Addressing Melanoma Risk in Families

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Addressing Melanoma Risk in Families

Rebecca B. Roy

A scholarly paper submitted to the faculty of
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Master of Science

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ABSTRACT

Addressing Melanoma Risk in Families

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Melanoma is one of the top 10 cancers in the United States. It is an aggressive and potentially fatal skin cancer. Up to 10% of all people diagnosed with melanoma have a positive family history of melanoma and a family history with a first degree relative can increase the risk for family members by 75%. Poor family communications often result in first degree relatives’ lack of knowledge about risk and prevention for melanoma. Considering this lack of knowledge, primary care providers have the opportunity to assess risk; teach and encourage prevention measures; teach and perform skin exams; and refer to dermatology. Therefore, the purposes of this paper are to: (1) describe familial melanoma and families at risk for melanoma, (2) discuss communication about melanoma within families, and (3) discuss clinical implications for primary care providers related to identifying those at risk for melanoma. Collaborative care for families with melanoma risk can increase the chance of diagnosing melanoma in the earlier stages and decrease the risk of mortality associated with melanoma.

Keywords: melanoma, family history of melanoma, risk factor, skin cancer, familial melanoma, melanoma genetics, CDKN2A, CDK4
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Introduction

Melanoma is an aggressive and potentially fatal skin cancer. Melanoma is one of the top 10 most common cancers diagnosed in the United States (U.S. Cancer Statistic Working Group, 2020). People with a positive family history of melanoma are at an increased risk and require more extensive screening. Most patients are not aware they are at higher risk for melanoma and require additional screening and prevention measures. Primary Care Providers (PCPs) play an important role in identifying people at-risk and directing the screening measures. Melanoma survival improves when individuals are aware of their risk and ways to modify it (Loescher et al., 2009). This paper will describe familial melanoma and discuss clinical implications for PCPs caring for patients at risk for melanoma.

Familial Melanoma

Familial melanoma is a genetic condition where elevated risk of melanoma can be inherited (Nijsten, 2016). The diagnosis usually applies to families in which two or more first-degree relatives have been diagnosed with melanoma. Approximately 10% of all people diagnosed with melanoma have a positive family history of melanoma (Nabil et al., 2018; Read et al., 2016; Rossi et al., 2019). Further breakdown of these numbers includes an estimated 8% of individuals with melanoma have a first-degree relative with melanoma. One to two percent of individuals with melanoma have two or more close relatives with melanoma (Bishop, 2002).

The clinical presentation of people diagnosed with familial melanoma is different from people with non-familial melanoma. Individuals with familial melanoma tend to be diagnosed in their thirties compared to those with non-familial melanoma who are diagnosed in their fifties (American Society of Clinical Oncology [ASCO], 2020). Additionally, individuals with familial melanoma exhibit an increased prevalence of squamous cell carcinoma and Spitz nevi, also
known as melanocytic BAP-1 mutated atypical intradermal tumors (MBAIT) (Huerta et al., 2018). Having either squamous cell carcinoma or atypical nevi would increase the likelihood of familial melanoma and the need for genetic testing (Rossi et al., 2019).

People may be predisposed to familial melanoma through specific genetic mutations. The most common mutations occur in the CDKN2A and CDK4 genes, which account for 20-40% of gene mutations in melanoma families (National Comprehensive Cancer Network, 2020; Rossi et al., 2019; Taylor et al., 2019). Mutations in other genes for melanoma are rare, accounting for 10% of familial clustering of melanoma. Despite having identified several high and intermediate risk variants, causative or contributory gene variants are only identified in about 50% of familial melanoma cases (Read et al., 2015). Thus, research for genetic mutations associated with melanoma is ongoing.

Families at Increased Risk for Melanoma

It is important to be aware of patients who meet the strict criteria for familial melanoma—having two first-degree relatives diagnosed with melanoma—and those who may carry specific genetic mutations. Focusing only on these patients, however, misses an important group at increased risk, including anyone with family history of melanoma (Ransohoff et al., 2016). For example, having just one first-degree relative diagnosed with melanoma increases lifetime risk of melanoma by 75% (Wei et al., 2019). Individuals with a family history of melanoma are at increased risk regardless of whether they are found to carry a melanoma-associated mutation (Ransohoff et al., 2016).

