Barriers to risk-understanding and risk-reduction behaviors among individuals with a family history of melanoma

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Practice points

- First-degree relatives of melanoma patients are at increased risk of developing melanoma themselves. Individuals with two or more affected first-degree relatives are at an even higher risk.
- Individuals with a family history of melanoma are frequently unaware of their risk and do not take steps to reduce it.
- Interventions in order to improve primary and secondary prevention behaviors in this population have demonstrated no clear consensus on the optimal approach. Further research is needed into ways to assist this at-risk population.

SUMMARY Family members of melanoma patients are often called upon to provide support, ranging from monetary to medical assistance. Consanguineal relatives of melanoma patients are also at greater risk of developing the disease themselves. However, as a group, they have limited understanding of their melanoma risk and they demonstrate inadequate primary and secondary prevention behaviors. The optimal intervention strategies for improving the consistent use of such behaviors (i.e., improving rates of sun-protection behaviors and screening) remains unclear, necessitating further research.

Melanoma incidence is rising [1], and with it, the number of family members affected by a relative's diagnosis. First-degree genetic relatives of melanoma patients, including parents, children and siblings, are at an increased risk of developing melanoma themselves [2]. However, as we will review, individuals with a family history of melanoma frequently neither fully understand their risk nor take steps to reduce it. Understanding the barriers to risk assessment and to the uptake of risk-reduction strategies is essential to the individual clinician for developing a clinical framework in order to promote understanding of risk and the adoption of risk-reduction strategies in this population. We review the literature on this topic, including information on factors affecting family members' risk perceptions, facilitators and barriers to prevention behaviors and interventions to improve such behaviors.

Defining family history

Individuals who are considered to have a family history of melanoma can be divided into two groups: at-risk and high-risk individuals. At-risk individuals are those with a single first-degree relative (parent, child or sibling) with melanoma. This group, which accounts for up to 8% of people with melanoma, has a melanoma risk that is more than twice that of the general population [3–5]. Teasing out the degree of environmental versus genetic causation is difficult, however, because melanoma

KEYWORDS

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risk-associated habits (e.g., sunny vacations) and phenotypes (e.g., light hair and eyes, light skin, mole number and skin type) both tend to run in families [6].

A smaller group of individuals with two or more first-degree relatives with melanoma are part of so-called ‘high-risk’ families, accounting for 1–2% of all melanomas [5]. Definitions of melanoma-prone families vary, but the presence of the cancer in more than two family members over two or more generations may indicate a familial cancer syndrome [7]. Some of these high-risk families probably harbor genetic mutations with incomplete penetrance that predispose them to melanoma. High-risk mutations in identified susceptibility genes, such as *CDKN2A/p16* and *CDK4*, confer a lifetime melanoma risk that is estimated at 28–91%, as compared with the risk of melanoma in the general population, which is 0.1–2% [8–13]. Individuals with these mutations may also be at higher risk of prostate, breast and colon cancers, as well as non-Hodgkin’s lymphoma and multiple myeloma [2]. Another group of high-risk individuals for melanoma (as well as other malignancies; e.g., pancreatic cancer) are those with familial atypical multiple mole melanoma syndrome, an autosomal-dominant disorder generally characterized by greater than 50 melanocytic nevi and a family history of melanoma [14].

Recommended risk-reduction behaviors & limited adherence

Recommended primary prevention (i.e., minimizing ultraviolet exposure) and secondary prevention behaviors (i.e., appropriate surveillance according to risk level, as well as education for early detection) differ depending on whether an individual is at risk or high risk based on their family history. For the at-risk group, the American Academy of Dermatology recommends annual full-body skin examinations [15]. This group is also advised to be especially adherent to daily sun-protection strategies, including sun avoidance at peak hours, shade seeking, use of hats and clothing to block ultraviolet radiation exposure and sunscreen on exposed body parts [16,17].

