Importantly, the relationships between sun exposure, geography, ethnicity, and familial melanoma incidence and survival are complex, with conflicting research findings, and in need of further investigation.^{3,16–18} However, if *CDKN2A/p16* testing motivated minors to engage in primary prevention behaviors through avoidance of ultraviolet radiation exposure, such improvements in photoprotection might reduce minors' melanoma risk.¹⁹ It is possible that adolescents may be more motivated to engage in prevention and screening behaviors if they learn that they are at increased genetic risk for melanoma. Further, parents may be able to structure a child's recreational activities to reduce ultraviolet exposure. Thus, in the particular case of melanoma, children and adolescents have many opportunities to make lifestyle changes and choices to potentially reduce melanoma risk. The medical benefits accrued through melanoma-related preventive behavior can be contrasted to other diseases for which genetic testing of minors is available but not recommended, such as testing for Huntington disease or *BRCA1/2* mutations, because there are either no known behavioral or prevention strategies or none that would confer benefit if implemented in childhood.

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In terms of secondary prevention behaviors, adolescent screening behavior in high-risk families may be particularly important. The mean age of melanoma onset is earlier in *CDKN2A/p16*-mutation-carrying families than in the general population (mean of 35 years compared with median of 59 years).^{3,20} Although melanoma occurs infrequently in children, children as young as 12 years in these high-risk families have been diagnosed.³ Accordingly, children in high-risk families are advised to start regular screening at ages 10–12 years. Early detection is a well-established factor for increasing melanoma survival. Five-year survival rates are 98.1% for localized cancer but are only 15.3% for distant metastatic disease.²⁰ These data suggest that minors who undergo close dermatological screening and perform monthly self-skin examinations could maximize the likelihood of early detection and subsequent survival. These potential medical benefits may make *CDKN2A/p16* testing appropriate for minors.

Some researchers and practitioners have argued that *CDKN2A/p16* testing is unnecessary because individuals who test negative for a familial mutation may still be at elevated risk for melanoma due to other predisposing phenotypic or environmental factors²¹ and would likely benefit from the same risk-reducing strategies as are recommended for carriers. This argument—that members of high-risk families do not need genetic testing to implement consistent photoprotection and screening behaviors^{22,23}—would apply to minors as well. It is unknown whether testing would motivate minors to engage in prevention and screening behaviors, as there are no data regarding the behavioral outcomes of melanoma genetic test reporting of minors. However, emerging evidence for the impact of such testing on high-risk adults suggests potential benefits in terms of improved screening and photoprotection.^{24,25} Specifically, unaffected carriers who underwent *CDKN2A/p16* genetic testing reported prospective improvements in both the frequency and thoroughness of skin self-examinations at 1 month²⁶ and 2 years after test reporting.²⁵ Further, unaffected carriers complied more to photoprotection recommendations after genetic test reporting.^{25,27} Although these findings have not yet been evaluated in comparison with a counseling-only control group, they do suggest important benefits of genetic test reporting and counseling in a population that had received extensive prior counseling based on family history alone. It is possible that minors who receive genetic counseling and test reporting may accrue similar benefits.

In addition to questions of medical benefit, arguments related to a child's autonomy and the potential psychological benefits and harms to children and their families have been raised with respect to the impact of genetic testing of minors.^{2,28,29} First, a child who undergoes genetic testing is deprived of the right to decide as an adult whether this is information he would like to know. However, when minors are prohibited from undergoing genetic testing, a different decision has been made for the minor.³⁰ Without such testing, the minor may be deprived of potentially important health information that could be used to make decisions about adolescent recreational and occupational activities or to make fully informed decisions about other risk behaviors, such as sunbathing or the use of tanning booths. There is reason to believe that individuals who know their familial risk but do not know their own genetic status may be insufficiently motivated to change their behavior.²⁷ For example, children aged 9–14 years in at-risk families in which a parent has received a diagnosis of melanoma do not report greater behavioral adherence than children of the same ages in families at population risk.³¹

Second, there are concerns that a positive genetic test result will induce psychological distress and/or a sense of fatalism about disease prevention and treatment.^{32–35} Research findings to date from two different research groups suggest, however, that test reporting does not create either psychological distress or cancer fatalism, at least for adults. In our research with *CDKN2A/p16* mutation-tested adults, we found psychological benefits of test reporting, including prospective increases in perceived control over the development of a new melanoma and decreased beliefs that disease is inevitable given a positive test result.²⁵ Further, a sample of Australian *CDKN2A/p16* mutation carriers reported decreases in anxiety and depressive symptoms 2 weeks and 1 year after testing.²⁴ However, it is important to note that this study also showed that members of high-risk families who declined testing had more fatalistic beliefs about melanoma than those who underwent testing.²⁴ With respect to psychological outcomes among minors, most research has examined psychological outcomes among minors receiving genetic test results for FAP. This research suggests that minors aged 5–17 years who have received genetic test results for FAP have not reported significant changes in psychological distress,³⁶ although another study found that children aged 10–16 years who received positive FAP test results reported levels of anxiety and depression that were slightly higher than those of children who received negative test results but not higher than adults who received positive test results.³⁷

A minor's emotional and behavioral responses to a positive test result may depend on his emotional or cognitive maturity.^{32,38} Several researchers have addressed the idea that a minor's maturity, or cognitive ability to understand the long-term implications of a test result (independent of age), should be considered when deciding whether minors should undergo genetic testing³⁸ and have accordingly suggested that testing should only be offered to mature minors. Individuals younger than 18 years vary in their ability to comprehend medical information and in their ability to make rational decisions (such as whether to undergo testing or how to manage their risk), and this individual variability should be taken into account both in terms of determining whether a minor should be allowed to decide on his own about testing and how to present medical information to minors.³⁹ Accordingly, it is important to understand whether members of high-risk melanoma-prone families would consider a minor's maturity or other developmental factors when deciding whether melanoma genetic testing is appropriate for minor children and grandchildren.

Finally, concerns about psychological distress and fatalism may be balanced against potential psychological benefits that may accrue to members of high-risk families, such as having greater certainty about their health and the likely future health of their children and grandchildren. One of the major benefits of undergoing genetic testing is its potential to reduce such uncertainty,²⁸ and adults who choose to undergo testing may be motivated by negative attitudes toward uncertainty or a desire to have certainty about their risk.^{40,41} Individuals may experience anxiety or worry before genetic testing because they know that they could be at risk but do not know for sure. Qualitative interview studies suggest that members of high-risk melanoma families believe that learning their mutation status would decrease psychological distress among both carriers and noncarriers.⁴² For minors already aware of the presence of the mutation in their family, a definitive test result could be psychologically beneficial not only for the child but also for the family unit as a whole.