In addition to considering family history of melanoma in relation to risk, it is also important to consider shared family traits and environment. Most cases of melanoma are related to a person’s phenotype and environment. Family members often share similar phenotypes
including hair, eye and skin color. Phenotypic expressions are important factors when considering melanoma risk. For example, phenotypic traits such as, fair skin, freckles and red hair can increase melanoma risk by two-fold (Ransohoff et al., 2016). Lack of melanin is a risk factor for melanoma since melanin blocks some of the harmful effects of UV radiation. Fair skinned patients have less melanin and therefore less protection and cannot block the harmful effects of UV radiation (D'Orazio et al., 2013).

While phenotype plays an important role in melanoma, environmental factors are equally important. Families often share the same environmental exposures. Sun exposure is the leading cause of melanoma. Eighty-six percent of all melanomas can be attributed to exposure to ultraviolet radiation from the sun (Parkin et al., 2011). Patients who live or grow up in sunny climates or higher elevations are at greater risk for melanoma (Richards et al., 2011) because they are more likely to experience sunburns. In a study of 108,916 women from the United States, those who had five or more blistering sunburns between the ages of 15 and 20, compared to those that had none, were 80% more likely to develop melanoma (Wu et al., 2014). The association between number of sunburns and melanoma risk, however, does not end at age 20. Risk for melanoma increases with the total number of sunburns during all life-periods (Dennis et al., 2008). Thus, both total number of sunburns, severity of sunburns, and age during development of sunburns may play a role in melanoma development.

The increased risk of melanoma is not limited to UV light from sun exposure. Tanning bed lamps also emit UV light and contribute to melanoma risk. There has been an increase in melanoma over the last 20 years in young women who frequent tanning beds (Zhang et al., 2012). Tanning beds are frequently used by multiple members of a family. Studies show that if a
parent uses a tanning bed, children, especially daughters, are more likely to use tanning beds (Falzone et al., 2017).

The US Department of Health and Human Services acknowledges that individuals who start using tanning beds in early adolescence or young adulthood are at higher risk of melanoma (U.S. Department of Health & Human Services [HHS], 2014). A person does not need to use tanning beds frequently for it to pose a threat to his/her health. One indoor tanning session before the age of 35 increases a person’s risk of melanoma by 75% (Boniol et al., 2011). One study found that out of 63 women who were diagnosed with melanoma before age 30, 97% had used tanning beds in their lifetime (Lazovich et al., 2016).

**Communication About Melanoma Within Families**

Because families as a whole may share increased risk, based on health and social history, all members of a family should be made aware of their melanoma risk. Having even one family member with melanoma significantly increases a person’s lifetime risk (Wei et al., 2019). Therefore, all melanoma patients are encouraged to tell unaffected family members about melanoma risk and risk reduction practices (Rodríguez et al., 2017).

Generally, melanoma survivors express desire to provide information to their families when they are aware of the increased risk to relatives. In one study, all participants perceived that it was important for them to talk to family members about melanoma risk-modifying behaviors, particularly in regard to UV radiation exposure (Loescher et al., 2009). Studies show that 74 – 94% of patients with melanoma discussed genomic risk information with at least some of their family members (Bowen, et al., 2017; Hay et al., 2009; Smit et al., 2017).

A family’s communication style prior to diagnosis and beliefs about disease play important roles in effective communication about melanoma. If a family has an open
communication style, information regarding melanoma is more frequently communicated to first degree relatives. Additionally, more communication takes place if family members have strong beliefs about an increased melanoma risk or belief in a genetic element of their diagnosis (Harris et al., 2010; Hay et al., 2009).

Unfortunately, even though melanoma survivors often express desire to share information with family members, many times crucial information is not shared either because of a lack of knowledge or lack of communication.

Melanoma survivors often lack knowledge about the increased risk for all first-degree relatives. For example, a population-based study of 170 melanoma survivors in the United States found that half were not aware their family was at an increased risk (Zapolska et al., 2016). Thus, although survivors may tell family members about their own diagnosis, they may not convey the message that family members are at risk and should take prevention and screening precautions.

