MITF p.E318K prevalence similar, regardless of CDKN2A

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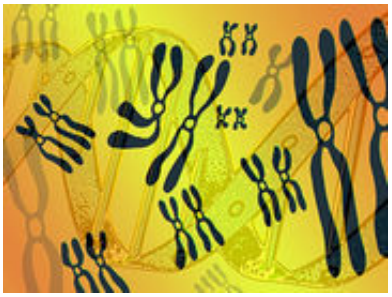

(HealthDay)—The prevalence of *MITF* p.E318K is similar in patients with melanoma, irrespective of the presence of *CDKN2A* mutations, according to a study published online Dec. 10 in *JAMA Dermatology*.

Miriam Potrony, from the Universitat de Barcelona in Spain, and colleagues examined the prevalence of *MITF* p.E318K in Spanish patients with melanoma. Genotyping was performed for 531 patients with melanoma: 271 with multiple primary melanoma (MPM) without mutations affecting p16INK4A; 191 probands from melanoma-prone families without mutations affecting p16INK4A; and 69 probands from families carrying *CDKN2A* mutations affecting p16INK4A. Controls were 499 age- and sex-matched cancer-free individuals from the Spanish National Bank of DNA.

The researchers found that the prevalence of *MITF* p.E318K was 1.9 percent in all melanoma patients with wild-type p16INK4A; 2.6 percent in patients with MPM; and 2.9 percent in probands of families with p16INK4A mutations. The *MITF* p.E318K variant correlated with increased melanoma risk (odds ratio, 3.3), especially in MPM and high nevi count (odds ratios, 4.5 and 8.4, respectively). Among two *MITF* p.E318K carriers, two fast-growing melanomas were detected during dermatologic digital follow-up.

"Testing for *MITF* p.E318K should not exclude patients with known mutations in p16INK4A," the authors write.

More information: Abstract
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