This brief review suggests that multiple aspects of the anticipated psychological, behavioral, and ultimate health impact of

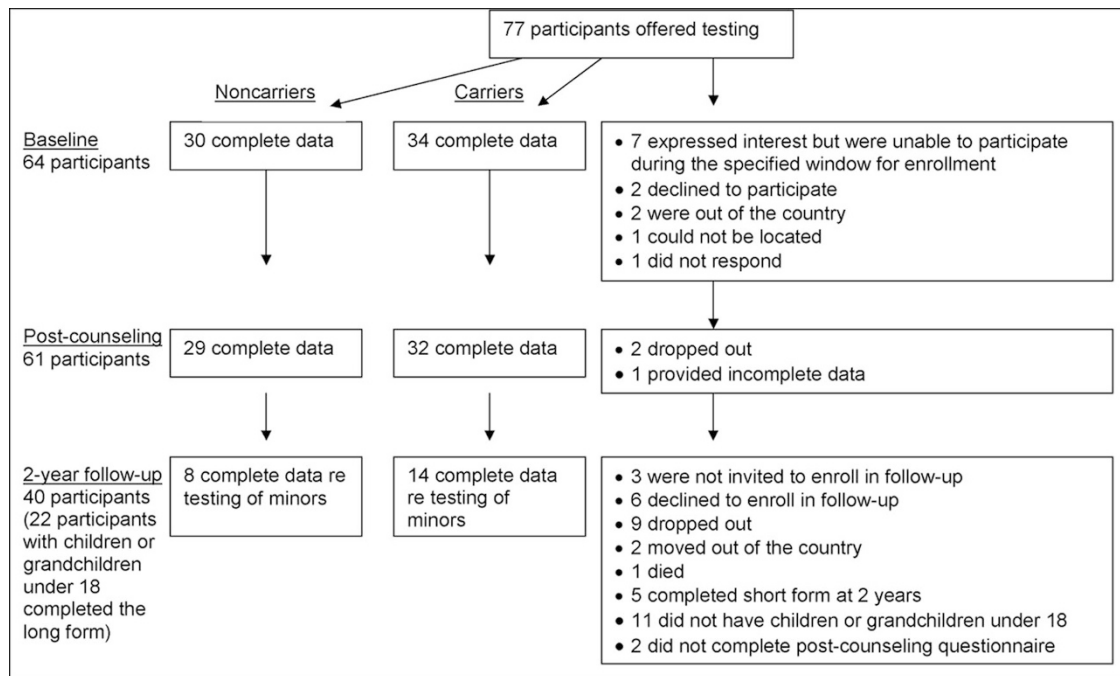


Fig. 1. Flowchart of respondent enrollment and attrition in the parent study of melanoma genetic test reporting and the follow-up study of long-term responses to genetic test reporting.

melanoma genetic testing of minors are important considerations in developing guidelines for genetic counseling and test reporting. Missing from this discussion to date are detailed analyses of the beliefs and preferences of adult members of high-risk melanoma-prone families. Such beliefs are particularly important because parents may be better able to assess their child’s ability to cope with a positive genetic test result than a genetic counselor who may be less familiar with the familial context or a child’s individual characteristics.^{7,43} Parents may also be able to assess whether their child would be more or less adherent to behavioral recommendations after a test result. These parental assessments could then determine the information their children receive, as genetic counselors could incorporate these preferences into individualized counseling sessions.

To address this gap in the ongoing discussion of melanoma genetic testing of minors, we asked members of two high-risk melanoma-prone kindreds who had recently undergone melanoma genetic test reporting and counseling to indicate their opinions and preferences regarding such testing for their minor children and grandchildren and to explain the reasons for their preferences. We assessed testing preferences again 2 years later.

MATERIALS AND METHODS

In companion test reporting and follow-up studies (Institutional Review Board no. 7916 and no. 13816), melanoma genetic counseling and test reporting were offered to a total of 77 adult research participants from two large melanoma pedigrees enrolled in previous *CDKN2A/p16* identification studies^{44,45} who had contributed DNA samples for research genetic testing. Each of these samples was subjected to genetic testing through a CLIA-certified laboratory (Myriad Genetic Laboratories or Yale University School of Medicine DNA Diagnostic Laboratories), and participants were offered the opportunity to receive

these results. None of the participants was aware either of his or her genetic status or of the presence of the *CDKN2A/p16* mutation in his or her family before participation in this study.

From May to November 2005, 64 (83.1%) of these individuals completed a baseline questionnaire and a genetic counseling and test reporting session, as shown in Figure 1. During individual predis closure genetic counseling sessions, participants received melanoma genetics education and, after informed consent, were offered the opportunity to receive their genetic test result. All 64 participants elected to receive their result. After result disclosure, the meaning of the result was reviewed, and tailored screening and management recommendations were provided. Participants were informed of their children’s risk for having inherited the mutation if they were positive, but they were not specifically counseled in detail about the pros and cons of potentially having their minor children tested nor was testing offered to minor children through this study. A complete description of the genetic testing protocol can be found in previously published supplementary materials.²⁶

Sixty-one participants provided complete questionnaire data at a postcounseling assessment regarding their attitudes and preferences with respect to genetic testing of minor children. The majority of the data presented in this article were obtained from this postcounseling assessment. We also present data from a follow-up assessment conducted 2 years later. The 2-year follow-up survey was completed by 40 total participants (62.5% of those completing the baseline). As shown in Figure 1, complete data regarding genetic testing of minors were obtained from 22 participants with either minor children or grandchildren. Of these, 14 were carriers, and eight were noncarriers, representing 66.7% and 40.0% of the originally enrolled carriers and noncarriers with children or grandchildren younger than 18 years, respectively. When we compared the postcounseling survey responses of participants who did or did not complete the 2-year follow-up assessment, there were no significant differ-

ences in beliefs about testing for children in general, one’s own children, children of carrier relatives, whether all children should be treated the same, or age at which children should be tested.

Measures

Demographic and medical history

Participants completed standard demographic questions, and the melanoma history of each participant was confirmed through the Utah Cancer Registry (a Surveillance Epidemiology and End Results Registry) and the Utah Population Database.

Preferences regarding genetic testing of minors

In a structured questionnaire administered after participants’ own genetic counseling and test reporting session, participants answered questions concerning specific attitudes and preferences related to melanoma genetic testing of minors. The survey items were jointly developed by two licensed genetic counselors with extensive experience in counseling members of high-risk cancer-prone families, a physician expert in cutaneous oncology who served as director of the clinic and a social psychologist with expertise in health cognitions. The items were developed to represent key aspects of the uptake of melanoma genetic testing for minors, as they focused on preferences for testing in general versus for one’s own children and carrier relatives’ children, the age at which participants would want their children to be tested, and reasons why participants would or would not want their children to be tested as minors. The protocol was also designed to elucidate whether participants’ views about testing would be the same for all of their children and whether particular factors, such as emotional and cognitive maturity, would be listed as important in making such decisions. The items were tested with the first few participants to receive *CDKN2A/p16* test reporting and evaluated for clarity and ease of completion. As no difficulties were reported, the items were retained for use in the study.

In a series of six questions, participants were asked to indicate “your personal opinion about testing children younger than 18 for *p16* gene changes.” (We used the term “gene change,” rather than “genetic mutation” throughout the questionnaire to reduce stigmatization and improve understanding of the ques-

tion.) Participants were not asked to differentiate responses based on whether they had minor children or grandchildren.

Participants were asked the following questions, each with the response options of “yes,” “no,” or “I don’t know”:

1. “In general, do you think children should be tested for the *p16* gene change?”
2. “Would you want your own children to be tested for the *p16* gene change?”
3. “Would your opinions and beliefs be the same for all of your children?”
4. “If there are some members of your family who have tested positive for the *p16* gene change, do you believe that their minor children (those under 18) should be tested?”

After each of these questions, participants were asked to provide additional information about their choices in an open-ended response format.

Next, participants completed a multiple-choice question asking them to indicate which aspects of the genetic testing process they would like their children to be involved in, if any, ranging from participation in a general melanoma genetics education session to undergoing genetic testing but not learning the results to having the test and being informed of the results (see Fig. 2 for complete set of response options). Participants were able to select as many choices as they wished.

Finally, participants were asked the following open-ended questions:

5. “At what age do you believe children should become involved in any aspect of the genetic testing process?”
6. “What other factors, if any, about the child would you take into account when making this decision?”

Two-year follow-up questionnaire

At the 2-year follow-up, participants with minor children were asked, “How interested are you in having your children 18 years of age or younger tested?” (1 = “not at all” and 5 = “very much”).

