

## Supplementary Online Content

Potrony M, Puig-Butille JA, Aguilera P, et al. Prevalence of *MITF* p.E318K in patients with melanoma independent of the presence of *CDKN2A* causative mutations. *JAMA Dermatol*. Published online December 9, 2015.  
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**eTable 1.** Primary Melanoma Characteristics From Patients With *MITF* p.E318K

**eTable 2.** Summary of the *MITF* p.E318K Reports in Melanoma Patients

This supplementary material has been provided by the authors to give readers additional information about their work.

**eTable 1. Primary Melanoma Characteristics From Patients With *MITF* p.E318K**

Patient ID	Famil y ID	MM	Year	Location	Clark level	Breslow thicknes s	Subtype
M0109-01	M010 9	NA	NA	NA	NA	NA	NA
M0881-01	M088 1	1st	1995	Trunk	IV	6.00	SSM
		2nd	2001	Trunk	III	0.50	SSM
		3rd	2004	Trunk	III	0.80	SSM
		4th	2013	Upper extremity	IV	1.30	NM
M1340-01	M134 0	1st	2003	Head	I	in situ	LMM
		2nd	2006	Trunk	III	1.60	SSM
M1545-01	M154 5	1st	2005	Lower extremity	II	0.50	SSM
		2nd	2005	Upper extremity	II	NA	SSM
M1569-01	M156 9	1st	2005	Trunk	I	in situ	SSM
		2nd	2005	Lower extremity	II	0.90	SSM
		3rd	2012	Lower extremity	I	in situ	SSM
		4th	2013	Trunk	NA	NA	SSM
M3824-01	M382 4	1st	2009	Lower extremity	IV	2.20	SSM
		2nd	2012	Foot	I	in situ	ALM
M3879-01	M387 9	1st	2003	Lower extremity	I	in situ	SSM
		2nd	2003	Trunk	II	0.60	SSM
M4182-01	M418 2	1st	1997	Trunk	NA	0.90	SSM
		2nd	2004	Lower extremity	NA	0.35	SSM
M4619-01	M461 9	1st	1995	Trunk	NA	NA	NA
M4713-01	M471 3	1st	2012	Scalp	IV	1.00	SSM
M4999-01	M499 9	1st	2014	Lower extremity	II	0.60	SSM

MM: melanoma; NM: nodular melanoma; SSM: Superficial Spread melanoma, LMM: Lentigo malignant melanoma; ALM: Acral Lentiginous melanoma. Breslow thickness is expressed in millimeters.

**eTable 2. Summary of the *MITF* p.E318K Reports in Melanoma Patients**

Study	Patients and origin	N cases	Melanoma Risk	Age of onset	High nevi count	non-blue eyes	Fair skin color	Dark hair color	Renal cancer	pancreatic cancer	Nodular melanoma
			OR (95% CI)		OR (95% CI)	OR (95% CI)	OR (95% CI)	OR (95% CI)	OR (95% CI)	OR (95% CI)	OR (95% CI)
Yokoyama et al. 2011	Australia + UK										
	All patients	3988	<b>2.19 (1.41–3.45)</b>		<b>2.5 (1.42–4.55)</b>	<b>2.0 (1.11–3.81)</b>	N.S.	N.S.			
	Australia										
	All patients	2059	<b>2.33 (1.21–4.70)</b>	N.S.							
	familial MM	575	<b>2.95 (1.23–6.92)</b>								
	MPM	270	<b>4.22 (1.52–10.91)</b>								
	familial MPM	105	<b>8.37 (2.58–23.80)</b>								
	UK										
	All patients	1929	<b>2.09 (1.14–3.94)</b>	N.S.							
	familial MM	110	N.S.								
MPM	89	N.S.									
Bertolotto et al. 2011	France, Italy and Spain										
	All patients	603	<b>4.78 (2.05–11.75)</b>						<b>5.2 (1.37–16.87)<sup>†</sup></b>		
	MPM	288	<b>7.79 (3.12–20.04)</b>								
Ghiorzo et al.	Italy										

2013	All patients	679	<b>2.85 (1.31-6.18)</b>						<b>8.0 (1.62-39.46)</b>	<b>30.9 (6.85-138.9)</b>	<b>4.48 (1.39-14.43)</b>
	Familial single MM	129	N.S.								
	Sporadic MPM	52	<b>6.40 (1.43-28.58)</b>								
Sturm et al. 2013	Australia	6	-		increased nevus count		All patients				no association
Berwick et al. 2014	Australia, Italy, Canada, USA										
	MPM	1194	<b>1.7 (1.1-2.6)*</b>		N.S.	<b>2.1 (1.2-3.8)</b>	<b>2.2 (1.2-3.8)</b>	<b>3.8 (1.5-9.6)</b>			
Present study	Spain										
	All patients	511	<b>2.9 (1.24-6.69)</b>	N.S.	<b>9.0 (2.30-35.45)</b>	N.S.	N.S.	N.S.	1/9 carriers	N.S.	N.S.
	MPM	271	<b>4.2 (1.67-10.44)</b>		<b>12.4 (2.58-59.7)</b>	N.S.	N.S.	N.S.			

MM: melanoma, MPM: multiple primary melanoma, N.S.: Non significant, OR: Odds Ratio, 95% CI: 95% Confidence Interval

\*Sporadic single melanoma patients were used as controls

†Renal cancer risk was evaluated in an independent cohort of 829 patients with renal cancer.