For members of high-risk families, the Melanoma Genetics Consortium recommends full-body skin examinations starting at 10 years of age. These examinations should be repeated every 6 months until the baseline nevi are considered to be stable, at which point examinations may become yearly. These individuals should

also perform monthly self-skin examinations or be examined by a parent, family member or partner [18]. Genetic testing for the *CDKN2A/p16* mutation is available commercially and offers the promise that test results might one day lead to tailored recommendations for primary and secondary prevention. However, melanoma is unique in that shared familial factors (e.g., sun-protection behaviors or phenotype) may increase the melanoma risk for members of high-risk families, independent of genetic test results [19]. Therefore, the test result – whether positive or negative – does not currently change the recommended clinical management of individuals from high-risk families [20,21].

Unfortunately, many studies have found that inconsistent sun protection – including use of sunscreen, protective clothing and hats, as well as shade seeking – is typical of individuals with a family history of melanoma. One study found that among first-degree relatives of malignant melanoma survivors, the mean score on a sun-protection practices scale corresponded to ‘sometimes’ [22]. Less than a third of first-degree relatives of melanoma survivors in one study used sunscreen routinely when in the sun; even fewer stayed in the shade or used protective clothing frequently [23]. One study found that adolescent children of mothers with a history of skin cancer (including melanoma) were slightly more likely to report frequent sunburns in the past year compared with peers, and approximately equally likely to report tanning bed use [24]. Another study determined that approximately half of children with a family history of melanoma in one study had recently been sunburned [25]. One study found that a smaller percentage of individuals with a family history of melanoma when compared with controls reported a sunburn (31 vs 41%) or suntan (55 vs 60%) the prior summer; however, the absolute percentages were still disappointingly high for this at-risk population [26].

Sun-protection behaviors for individuals from high-risk families are equally disappointing. In one study of young, unaffected members of high-risk families, a third of individuals reported sunbathing ‘often’ or ‘very often’, and 35% reported current tanning bed use [27].

Rates of screening behaviors (both self-screening and physician screening) are also relatively low among family members of melanoma survivors. Despite recommendations that individuals with a family history of melanoma receive regular full-body skin examinations (for both

the at-risk and high-risk groups) and perform monthly self-skin examinations (for the high-risk group) [28], studies have found that as many as 70% of first-degree relatives of melanoma patients had not performed a self-skin examination and 45% had never had a total cutaneous examination by a healthcare provider [22,29].

High-risk families are no more consistent with screening: a study of individuals with known *CDKN2A/p16* mutations found that 15% had not performed a skin self-examination in the past 12 months and 27% had never undergone a clinical skin examination by a healthcare provider [30]. Similarly, a study of members of familial atypical multiple mole melanoma syndrome families found that 8% had never performed a skin self-examination [31]. This lack of surveillance leads to multiple missed opportunities for the earlier detection and treatment of melanoma in individuals with a family history: one study found that a personal melanoma history – but not a family history of melanoma – was associated with a thinner Breslow depth at melanoma diagnosis, which is a key prognostic factor [32]. The key clinical question raised by these findings relates to why individuals with a clearly elevated risk of melanoma choose not to take steps to identify disease at an early stage.

Barriers to melanoma risk assessment in individuals with a family history

The most glaring factor that may prevent individuals from the at-risk (as opposed to high-risk) group from understanding the melanoma risk conferred by their family history is a lack of awareness or understanding of their family history. The percentage of patients who report having melanoma risk discussions with their families has been found to range from 50 to 94% [33,34]. The attributes of both the patient and the family member have been found to affect the likelihood that such a conversation will occur. For example, in one study, patients whose melanoma arose on a body site not exposed to the sun shared less regarding their disease, perhaps because they felt that sun-protection behavior was less likely to prevent family members from developing a similar melanoma [35,36]. Another study found that patients were more likely to have a discussion regarding melanoma risk with female family members [33]. A survey of mothers with melanoma found that their reasons for telling their children of the increased risk included a similar physical appearance (including skin type) and the child's