Lack of communication can occur in either close or emotionally distant families because of family dynamics. Some family members may be estranged or geographically isolated. In a study of melanoma survivors and first-degree relatives, lack of general family contact was cited by 67-74% of survivors as the main reason for not communicating with family members about familial melanoma risk (Bowen, et al., 2017). Alternatively, in emotionally close families a desire to avoid upsetting family members is a reason melanoma affected individuals may not share information about cancer diagnosis or downplay the risk. Fear of burdening others and a relative’s inability to cope, are some of the reasons information is not shared (Healey et al., 2017; Loescher et al., 2009). People diagnosed with cancer often express desire to manage anxiety for others in their family (Hay et al., 2009). Similarly, unaffected family members may not ask for information about melanoma and risk for other family members in fear of upsetting
the person diagnosed with cancer. Instead, family members may search the internet to find more information; however, information found on the internet may not accurately describe increased risk of melanoma within families (Hay et al., 2009).

Accurate risk perception is important if unaffected family members are to follow through with prevention recommendations and share risk concerns with their own PCPs. However, communication does not always occur between family members, and melanoma survivors generally are provided with little guidance for having these conversations (Bowen et al., 2017). Lack of effective communication and awareness of melanoma risk leads to decreased screening, lack of prevention behaviors, and delayed diagnosis of melanoma.

**Clinical Implications for PCPs**

Considering a lack of knowledge about increased risk for developing melanoma in people with a family history of melanoma, a PCPs role is vital. PCPs should not assume patients with a family history of melanoma understand they are at increased risk. Additionally, even if an unaffected family member has been told about melanoma in the family, he/she may not disclose the family history to the PCP. Despite the fact that over half of melanoma survivors encourage family members to inform their healthcare provider about family history of melanoma (Rodríguez et al., 2017), studies have shown that unaffected family members with a family history of melanoma communicate with a healthcare provider about their family history only 12% to 41% of the time depending on their risk perception (Bowen, et al., 2017; Smit et al., 2017). Thus, PCPs need to actively engage patients on the topics of family history and melanoma risk.
A PCP’s role in caring for a patient with a family history of melanoma includes two main areas; (1) assess risk and teach and (2) encourage prevention and screening measures.

**Assessing Risk**

Assessing a patient’s risk involves three elements, and sometimes a fourth: (1) reviewing family history of melanoma, (2) evaluating phenotype, (3) assessing environmental and behavioral risk factors, and sometimes, (4) ordering or referring for genetic testing if warranted.

**Review Family History of Melanoma**

PCPs need to ask about family history of melanoma and other cancers. The age at which the family member was diagnosed with melanoma is important. A melanoma diagnosis before the age of 30 increases the risk for melanoma in family members (Wu et al., 2018).

**Evaluate Phenotype**

One helpful tool for assessing risk by phenotype is the Fitzpatrick skin type classification. The Fitzpatrick skin type is based on the amount of melanin pigment in the skin, which is linked to the color of the skin and the likelihood for sunburns (see Table 1). Patients with skin type I and II face the highest risk of developing melanoma with types V and VI having the lowest risk (Skin Cancer Foundation, 2018).
Table 1

Scheme used in the Fitzpatrick’s Skin Type Classification

<table>
<thead>
<tr>
<th>Skin Type</th>
<th>Skin Color</th>
<th>Hair and Eyes</th>
<th>Burning and Tanning during first exposure in summer</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Pale</td>
<td>Red hair, blue eyes, freckles</td>
<td>Always burns, never tans</td>
</tr>
<tr>
<td>II</td>
<td>Pale</td>
<td>Blonde hair, blue eyes</td>
<td>Usually burns, tans less than average</td>
</tr>
<tr>
<td>III</td>
<td>Pale or lightly dark</td>
<td>Blonde or Brown hair, blue or brown eyes</td>
<td>Sometimes burns, tans about average</td>
</tr>
<tr>
<td>IV</td>
<td>Brown</td>
<td>Brown hair and eyes</td>
<td>Rarely burn, tans more than average</td>
</tr>
<tr>
<td>V</td>
<td>Dark Brown</td>
<td>Brown hair and eyes</td>
<td>Rarely burn, tan profusely</td>
</tr>
<tr>
<td>VI</td>
<td>Black</td>
<td>Brown hair and eyes</td>
<td>Never burn, always tan</td>
</tr>
</tbody>
</table>

(Eilers et al., 2013; Fitzpatrick, 1988; Matts et al., 2007; Pathak et al., 1976)

Assess Environmental and Behavioral Risk Factors

In addition to phenotype, it is important to assess patients’ environmental and behavioral risk factors. This includes considering number of sunburns, especially the number of blistering sunburns, exposure to tanning beds and whether a patient grew up in a sunny climate or higher elevations. PCPs should evaluate this information to assess patients’ risk for melanoma.

