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General Information for Individuals with a *MC1R* Risk Allele or Mutation

Cancer Risks and General Management Considerations

The cancer risks for individuals who have an *MC1R* mutation are still being understood. However, it has been suggested that individuals with one mutation in the *MC1R* gene may be at an increased chance to develop skin cancer, particularly melanoma. The exact lifetime risk for individuals with a mutation in the *MC1R* gene have not been established, but may indicated a slightly increased risk for melanoma (2.6%-11.8%)[1-3] and may confer a slightly increased risk for basal cell carcinoma. Associations with other cancers have not been reported for individuals with a *MC1R* risk allele or mutation at this time.

No consensus management recommendations or guidelines have been published for individuals with a mutation in the *MC1R* gene at this time. Individuals may consider a formal dermatological evaluation based on the increased chance to develop skin cancer.

Implications for Family Members/Reproductive Considerations

- First-degree relatives (i.e., parents, siblings, and children) have a 50% chance to have the familial *M1CR* mutation. Second-degree relatives (i.e., nieces/nephews, aunts/uncles, and grandparents) have a 25% chance to have the familial mutation.
- Individuals with two *MC1R* mutations (one inherited from each parent) have a chance for red hair and paler skin.
- Family members may pursue genetic counseling to clarify their risks. Family members can visit www.FindAGeneticCounselor.com to find genetic services near them.

References

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2. Landi Mt, Kanetsky PA, Tsang S, et al. 2005. MC1R, ASIP, and DNA Repair in Sporadic and Familial Melanoma in a Mediterranean Population. J Natl Cancer Inst. 97(13):998-1007.
3. Williams PF, Olsen CM, Hayward NK, Whiteman DC. 2011. Melanocortin 1 Receptor and Risk of Cutaneous Melanoma: a Meta-analysis and Estimate of Population Burden. Int J Cancer. 129(7);1730-40.
4. King RA, Willaert RK, Schmidt RM, et al. 2003. MCR1 Mutations Modify the Classic Phenotype of Oculocutaneous Albinism Type 2 (OCA2). Am J Hum Genet. 73(3): 638-645.