skin cancer risk behavior, such as deliberate tanning (56%) [29]. Similarly, in a focus group study on melanoma patients, survivors reported singling out certain family members they perceived to be at high risk for risk and prevention discussions [37]. It may be beneficial for clinicians to encourage patients to share the news of their diagnosis and initiate a risk discussion with all first-degree family members. Family dynamics have also been found to both promote and hinder conversations regarding melanoma. Factors associated with open communication about melanoma include familial cohesion, adaptability and strong beliefs regarding melanoma risk [33]. Individuals who are members of families that consider themselves to be health conscious also have more conversations regarding melanoma with their family members [35]. Conversely, barriers to communication regarding melanoma risk have been found to include little time to talk, geographic separation from family members, the feeling that the conversation would overburden an elderly and ill family member and the feeling that “whatever happens, it's up to the good Lord now” [38]. Awareness of the factors that may help determine which individuals are more likely to share news of their illness with family members is essential for clinicians, who are in a unique position to encourage individuals to disclose their melanoma diagnosis to relatives. Similarly, it may be beneficial for clinicians to elicit information regarding family communication dynamics that may be functioning as barriers to conversations about melanoma risk and encourage individuals to surmount them for the purpose of conveying important health information.

Simply ensuring that the individual discloses his or her diagnosis, however, is not sufficient. Clinicians should also work to ensure that patients and their family members understand the risk conferred by the family history, given research that suggests family members frequently do not. In studies of first-degree relatives of individuals diagnosed with malignant melanoma, only 60–64% of respondents considered their risk of malignant melanoma to be ‘a little’ or ‘much’ higher than that of others [22,39]. In one of these studies, less than half of the first-degree relatives surveyed believed that heredity/genetics contributed to their melanoma risk, compared with 80% who cited ultraviolet radiation exposure as a factor. Participants with higher education levels were more likely to cite heredity/genetics as a factor in their melanoma risk [39]. A study on members of families with multiple cases of

melanoma found that while 79% considered themselves to be at increased risk for melanoma as compared with peers, some (as many as 39% in Sweden) believed themselves to have a similar or lower risk. Participants with multiple moles and freckles were more likely to believe themselves to be at increased risk for melanoma [40].

As relates specifically to individuals from high-risk families, genetic testing may be a particular point of misunderstanding. Genetic testing to identify germline mutations in *CDKN2A* is available both in the USA and abroad. However, the clinical utility of such testing is controversial. In a study that followed unaffected individuals who received *CDKN2A/p16* genetic test results over a 2-year period, individuals testing positive did not change their estimations of their lifetime risk; individuals testing negative, however, demonstrated a significant decrease in their lifetime risk estimation immediately following receipt of their test results, followed by a rebound, so that 1 month following the test, their estimation of their lifetime risk did not differ from baseline [41]. In the area of behavior change, one study found that following *CDKN2A/p16* genetic test reporting and genetic counseling, intentions to practice sun-protection behaviors, as well as the implementation of new sun-protective behaviors, increased in all participant groups, regardless of the test results [42]. Similarly, a study of 37 members of *CDKN2A/p16* families found that, 2 years following genetic testing, adherence to recommendations for annual full-body skin examinations increased among unaffected carriers from 40 to 70%. However, adherence among unaffected noncarriers decreased from 56 to 13% [21]. Given the multifaceted nature of melanoma risk (i.e., the contribution of nongenetic shared familial risk factors; e.g., sun-related behaviors and phenotype), these individuals may still be at an increased risk of developing melanoma, meaning they should still abide by the rigorous screening recommendations for the high-risk population. Thus, more research is needed on how to prevent reductions in adherence to screening recommendations following a negative genetic test result and on the overall benefit of genetic testing with regards to behavioral change [21].

Barriers to melanoma risk reduction behaviors in individuals with a family history

The limited understanding of melanoma risk reviewed above is surely a major factor leading

to the inconsistent prevention and screening behaviors among individuals with a family history of melanoma. However, a wide range of contextual factors have also been identified. For at-risk individuals (i.e., those with a first-degree relative with melanoma, as opposed to individuals from high-risk families), facilitators of sun protection behaviors have been found to include being at a beach, pool or other water setting and sunny weather. Some at-risk family members report being more likely to use sunscreen during physical activity (e.g., while golfing or gardening), while others are actually less likely to use sunscreen under these conditions. At-risk family members are also less likely to use sun protection when they anticipate shade being available [43].