Order or Refer for Genetic Testing

Patients with a family or personal history of melanoma may benefit from genetic testing to further clarify their risk. Genetic testing is not recommended for all families with a history of melanoma. According to the American Academy of Dermatology Association (Leachman et al., 2009; Swetter et al., 2019), genetic testing may be recommended for individuals with the following risks:
- Three or more melanomas that have grown deep into the skin (or spread), especially if one melanoma was diagnosed before their 45th birthday
- Three or more blood relatives on one side of the family who have had melanoma or cancer of the pancreas
- Two or more unusual-looking moles called Spitz nevi, (a rare non-cancerous mole that usually affects younger adults and children and often looks like melanoma. Also known as MBAIT)
- One or more Spitz nevi and a close blood relative has (or had) mesothelioma (a type of cancer), meningioma (a type of brain tumor), or melanoma of the eye

Patients who meet requirements for genetic testing may be referred to a genetic counselor or a dermatologist that does genetic testing. Alternatively, PCPs may order genetic testing if they have developed expertise in pre- and post-test genetic counseling and have skill in interpreting genetic test results.

Because melanoma can be a component of many hereditary cancer syndromes, it is important to consider broader causes of melanoma if the family history includes multiple types of cancer. It is beyond the scope of this paper to discuss all cancer predisposing syndromes that include melanoma. Leachman et al. (2017) provides an in-depth discussion of these syndromes and a decision-making instrument to help providers determine when genetic panel testing would be appropriate. Panel testing is available commercially and costs approximately the same as targeted gene testing. It is important for healthcare providers to work with diagnostic laboratories before tests are drawn to ensure proper documentation for billing. Most insurance companies will cover the cost of testing if patients meet testing criteria. Laboratories will work with patients and can often run a panel test for under $300 for self-pay patients.
Teach and Encourage Prevention and Screening Measures

PCPs should counsel all patients with a family history of melanoma about lifestyle prevention measures, self-skin exams, PCP skin exams, and regular dermatology screening (Rossi et al., 2019).

Lifestyle-related Prevention Measures

Lifestyle-related prevention measures to reduce melanoma risk include appropriate use of sunscreen, and minimization of UV radiation exposure.

Sunscreen use can reduce the risk of sunburns, if it is used correctly and there is no increased time in the sun due to a false sense of security by applying the sunscreen (Olsen et al., 2018; Watts et al., 2018). Sunscreen should be at least 15 SPF, cover both UVA and UVB rays, and be applied to the skin every two hours (Gallagher, 2005; Strauss & Michele, 2020).

Minimization of UV exposure is accomplished by avoiding sun exposure during peak hours of 10 am to 4 pm and seeking shade when outdoors (Gallagher, 2005; Linos et al., 2011; Saraiya et al., 2004). Shade does not provide complete UV protection; however, it does reduce the total UV exposure (Downs et al., 2014; Parisi & Turnbull, 2014). Additionally, UV exposure can be reduced through protective clothing such as UV-protective fabrics, wide-brimmed hats, full-length clothing, and UV-protective sunglasses. UV protective fabrics are made with ultraviolet protection factor (UPF). A UPF rating of 30 means that the fabric will only allow 1/30th of UV radiation to pass through the material. Clothing with a UPF rating over 25 is considered very good, and over 40 is considered excellent (Aguilera et al., 2014). Finally, tanning beds should be avoided completely to avoid any unnecessary UV exposure.
Self-skin Exams

Self-skin exams (SSEs) should be recommended and taught to patients who have a first-degree relative with melanoma. There is evidence that the practice of SSEs is beneficial for high-risk individuals (Coroiu et al., 2020). In fact, in one study patients successfully detected 33% of melanomas and patient spouses found 16% of melanomas during SSEs (De Giorgi, 2012). Another study demonstrated that SSEs may reduce melanoma mortality by 63% (Berwick et al., 1996). Patients that receive provider advise on performing SSEs and use a mirror or have a partner available, are more likely to do SSEs and more confident in their ability to examine their skin (Chiu et al., 2006; Robinson et al., 2007). Providing written information on how to complete a SSE, improved exam rates from 30% to 80% in women (Glanz et al., 2015; Robinson et al., 2019).