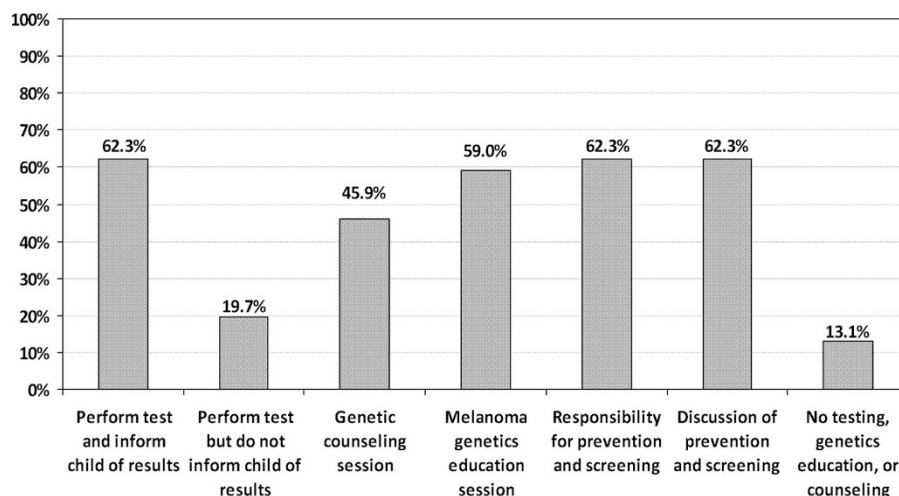


Fig. 2. Percentage of respondents who indicated they would want their children to be involved in different aspects of the genetic counseling and test reporting process.

Table 1 Demographics, medical history, and parental status of *CDKN2A/p16* carriers and noncarriers at baseline

Variables	Noncarriers (n = 29)	Carriers (n = 32)	Total (N = 61)
Age, mean (SD), yr	46.34 (15.55)	44.50 (15.22)	45.38 (15.28)
Male (%)	41.4	59.4	50.8
Married (%)	79.3	81.3	80.3
Have children and/or grandchildren <18 yr (%)	82.8	71.9	75.4
Education > high school (%)	89.7	87.5	88.5
Church of Jesus Christ of Latter-day Saints (LDS; %)	93.1	96.9	96.7
Income (median)	\$50–\$59K	\$60–\$69K	\$50–\$59K
Melanoma history (no.)	2	18	20
Confirmed melanomas, mean (SD)	2.5 (0.71)	1.25 (1.98)	2.25 (2.10)
Years since most recent melanoma diagnosis, mean (SD)	20.0 (18.39)	11.06 (11.52)	11.95 (12.01)

Coding system for qualitative responses to genetic testing-preference questions

As noted earlier, participants were asked to explain their answers to the three yes/no questions about their preferences (testing for minors in general, testing for their own children, and testing for children of carrier relatives), to explain why their beliefs would or would not be the same for all of their children, and to describe any other factors they would take into account in making a decision about genetic testing for a particular child. Our first step in developing a coding scheme for these responses was to conduct a literature review to identify common reasons cited for and against genetic testing of minors and to identify factors that may influence whether an individual child is ready for genetic testing, such as maturity, age, or personality type. These reasons and factors gleaned from the literature were used to identify themes in participant responses, which were then organized into five categories: (1) reasons for testing children, (2) reasons for not testing children, (3) reasons for treating all children the same, (4) reasons for not treating all children the same, and (5) other factors to consider when making a decision to test children. A certified genetic counselor with more than 10 years of experience in pediatric oncology genetics reviewed and helped to develop the coding scheme, which was used by two independent raters to code participant responses. The raters reached 91% agreement and resolved all disagreements in conference.

Overview of analyses

We first present standard demographic and medical history data for the sample, including a detailed comparison of these factors in the two large families (Kindreds A and B) from which all but two participants were recruited. We next present testing-preference data obtained from the postcounseling questionnaire and 2-year follow-up data concerning the stability of such preferences. We used logistic regression analyses to examine predictors of the support for genetic testing of minors in general, of one's own minor children, and of carrier relatives' minor children. Participants who indicated they were "unsure" about any question were coded as "no" to create dichotomous no = 0/yes = 1 outcomes. Each regression model included the following predictors: kindred ($A = 0/B = 1$), respondents' melanoma history (no = 0/yes = 1), *CDKN2A/p16* mutation status (negative = 0/positive = 1), respondents' age (continuous), sex

(female = 0/male = 1), and parental/grandparental status (absence of children or grandchildren younger than 18 years at baseline = 0/presence = 1). We conducted a linear regression analysis using the same model to examine predictors of the age at which individuals believed children should become involved in the genetic testing process. We also present descriptive data concerning the particular aspects of the genetic counseling process in which participants wanted their children to be involved. Finally, we present a detailed analysis of the reasons respondents provided for and against melanoma genetic testing of minors and the factors they said they would take into account in making such a decision for a particular child.

RESULTS

Participant characteristics and demographics

Table 1 presents demographic, medical history, parental status, and *CDKN2A/p16* mutation status data for our sample. Thirty women (49.2%) and 31 men (50.8%) were enrolled, with an average age of 45 years (SD = 15.28, range = 21–80). All participants were white. All participants were high school graduates, and more than half (55.8%) had completed a bachelor's degree or higher. Median annual income was \$50–\$59,999. The majority were married (80%) and reported having children or grandchildren younger than 18 years (75.4%). Thirty-two (52.5%) received positive test results, and 20 (32.8%) had a confirmed personal history of one or more melanomas. There were no statistically significant differences in demographic characteristics or parental status between carriers and noncarriers.

Table 2 presents a detailed comparison of the participants from the two large kindreds from which our participants were originally recruited (hereafter referred to as Kindreds A and B). All but two participants came from these two kindreds (Kindred A, 44.3%; Kindred B, 52.5%; and other kindreds, 3.3%). We first determined whether the kindreds were comparable with respect to demographic and medical history variables that might influence preferences for genetic testing of minors. Respondents from each kindred did not differ in age, gender composition, marital status, education, or income, although members of Kindred A were somewhat more likely to report having children or grandchildren younger than 18 years ($t(55) = 1.94, P < 0.06$). Further, as presented in Table 2, Kindreds A and B did not differ in the proportion of respondents with a melanoma history or who

Table 2 Age of earliest onset and other melanoma history characteristics associated with kindred membership

Variables	Kindred A (n = 27)	Kindred B (n = 32)	Test statistic	P
Have a personal melanoma history (%)	25.9	34.4	$\chi^2(1) = 0.49$	0.48
<i>CDKN2A/p16</i> mutation carriers (%)	48.1	53.1	$\chi^2(1) = 0.15$	0.70
Age of affected respondents' earliest melanoma, mean (SD), range (yr)	34.1 (9.3), 25–53	29.8 (9.0), 18–47	$t(16) = 0.98$	0.34
Years since affected respondents' earliest melanoma, mean (SD), range	18.7 (15.4), 4–41	22.0 (11.1), 5–39	$t(16) = 0.53$	0.61
Years since affected respondents' most recent melanoma, mean (SD), range	5.9 (4.1), 2–14	17.0 (14.2), 1–39	$t(16) = 2.45$	0.03
Age of earliest melanoma in first-degree relatives (yr)	20	16		

tested positive for the *CDKN2A/p16* mutation. We next examined age of melanoma onset. As presented in Table 2, the age at which participants from Kindred A had their earliest melanoma diagnosis ranged from 25 to 53 years ($M = 34.1$), whereas for Kindred B, age of onset ranged from 18 to 47 years ($M = 29.8$). Kindreds A and B did not differ in the average age of onset or in the number of years from earliest onset to the time of their genetic counseling session. Next, we compared the recency of respondents' melanoma diagnosis in the two kindreds. As presented in Table 2, both kindreds had experienced a melanoma diagnosis in the past 2 years. However, the average number of years between participants' most recent melanoma and the time of the genetic counseling session was significantly shorter for members of Kindred A than it was for members of Kindred B ($M_A = 5.86$, $M_B = 17.0$; $t(16) = 2.45$, $P < 0.030$). Finally, we reviewed the pedigrees to determine the age of onset in first-degree relatives of our respondents, regardless of their participation in the test-reporting study. The youngest age of first melanoma diagnosis was 20 years in Kindred A and 16 years in Kindred B. Thus, members of both kindreds had experienced a diagnosis of melanoma among either teenaged or young adult family members.