In another study, at-risk individuals who reported higher sun protection tended to be more educated, to report greater sunscreen self-efficacy (a measure of the belief in one's own ability to use sunscreen) and to be more concerned about photo-aging and about social norms regarding sun protection. Conversely, at-risk individuals reporting more sunbathing were more likely to be younger, female and to perceive more benefits of sunbathing, among other factors [44]. These findings may help identify family members who are less likely to practice sun-protection behavior and target them for extra counseling.

In the area of screening among at-risk individuals, one study found that the children of mothers with melanoma or breast cancer who reported open communication about cancer risk with their mothers took significantly more action to screen themselves for cancer than children who did not have open conversations [29]. Another study demonstrated that only 46% of siblings of melanoma patients told their primary care doctor about their sibling's melanoma within 2 months of diagnosis; however, such a conversation was associated with a higher likelihood of performing skin self-examination and undergoing an examination by a dermatologist. A similar association was observed for communication with dermatologists [45]. These findings suggest that it may be beneficial for clinicians, including primary care physicians, to directly ask their patients about whether they have a family history of melanoma in the course of eliciting a family history of other illnesses.

Indeed, inadequate communication with physicians has been explicitly demonstrated to contribute to both inconsistent sun-protection behavior and inconsistent screening among at-risk family members. Nearly 85% of dermatologists report

that they often or always communicate risk to individuals with melanoma regarding their first-degree relatives; however, less than 50% routinely offer to screen first-degree relatives who live nearby, and less than 20% use medical record reminders to note communication of melanoma risk to family members [46]. In a study of mothers with either melanoma or breast cancer, no mothers with melanoma reported that a physician's recommendation was a factor in their decision to tell their child of their increased risk for melanoma, as compared with 30% of mothers with breast cancer [29].

Similarly, in a study of high-risk families' perceived melanoma risk communication from healthcare providers, participants reported few provider discussions of melanoma risk or risk-modifying behaviors, despite a desire to trust such information from providers [47]. These findings suggest that physicians may be able to make a positive impact by explicitly recommending screening behaviors; however, this idea remains untested.

Interventions to improve sun protection & skin surveillance behaviors

There is no clear consensus regarding the best way to effect behavioral change in the areas of sun protection and skin surveillance for family members of melanoma patients. A randomized trial found that tailored educational materials and telephone counseling were more effective than usual care at improving self-skin examination among the siblings of melanoma patients, but not at improving sunscreen use or the frequency of physician screening [48].

Another study of first-degree relatives of melanoma patients found that, when compared with a generic intervention, a tailored intervention resulted in greater improvements in sun-protection behaviors and a greater likelihood of undergoing a total cutaneous skin examination, but that the tailored and generic interventions were equally effective at improving

self-skin examination behaviors. In both groups, even behaviors that were improved remained inconsistently practiced [49].

Conclusion & future perspective

Individuals with a family history of melanoma – whether with a single affected family member or multiple family members – are at greater risk for developing the disease themselves. However, they are frequently unaware of their risk, a knowledge gap that, along with other factors, translates into inconsistent sun-protection and skin-surveillance behaviors among family members of melanoma patients.

Further research is needed into effective interventions in order to improve family members' risk awareness and protective or surveillance behaviors. Additional studies are also needed into ways to improve physician communication with family members regarding melanoma risk. Furthermore, melanoma is not a single, unified disease; rather, there are many forms, including acral lentiginous melanomas and ocular melanomas, for which sun exposure may be a less important risk factor, and therefore the cost–benefit calculation for genetic testing, for example, may be different between forms of melanoma. As such, additional research exploring the specific family issues involved with the diagnosis of these less common types of melanoma would be valuable and may reveal unique needs and issues among these families affected by melanoma.

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