Screening PCP Skin Exams

Often PCPs are the first point of contact for skin concerns. PCPs should be comfortable with providing full skin exams to help identify suspicious looking moles to excise for biopsy. Currently, PCPs perform 1.4%-13% of all initial melanoma biopsies (Curiel-Lewandrowski et al., 2012). However, only 37% of PCPs are conducting annual skin exams on their patients (Altman et al., 2000). This low percentage may be due to short amount of time allowed during an annual visit, PCPs not feeling comfortable doing a skin exam and diagnosing skin cancer, or PCPs trying to reduce discomfort for the patient (Geller et al., 2004; Oliveria et al., 2011).

Refer for Regular Dermatology Screening

Referral to dermatology is important for patients that meet familial melanoma criteria, genetic testing criteria, or have been diagnosed with melanoma. Although it can take time and effort to become established with a dermatologist (Hawkins, 2017), annual dermatologic
evaluation is important for high-risk individuals. Most melanoma is identified through annual skin exams by dermatologists. In fact, in a study of 802 melanoma patients, their lesions were first identified by dermatologists 36% of the time, by patients themselves 33% of the time, by a spouse 16% of the time and 15% by a PCP (De Giorgi, 2012). Because dermatologists identify most melanomas, it is essential for patients with a family history of melanoma to see both their PCP and a dermatologist on a regular basis, in addition to conducting SSEs. Patients visiting both a dermatologist and PCP before diagnosis of melanoma, have greater odds of diagnosis at an earlier stage and lower mortality. The lower mortality was due to an earlier staging at diagnosis and an earlier staging has higher survival rates (see Table 2) (Roetzheim et al., 2013).

**Table 2:**

*Five-Year Survival Rate by Melanoma Stage*

<table>
<thead>
<tr>
<th></th>
<th>Localized Melanoma</th>
<th>Regional Melanoma</th>
<th>Metastatic Melanoma</th>
</tr>
</thead>
<tbody>
<tr>
<td>General Description</td>
<td>No sign cancer has spread beyond the skin where it started</td>
<td>Cancer has spread beyond the skin to nearby structures of lymph nodes</td>
<td>Cancer has spread to distant parts of the body, such as the lungs, liver or skin on other parts of the body</td>
</tr>
<tr>
<td>Stage at Diagnosis</td>
<td>Stage 0, I and II</td>
<td>Stage III</td>
<td>Stage IV</td>
</tr>
<tr>
<td>Five Year Survival Rate</td>
<td>99%</td>
<td>65%</td>
<td>25%</td>
</tr>
</tbody>
</table>

(Godman, 2020; Loescher et al., 2009)

Some dermatologists offer mole mapping services to monitor skin changes. Patients with a significant number of moles may benefit from mole mapping, which includes receiving photographs of the patient’s body showing all moles to use as a reference for SSEs. Studies show patients that use mole mapping photos for monthly SSEs were more likely to identify new lesions (Chiu et al., 2006).
Conclusion

Melanoma is a deadly form of skin cancer. People with a family history of melanoma are at significantly increased risk of developing melanoma themselves regardless of whether or not the family meets the strict criteria for familial melanoma. In families with a history of melanoma, unaffected family members may not be aware of their increase in risk because of lack of family communication. Thus, it is important for PCPs to be aware of the family history of melanoma and to educate patients with a family history of melanoma about increased risk. PCP care of patients at risk for melanoma requires risk assessment, patient teaching about prevention measures, and inter-disciplinary collaboration to perform skin exams for early diagnosis.
References


PennSCAPE randomized trial. *Cancer Epidemiology, Biomarkers & Prevention*, 24(2), 415-421. https://doi.org/10.1158/1055-9965.EPI-14-0926


https://doi.org/10.1002/pon.4315

https://doi.org/10.3122/jabfm.2013.06.130042

https://doi.org/10.5826/dpc.0901a03

https://doi.org/10.1016/j.amepre.2004.08.009

Skin Cancer Foundation (2018). Are you at risk for skin cancer?  
https://www.skincancer.org/blog/are-you-at-risk-for-skin-cancer/

https://doi.org/10.1111/bjd.15744


the American Academy of Dermatology, 81(2), 489-499.

https://doi.org/10.1016/j.jaad.2019.04.044