Preliminary analyses involving kindred as a predictor of preferences for genetic testing

We first examined whether there were any kindred effects in responses to our primary testing-preference outcomes. These analyses yielded only a single marginally significant difference between the kindreds, such that members of Kindred B were somewhat more in favor of genetic testing for their own children than were members of Kindred A (78.1% vs. 56.0%, respectively; $\beta = 1.03$, standard error [SE] = 0.59, odds ratio [OR] = 2.81, 95% CI = 0.89–8.88, $P < 0.08$). The kindreds did not differ in responses to our other major outcomes, including preferences for genetic testing of minors in general ($\beta = 0.05$, SE = 0.60, OR = 1.05, CI = 0.32–3.40, $P < 0.94$) and beliefs about the age at which children should be tested ($\beta = 0.01$, SE = 0.61, OR = 1.01, CI = 0.31–3.32, $P < 0.98$).

Next, we examined whether kindred interacted with any of the other demographic and medical history variables in the model in predicting responses to any of the three quantitative testing-preference questions. As no such interactions were obtained, none were included in the final model. To account for the marginally significant difference in preferences for testing one's own children based on kindred, we retained the main effect of kindred in the final regression model as a predictor of all genetic testing-preference outcomes.

Primary analyses of *CDKN2A/p16* testing preferences

Preferences for testing in general

Table 3 presents the proportion of respondents supporting melanoma genetic testing in response to the three major testing-preference questions. Overall, the majority of participants (73.8%) supported genetic testing for children in general, whereas approximately one in five participants (19.7%) believed children should not undergo genetic testing. The remainder (6.6%) reported being unsure. When we conducted a logistic regression with the multivariate model presented in Table 4, this analysis yielded no significant demographic, medical history, or parental status predictors of support for genetic testing of minors in general (all P values > 0.10).

Preferences for testing own children and/or grandchildren

The majority of participants (69.0%) wanted their own minor children or grandchildren to undergo genetic testing, whereas 29.3% did not, and 1.7% were unsure (Table 3). As expected, participants who tested positive for the *CDKN2A/p16* mutation were significantly more likely to express support for testing their minor children ($\beta = 2.63$, SE = 1.03, OR = 13.82, CI = 1.82–104.96, $P < 0.02$; Table 4). As presented in Table 3, *CDKN2A/p16* mutation carriers overwhelmingly wanted their children to be tested (86.7%), whereas only four carriers (13.3%) did not want their children to undergo genetic testing. Among noncarriers, 50% indicated that their own children should undergo melanoma genetic testing (see subsequent analyses of parents' reasons for such preferences). When we further stratified these testing preferences by parental status, 82.6% of *CDKN2A/p16* mutation carriers with minor children or grandchildren wanted them to be tested. As presented in Table 4, the main effect of kindred, which was marginally significant in the preliminary analyses, was not significant in the multivariate model, and no variables other than mutation status significantly predicted support for genetic testing of one's own minor children (all P values > 0.10).

Age of melanoma onset as a predictor of testing preferences

We conducted supplementary analyses to examine whether responses to our primary testing-preference outcomes were associated with the age at which participants were first diagnosed with a melanoma. This variable could not be tested in the full multivariate model, as only participants with a melanoma his-

Table 3 Preferences for *CDKN2A/p16* genetic testing for minors for children in general, for their own children or grandchildren, and for the children of mutation-positive family members, as a function of respondents' mutation status and in the total sample

Testing preference question	Noncarriers % (n = 29)	Carriers % (n = 32)	Total % (N = 61) ^a
Should children in general undergo melanoma genetic testing?			
Yes	72.4	75.0	73.8
No	20.7	18.8	19.7
I don't know	6.9	6.3	6.6
Should your own children undergo melanoma genetic testing?			
Yes	50.0	86.7^b	69.0
No	46.4	13.1	29.3
I don't know	3.6	0	1.7
If some members of your family have tested positive, should their children undergo melanoma genetic testing?			
Yes	78.6	69.0	73.7
No	7.1	13.8	10.5
I don't know	14.3	15.6	15.8
Are your opinions about melanoma genetic testing the same for all your children?			
Yes	92.8	90.0	91.4
No	3.6	0	1.7
I don't know	3.6	10.0	6.9

^aThe proportions of responses in each column reflect the number of respondents who answered each question. All respondents answered the first question, but 1 noncarrier and 2–3 carriers did not answer some of the subsequent testing-preference questions.

^bProportion in bold denotes significant difference between carriers and noncarriers, $P < 0.01$.

tory would have values. Among participants with a melanoma history, participants who had melanoma diagnoses at earlier ages were more likely to support genetic testing for their own children than were participants who had their first melanoma diagnosis at later ages ($r = -0.48$, $P < 0.042$).

Uniformity of testing preferences for all of one's children

As presented in Table 3, participants overwhelmingly (91.4%) indicated that they would have the same opinions about melanoma genetic testing for all of their children. When asked

to explain their responses, 30.2% simply stated that children should be treated the same and that they did not see a reason why anyone would treat their children differently, 22.6% stated that all the children may be at risk and should be treated the same, 7.5% stated that all children should have the chance to protect themselves and understand their risk, and another 7.5% stated that it is important for all children to know their risk. Additionally, six noncarriers (11.3%) stated that their opinions and beliefs were the same for all of their children because they had themselves tested negative for the *CDKN2A/p16* mutation, and therefore, their children could not have inherited the mutation. One participant (1.7%) stated that his or her opinion would differ by child, mentioning that children who conduct proper and thorough skin examinations and engage in precautionary behaviors do not need to be tested for the *CDKN2A/p16* mutation because they are already compliant and safe. Four participants (6.9%) stated that they did not know whether their beliefs and opinions were the same for all of their children; of these, one participant said this is because it is up to the child to decide whether he wants to be tested; another stated that he can tell who has the gene; and finally, one stated that some children may not be able to fully understand and manage the implications of testing. Because of the high rate of endorsement for treating all of one's children the same, we did not perform logistic regressions to examine predictors of this outcome.

Preferences for testing children of carrier relatives

When we asked participants if they supported testing for children of their relatives who tested positive, the pattern of results for carriers and noncarriers was highly similar to that seen for beliefs about testing for children in general, with 73.7% in favor of testing, 10.5% opposed to testing, and 15.8% unsure (Table 3). As presented in Table 4, the logistic regression yielded no significant predictors of support for genetic testing of carrier relatives' children (all P values >0.10).

Preferences regarding minors' degree of involvement in the genetic counseling and testing process

Figure 2 presents the proportion of respondents who wanted their children to be involved in each of the following aspects of the genetic counseling and test reporting process: 62.3% said they would want their children to undergo genetic testing and learn the result, and 19.7% said they would want their children to have the test but not necessarily learn the results. In terms of genetic counseling and education about the management of melanoma risk, 45.9% indicated that they would want their children to attend a genetic counseling session similar to the one they had just completed, 59.0% said they would want their children to attend a general melanoma genetics education session, 62.3% said they wanted their children to be involved in a discussion of the impact of their genetic testing result on prevention and screening behaviors, and 62.3% said they would want their children to take responsibility for changing their own prevention and screening behaviors based on the genetic test result. Of note, 13.1% did not select any of the aforementioned options.

Preferences regarding age at which children should become involved in the genetic testing process

Respondents' estimates of average age at which children should become involved in any aspect of the genetic testing process ranged from 0 to 25 years, with a mean of 11.76 years ($SD = 5.13$). The most common response was age 12 years, listed by 27% of participants. Entered together, the demo-

Table 4 Logistic regression analyses predicting support for melanoma genetic testing of children in general, one’s own children, and children of carrier relatives

Testing-preference question	β	SE	Odds ratio	95% CI	<i>P</i>
Children in general					
Kindred	0.11	0.66	1.11	0.31–4.06	0.87
Melanoma history	−0.94	0.90	0.39	0.07–2.27	0.29
<i>CDKN2A/p16</i> status	0.92	0.83	2.50	0.49–12.66	0.27
Age (yr)	−0.02	0.02	0.98	0.94–12.66	0.34
Sex	−0.65	0.66	0.52	0.14–1.91	0.32
Children <18 yr	0.95	0.73	2.58	0.62–10.71	0.19
Own children					
Kindred	0.81	0.71	2.25	0.56–9.03	0.25
Melanoma history	−1.30	1.10	0.27	0.03–2.36	0.24
<i>CDKN2A/p16</i> status	2.63	1.03	13.82	1.82–104.96	0.01
Age (yr)	−0.02	0.03	0.98	0.93–1.03	0.36
Sex	0.07	0.73	1.07	0.26–4.42	0.93
Children <18 yr	−1.41	1.41	0.24	0.02–2.73	0.25
Children of carrier relatives					
Kindred	−0.01	0.67	0.99	0.26–3.71	0.99
Melanoma history	0.44	0.88	1.55	0.28–8.70	0.62
<i>CDKN2A/p16</i> status	−0.57	0.77	0.57	0.13–2.54	0.46
Age (yr)	−0.003	0.02	1.00	0.96–1.04	0.89
Sex	−0.73	0.68	0.48	0.13–1.83	0.28
Children <18 yr	0.47	0.78	1.61	0.35–7.43	0.54

graphic, medical history, and parental status variables did not significantly predict beliefs regarding the age at which children should become involved in the process ($F(6,43) = 0.88, P < 0.52$). Participants’ mutation status was a marginally significant predictor, such that participants who tested positive for the *CDKN2A/p16* mutation reported that children should become involved in the process at a somewhat later average age than participants who tested negative ($M_{pos} = 12.71$ years versus $M_{neg} = 10.61$ years; $\beta = 3.38, SE = 1.83, P < 0.08$). No other predictors in the model were significant (all *P* values > 0.10).

Qualitative data concerning testing preferences

The major categories and subcategories of the coding system, along with verbatim qualitative examples from respondents, are presented in Tables 5–7.

Reasons given in favor of melanoma genetic testing of minors

Overall, across the three eliciting questions (children in general, own children, and carrier relatives’ children), 53 participants (86.9%) expressed support for melanoma genetic testing of minors. Table 5 presents the most frequent reasons given in support of melanoma genetic testing in response to each of these three ques-

tions and for the total survey protocol. Overall, 77.4% of respondents who supported testing expressed the belief that it is important for a child to be aware of his or her own risk for melanoma, and 69.8% expressed the belief that testing would result in health benefits with respect to improved prevention and/or screening behavior. The next most frequent response given in support of testing was that information and knowledge are important to have for their own sake (24.5%). Additional responses given by fewer participants are summarized in Table 5.

It is important to note that these benefits were frequently mentioned by noncarriers. Although Table 5 is not stratified by mutation status, we specifically examined the reasons provided by the nearly 50% of noncarriers who wanted their own children to undergo genetic testing. Of these 14 noncarriers, 10 expressed the belief that testing would result in health benefits with respect to improved prevention and/or screening behavior, five expressed the belief that it is important for a child to be aware of his or her own risk for melanoma, one expressed the belief that information and knowledge are important to have for their own sake, and one expressed the belief that only older children should be tested. Only three stated in their open-ended responses that they had tested negative and could not pass on the mutation.

Table 5 Most frequent open-ended responses given in favor of genetic testing of children, their frequency of endorsement among participants supporting such testing, and qualitative examples of each category and subcategory of response

	Children in general % (n = 45)	Own children % (n = 40)	Carrier relatives' children % (n = 42)	Total % ^a (n = 53)	Qualitative examples of each coding category
It is important for child to be aware of own risk for melanoma	51.1	70.0	38.1	77.4	Because children are in the sun a lot and it would be great to know their risk.
Testing would result in health benefits	73.3 ^b	45.0	38.1	69.8	
Increase preventive behavior/reduce risky behavior associated with getting melanoma	55.6	35.0	28.6	58.5	Because it gives special attention to preventive measures they may not be taking seriously if it were not known.
Increase screening behavior so that early detection of melanoma is possible	17.8	7.5	7.1	26.4	(To) monitor skin changes/ moles.
Improve the child's health (unspecified)	11.1	7.5	4.8	13.2	For better health.
Information about the <i>CDKN2A/p16</i> mutation is important and useful	20.0	15.0	7.1	24.5	Further knowledge is always good.
It is important for the child to understand how the <i>CDKN2A/p16</i> mutation is passed down in the family	0	7.5	4.8	11.3	See how the gene is skipping through the family.
Testing may lead to melanoma research and/or prevention	0	2.5	7.1	7.6	If it may lead to research that can help others and possibly prevent melanoma.
Children are emotionally capable of receiving test results	0	5.0	2.4	3.8	I believe children can handle scary subjects like death better than most adults.
Children are cognitively capable of receiving test results	2.2	0	0	1.9	Children are smart enough to comprehend a need to protect one's self.

^aThe total column presents the unique proportion of respondents who mentioned a specific potential benefit of melanoma genetic testing in response to any of the five testing-preference questions (children in general, own children, children of mutation-positive relatives, whether all children would be treated the same, and other factors that would be considered in making a decision about testing for a particular child). A respondent who mentioned a particular benefit multiple times is counted only once in the total column. All proportions were calculated based on the number of participants who expressed support for melanoma genetic testing of minors.

Reasons given against melanoma genetic testing of minors

As reported previously (Table 3), approximately 20% of respondents opposed genetic testing for children in general. Table 6 presents the most frequently listed reasons against genetic testing of minors. Because reasons against melanoma genetic testing for one's own children differed between carriers and noncarriers, Table 6 is also stratified by mutation status. As presented in Table 6, most noncarriers who opposed genetic testing explained their decision by stating that mutation-negative persons could not pass on the gene. Specifically, of the noncarriers who indicated that they did not want their children to undergo genetic testing, 11 of 13 (84.6%) accurately stated in open-ended responses that their negative status meant that their children were not at risk for the mutation, thus precluding the need for genetic testing. Among mutation carriers who opposed testing of their own children ($n = 4$), two indicated that a child should make his own decision, with one noting that children should decide on their own as adults with

the input of a future spouse, and two indicated the test is unnecessary because they already encourage high levels of behavioral compliance in their children.

Overall, the next most frequent reasons given against testing for children in general included concerns that children were not cognitively or emotionally capable of receiving test results ($n = 5$, 8.2% of all respondents), a belief that children should not be tested because the risk of children developing melanoma is low ($n = 2$, 3.3% of all respondents), and specific concerns related to the autonomy of children (children should make own decision, $n = 3$, 4.9% of all respondents; only older children [$n = 4$, 6.6% of all respondents] or adults [$n = 2$, 3.3% of all respondents] should be tested). A small subset of all respondents (4.9%) indicated that they believed that testing would not change sun-related behavior, either because they already encourage high levels of compliance ($n = 2$) or because compliance should not depend on genetic test results in these high-risk families ($n = 1$).

Table 6 Most frequent open-ended responses given against genetic testing of children, their frequency of endorsement among participants opposing such testing as a function of respondents' *CDKN2A/p16* mutation status, and qualitative examples of each subcategory of response

	Children in general % (n = 12)		Own children % (n = 17)		Total ^a % (n = 24)	Qualitative example of each coding category
	<i>p16</i> - (n = 6)	<i>p16</i> + (n = 6)	<i>p16</i> - (n = 13)	<i>p16</i> + (n = 4)		
Lack of <i>CDKN2A/p16</i> mutation should preclude testing	66.7	0	84.6	0	54.2	
I tested negative/can't pass on gene	50.0	0	84.6	0	50.0	I tested negative therefore I cannot pass it on.
Testing should be done only if parent is a carrier	16.7	0	0	0	4.2	Should be tested if one of the parents has the gene.
Child's maturity level	50.0	33.3	7.8	0	20.8	
Children are not cognitively capable of receiving test results	33.3	0	0	0	8.3	Small children would not understand about protecting their skin from the sun.
Children are not emotionally capable of receiving test results	16.7	16.7	7.8	0	8.3	I think it would only make a child feel worried about something that can't change.
Risk of developing cancer is low as a child; therefore, the test should be postponed	16.7	16.7	7.8	0	8.3	Unless science can show that children may develop melanoma before age 18, then I do not believe children should be subject to testing.
Child's autonomy						
Child should make own decision	0	33.3	0	50.0	12.5	Whether they want to or not.
Only older children should be tested	16.7	16.7	7.8	0	16.7	Very small children I don't think should be subject to the test. Those around 13–18 could handle it.
Only adults should be tested	16.7	16.7	0	0	8.3	They can wait until 18 to be tested.
Future spouse and social support are necessary before deciding to undergo testing	0	16.7	0	25.0	4.2	Because their future spouse may have different thoughts, I want the child to be older & decide on their own.
Give child information about the risk of melanoma, but not <i>CDKN2A/p16</i> mutation status	16.7	0	0	0	4.2	It's good to know but only if it is kept confidential.
Parents do not want testing	0	0	0	0	4.2	Their parents do not want them to be tested.
Testing would not change behavior	66.7	0	84.6	0	50.0	
Test unnecessary because parents already encourage high levels of compliance	0	0	0	50.0	8.3	It wouldn't matter right now as we would make them all do the same things for sun protection.
Genetic test results would not change any sun-related behaviors	0	16.7	0	0	4.2	Because having the gene or not having the gene shouldn't change their sun behaviors.

^aThe total column presents the unique proportion of respondents who mentioned a specific potential liability of melanoma genetic testing of minors in response to any of the five testing-preference questions (children in general, own children, children of mutation-positive relatives, whether all children would be treated the same, and other factors that would be considered in making a decision about testing for a particular child). A respondent who mentioned a particular liability multiple times is counted only once in the total column. All proportions were calculated based on the number of participants who expressed opposition to melanoma genetic testing of minors.

Table 7 Respondents' views of other factors they would take into consideration about genetic testing for a particular child or grandchild and qualitative examples of each subcategory of response

	All respondents % (n = 53 ^a)	Qualitative example of each coding category
Child's maturity level	35.9	
Child's emotional maturity	15.1	Personality (extremely sensitive, a worrier, anxiety level of child, etc.).
Child's cognitive capabilities	9.4	Ability to understand a need to protect one's self.
Child's maturity level (unspecified)	17.0	Maturity of children. Readiness to deal with the information.
Child's personal history	28.3	
Child's phenotypic characteristics (moles, red hair, freckles, fair skin, and/or burns easily)	17.0	Red hair and freckles, especially easily sunburned.
Child's sun exposure history	9.4	How much sun exposure does the child receive.
Child's health or cancer history	5.7	If they had a cancerous mole removed already.
Whether testing would have health benefits	15.1	
Whether testing could be used to increase preventive behavior/reduce risky behavior in a particular child	11.3	We start teaching our children early ... so we can prepare for adulthood. Why not teach them about potential areas of concern within their bodies and how to prevent future problems.
Whether testing could be used to increase screening behavior so that early detection of melanoma is possible	5.7	Make (self-examinations) part of their regular lives.
Child's ability to perform prevention or screening behaviors	5.7	Their ability to ... take precautionary measures.
Only if a parent is a carrier	7.5	If the parent carries the gene, the children should be tested.
Child should make own decision	5.7	Whether they want to or not.
Only older children should be tested	1.9	If they object too strongly, let them be older.
Family should make own choice	1.9	It's a personal choice. I think each family should make that choice themselves.

^aFifty-three respondents provided answers to this open-ended question.

Factors respondents would consider in making a decision about genetic testing for a particular child

As presented in Table 7, participants listed several factors they would take into account when making a decision about genetic testing for a particular child. Specifically, 35.9% of respondents stated they would consider the child's emotional and/or cognitive maturity level, and 28.3% said they would consider the child's personal history, including phenotypic characteristics (17.0%), history of sun exposure (9.4%), and cancer or more general health history (5.7%). In addition, several participants indicated that they would take into account whether the test would be likely to have health benefits (15.1%), whether the parent is a carrier of the mutation (7.5%), and the child's own desire to be tested (5.7%). Additional responses are presented in Table 7.

Stability of parental preferences for genetic testing for their minor children at the 2-year follow-up among carriers and noncarriers

Carriers

As noted earlier, 22 participants with minor children or grandchildren (14 carriers and eight noncarriers) provided complete data regarding their testing preferences at the 2-year follow-

up. At 2 years, carriers reported strong preferences for genetic testing for their children ($M = 4.21$ of 5), with nine carriers (64.3%) endorsing the highest option on the scale, and no carriers indicating that they did not want their children to be tested.

We next examined the stability of parental preferences at 2 years, when compared with preferences expressed on the post-counseling questionnaire. Because the questions were asked on different scales (yes/no/unsure at post counseling and 1 = "not at all" to 5 = "very much" at 2 years), participant responses at the 2-year follow-up were recoded to "yes" if they answered 3 = "somewhat" to 5 = "very much" and "no" if they answered 1 = "not at all." Among carriers, preferences for genetic testing for one's own children were remarkably stable—13 of 13 carriers who indicated such a preference at the postcounseling assessment also did so at 2 years. Additionally, the lone carrier who did not want his or her children to undergo testing at the postcounseling assessment who provided data at follow-up reported "very much" wanting such testing at 2 years.

Noncarriers

We examined testing preferences in the smaller group of noncarriers with minor children. At 2 years, five of these eight noncarrier parents indicated that they had some interest in

genetic testing for their children younger than 18 years ($M = 2.75$ of 5), and three indicated that they did not at all want their children to be tested. When we examined the stability of such preferences, we found some evidence of change over time. Among noncarriers who initially opposed testing for their children, two did not want testing at 2 years, but three wanted their children to be tested at 2 years ($M = 4.00$). Among three noncarriers who initially wanted their children to be tested, two still wanted such testing at the 2-year follow-up, whereas one noncarrier no longer wanted such testing. We did not ask participants to explain their responses at the 2-year assessment; however, we examined the reasons these participants provided at the postcounseling assessment. Each of the three noncarriers who did not want testing at postcounseling but did at 2 years stated at postcounseling that because they had tested negative they could not pass the mutation on to their children. When the two noncarriers who wanted their children to be tested at both postcounseling and 2 years were asked why they supported testing, one said “Just to be sure” and the other, “Since it’s in my family.”

DISCUSSION

Both quantitative and qualitative data indicate that the vast majority of *CDKN2A/p16* mutation-tested adult members of two high-risk melanoma kindreds supported genetic testing for their own children or grandchildren, for the children of carrier relatives, and/or for children in general. In particular, immediately after genetic counseling and test reporting, 86.7% of mutation carriers reported that they wanted their children to undergo genetic testing. These preferences remained stable at 2 years, as all mutation-carrying participants who completed the follow-up who indicated at our initial assessment that they wanted their children to be tested also did so at 2 years. Although these follow-up data are based on a smaller number of respondents, we believe that the preferences expressed 2 years after parents’ own test reporting and counseling session are particularly meaningful, as these respondents have had considerable time to assess the risks and benefits of testing and to evaluate their preferences about testing for their minor children in light of their own experience.

Qualitative analysis of the reasons that participants gave in support of genetic testing of minors yielded two major potential benefits: increased awareness of melanoma risk and improved prevention and screening behavior. The highly frequent listing of the anticipated health benefits of genetic testing and test reporting suggests that participants support genetic testing for their children because they believe that their children will increase prevention and detection behavior performance once they know their mutation status, with some respondents noting that such behaviors learned in childhood would be more likely to carry through to adulthood. Without knowledge of individual genetic status or a personal history of melanoma, children and adults in high-risk families report levels of behavioral adherence similar to individuals in families at population risk.^{27,31,46,47} Thus, parents may believe that additional motivation in the form of a genetic test result will prompt their children to protect themselves from the sun. Such expectations are similar to the results reported by Kasparian et al.⁴² in which one third of interviewed adults from high-risk families believed that genetic testing would motivate increased prevention behavior.

A minority of respondents—approximately 20%—opposed melanoma genetic testing of minors in general. It is important to note that the majority of respondents who reported that they did

not want their children to undergo testing were individuals who had tested negative for the mutation, and the majority of these accurately stated that their negative status rendered them unable to pass the mutation on to their children. Nonetheless, 50% or more of the noncarriers indicated that their children should undergo melanoma genetic testing at either the initial assessment or the 2-year follow-up. The prevalence of support for testing and the overall inconsistency of noncarriers’ beliefs from our initial assessment to the 2-year follow-up suggest that additional educational sessions might be appropriate for noncarriers from melanoma-prone families to ensure accurate understanding of heritability. However, other reasons may account for such preferences; for example, respondents might believe that it is important or impactful for each child in a high-risk melanoma-prone family to see his or her own specific results and to receive intensive counseling regarding familial risk and its management.

The next most frequently cited reasons to oppose genetic testing of minors were the concern that children are not cognitively or emotionally capable of receiving test results and the belief that risk of developing cancer as a child is low. It is important to note that such concerns were cited by a very small proportion of the overall sample, suggesting that although such concerns are clearly important, they are not widely shared. Further, as noted earlier, melanomas have been detected in minors from high-risk families,³ suggesting that improved screening might confer benefit. Similarly, given emerging evidence about the role of childhood and adolescent sun exposure in the etiology of melanoma,¹¹ it is likely that children and adolescents would also benefit from improved practice of photoprotection even though melanoma may not develop in some cases until later in life.

Respondents overwhelmingly stated that they would treat all of their children the same when considering genetic testing, suggesting that respondents did not believe that there are subsets of children who should not undergo testing. However, respondents listed multiple characteristics of the child they would consider in determining when to test, such as emotional or cognitive maturity (e.g., the child’s propensity to worry), the child’s phenotype, sun exposure or health history, numerous aspects of the child’s autonomy with respect to the decision, and whether the test results would be necessary to promote or maintain behavior change. Genetic counselors might consider such issues with high-risk patients by aiding parents in determining when and whether to test children.

Implications for understanding the costs and benefits of melanoma genetic testing of minors

With respect to various aspects of the debate about genetic testing of minors, it is important to note that concerns about inducing psychological distress and compromising the autonomy of minors were relatively infrequent in our protocols. Notably, although more than one third of respondents said they would take the child’s maturity level into account in making a decision about genetic testing, only two respondents opposed testing for all children because they believed it would induce worry and distress. Similarly, only a few respondents expressed the idea that it may be better for a child to make a decision about genetic testing as an adult or that the child may not want to be tested. Thus, with a few exceptions, participants’ responses did not seem to correspond to some of the major concerns about genetic testing of minors.^{2,28,29} Although some medical professionals are concerned that a child who undergoes genetic testing is deprived of the right to decide as an adult whether this is

information he wants, only three participants in our sample gave this reason. Instead, as noted earlier, the majority of respondents indicated that they believed that genetic test results would improve prevention and screening behaviors in children younger than 18 years. Thus, these responses may be consistent with the argument that parents or genetic counselors who do not let minors undergo genetic testing are depriving the minor of health information that could be used to make behavioral choices as adolescents.³⁰

Interestingly, in contrast to findings for FAP,⁴⁰ the potential benefit of reduction of uncertainty was not explicitly mentioned by our respondents. Instead, respondents expressed a more general preference for having information about their child's mutation status, either because it would be important to alert a child to his or her risk or for the family to have this information. This strong desire for genetic test results is consistent with the results of interview studies conducted in members of Australian *CDKN2A/p16* mutation-carrying families.⁴²

Finally, because all individuals who undergo testing may still be at elevated risk for melanoma, some researchers have argued that testing to distinguish between carriers and noncarriers is unnecessary.^{22,23} However, only a few respondents mentioned that the genetic test result should not be a determinant of whether children should protect themselves in the sun, because all children should do so regardless of mutation status. Instead, the vast majority of respondents reported believing that a positive test result would be useful in improving sun protection and screening behaviors in their children and grandchildren.

Strengths and limitations of the qualitative assessment of parental preferences for genetic testing of minors

Respondents expressed reasons for and against melanoma genetic testing of minors in written answers to open-ended questions. Thus, respondents were required to articulate their own reasons, rather than to indicate their agreement with statements that were provided to them as part of a structured inventory. It is possible that respondents may have expressed greater agreement or disagreement with the different reasons described in this study if we had used structured inventories (i.e., that more respondents might have expressed agreement or disagreement with a particular statement had it been presented to them). However, one advantage of the qualitative approach taken in the present assessment is that we can be confident that these answers represent participants' own reasons and not those suggested or seemingly endorsed by researchers through their inclusion on a list of potential reasons families should consider in making decisions about genetic testing.⁴⁸ Future studies might profitably examine whether different rates of agreement with the various reasons for and against melanoma genetic testing of minors are obtained using a structured quantitative assessment based on the reasons reported in this study.

Implications for options for melanoma genetic counseling and testing of minors

When we asked participants whether they would like their children to be involved in the genetic testing process, more than 60% wanted their children to receive both testing and test results. Respondents may be considering specific aspects of melanoma prevention and early detection, as parental preferences in our study differ considerably from those of respondents counseled for breast cancer at the same institution. Approximately 25% of individuals who had undergone genetic testing for breast cancer supported testing for children in general, and

less than one fifth supported testing for their own children⁴⁹; other studies of *BRCA1/2* mutation carriers have yielded similar results.⁵⁰ These parental preferences for *BRCA1/2* testing of minors cohere with medical guidelines that recommend against testing children for adult-onset conditions for which no medical intervention or established prevention measures are available. Genetic testing for melanoma differs from genetic testing situations such as these because there is a well-established and avoidable environmental influence on melanoma development and because melanoma outcomes can be improved by self- and physician-performed skin screenings. Our findings suggest that genetic conditions with associated prevention measures may require a new model for genetic counseling and testing in minors, particularly if the testing process is shown to improve compliance with these prevention measures.²⁷

A significant minority of respondents wanted the genetic test performed but did not want children to receive the results. Withholding genetic information from children is not generally considered standard of care, and there are no data concerning the prevalence or the short- or long-term impact of this approach. Current recommendations for genetic testing of minors suggest that the child participate in the pre- and posttest counseling process on an age-appropriate basis and that counselors and parents agree on a plan to disclose results to the children during the pretest counseling.⁵ If parents are unwilling to inform children of test results, then it is recommended that testing be postponed until parents believe their children are ready to learn that information.⁵

However, parental autonomy should be respected in addition to the child's autonomy when making decisions about genetic testing and reporting.⁵¹ Parents may find the option of testing but not reporting results to the child attractive because they believe the child could benefit from behavior change that could occur without the child knowing his or her actual test result. Parents could implement photoprotection, regular skin examinations, and biopsy of suspicious lesions for both carrier and noncarrier children without the child knowing his or her genetic status. It may be difficult for parents to withhold information from children about other diseases because of extensive medical management, which could prompt the child to ask questions (e.g., colectomy in FAP), but these practical barriers are less applicable to melanoma. Although withholding information from the child may be logistically possible, we currently caution against informing the child in a context away from the genetic counseling session or at a date too far removed from the original counseling session, so as to guarantee that the child receives accurate information and appropriate support. Again, our results suggest that the model of complete disclosure of genetic test results to minors with FAP may not be completely satisfactory for familial melanoma.

Of the respondents who wanted their children to be tested but not to receive the test results, two thirds indicated that they would want their child to be involved in a counseling or education session, or have a discussion regarding prevention and detection behaviors. This suggests that parents plan to give their children some information but believe that the child should not be given the actual test result. Accordingly, many respondents mentioned that they would consider the child's emotional and cognitive maturity when making decisions about testing. In fact, it is recommended that genetic counselors tailor the counseling protocol to the developmental stage of the child.⁵⁰ Genetic counselors could facilitate tailored disclosure by adopting a flexible, multistep procedure in which parents are first informed of the child's test result, so they may react without the child present and ask questions that are more detailed than the child

could likely understand. In a subsequent meeting with both parent(s) and child, the genetic counselor would present the child with age-appropriate risk-management information (e.g., discuss social issues with peers regarding a child's practice of prevention behaviors). Additionally, because genetic test results will have lifelong implications, ongoing genetic counseling interventions ("booster" sessions) could reinforce appropriate behaviors and introduce developmentally appropriate information, such as specifics of passing the mutation onto one's future children. One respondent's comment particularly exemplified the spirit of such ongoing counseling: "Maybe testing [should be done] for the parents to know from birth to start preventive steps and then more education as the child matures to take responsibility for themselves to watch for changes, etc."

Finally, several respondents said they would consider issues of the child's autonomy, and we also recommend involving the child in the decision-making process at an age-appropriate level.⁵⁰ Future research might profitably examine the experiences of adults who were either informed or not informed of their genetic risk as children to identify additional risks and benefits families may wish to consider in making such decisions.⁵² As our ability to prevent genetically defined disease improves, new models that take the benefit of increased adherence to prevention guidelines into account will become increasingly important as we investigate the impact of melanoma genetic counseling and testing in minors.

Limitations and generalization to other samples

Our respondents were a relatively homogenous sample of well-educated and affluent white individuals, nearly all of whom reported an affiliation with The Church of Jesus Christ of Latter-day Saints. Although we believe that these kindreds are representative of the high-risk melanoma families in Utah, the generalizability of our findings may depend on these demographic factors, as well as cultural and religious beliefs regarding both the value of genetic testing and the meaning of cancer and other illnesses.^{53,54} That our respondents were willing to receive genetic counseling and their own test results may also indicate that our sample is biased in favor of genetic testing and, thus, potentially unrepresentative of the larger population of high-risk individuals (although still representative of a population who might elect clinical testing). Finally, it should be noted that our respondents were drawn almost exclusively from two extended kindreds. Although no statistically reliable differences in testing preferences between the kindreds were found and there did not seem to be differences between the kindreds in the relationship of other demographic and medical history variables to testing outcomes, it will be important to investigate beliefs and preferences in a larger and more diverse set of families.

Study strengths were the inclusion of both unaffected and affected family members, including several with a melanoma diagnosis in young adulthood, and the wide age range of the sample and its corresponding inclusion of parents, grandparents, and respondents with no children, all of whom had undergone genetic counseling and test reporting themselves. None of the findings reported in this study were qualified by age, melanoma history, parental status, or sex of respondent, although earlier age of onset predicted greater support for melanoma genetic testing of minors among affected participants. It is important to note, however, that none of these respondents had undergone specific counseling regarding the pros or cons of, or approaches to, genetic testing of minors; thus, this study represents participants' own thoughts and ideas on this issue. It is unknown whether the testing preferences reported in this study would be altered by more detailed counseling about this issue. An under-

standing of the degree to which these findings about preferences for melanoma genetic testing of minors may generalize to other groups at risk for melanoma or to other cancer syndromes awaits future study.

Another potential limitation of the study concerns the interpretation of the apparent stability of testing preferences at the 2-year follow-up. Although we have interpreted participants' strong support for melanoma genetic testing of minors over a 2-year period as evidence of consistent belief in the utility of such testing, other factors may account for the consistency of responses, especially among mutation carriers. Specifically, it is possible that support for such testing represents a post-hoc rationalization of participants' decisions to learn their test results. Although we cannot know for sure whether participants regretted their decision to learn their melanoma genetic test results at any point after their counseling session, we can say that participants' responses to questions concerning multiple aspects of their responses to genetic test reporting, such as responses to an inventory specifically designed to yield both negative and positive outcomes of learning one's test result (the Multidimensional Impact of Cancer Risk Assessment inventory),⁵⁵ reveal no evidence of such regret. Direct open-ended questions concerning participants' evaluations of the pros and cons of having received the results similarly revealed few reported cons.

Finally, it should be emphasized that these parental preference data cannot inform us about the outcomes of melanoma genetic testing of minors or about minors' own preferences with respect to testing and counseling. Future research should examine psychological and behavioral outcomes of minor children in *CDKN2A/p16* mutation-carrying families in which a parent has received a positive test result and survey minors themselves about whether and when they would like to undergo testing. Additionally, the behavioral and psychological outcomes of any minors who do receive melanoma genetic testing should be prospectively monitored to evaluate both the short- and long-term costs and benefits of such testing.

CONCLUSION

In our sample, a substantial majority of *CDKN2A/p16* mutation-tested individuals expressed strong and consistent support for melanoma genetic testing of minors over a 2-year period. The majority of respondents indicated that children would both be informed about their melanoma risk and improve their prevention and screening behaviors and overall health as a result of the genetic test. Relatively, few respondents expressed concerns that genetic testing would have substantial risks for minors in terms of psychological distress or harm. We believe that parental preference data such as these may play an important role in shaping formal recommendations for melanoma genetic testing of minors.

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