

Supplementary Table 1. *Distribution of pigmentation phenotypes in CDK4-positive and CDK4-negative family members*

Family members	Skin color						Hair color					
	N	Very fair	Fair	Olive	Brown	P-value*	N	Red	Blond	Brown	Black	P-value*
Affected and <i>CDK4</i> -positive	46	8 (17.4)	27 (58.7)	7 (15.2)	4 (8.7)	NS	49	8 (16.3)	20 (40.8)	17 (34.7)	4 (8.2)	NS
Unaffected and <i>CDK4</i> -positive	19	0 (0.0)	14 (73.7)	3 (15.8)	2 (10.5)	NS	19	0 (0.0)	8 (42.1)	10 (52.6)	1 (5.3)	NS
<i>CDK4</i> -negative	53	4 (7.6)	33 (62.3)	10 (18.9)	6 (11.3)	-	62	6 (9.7)	26 (41.9)	30 (48.4)	0 (0.0)	-

* *CDK4*-negative family members were compared with affected and with unaffected *CDK4*-positive family members, respectively.

NS = non-significant *P*-value

Supplementary Table 2. *Phenotypic characteristics of family members testing positive for the CDK4 mutations R24C or R24H*

Variable	R24C	R24H	P-value
<i>Age at first diagnosis (N=62)</i>			
N	21	41	
Median (years)	33.0	39.0	NS
Mean (years)	36.2	40.0	
	N (%)	N (%)	
<i>Number of primary melanomas (N=62)</i>			
SPM	10 (47.6)	21 (51.2)	NS
MPM	11 (52.4)	20 (48.8)	
<i>Atypical nevi (N=70)</i>			
Present	19 (82.6)	31 (66.0)	NS
Not present	4 (17.4)	16 (34.0)	

NS = non-significant P-value; SPM = single primary melanoma; MPM = multiple primary melanoma

Supplementary Table 3. *Presence of non-melanoma cancers in melanoma cases and CDK4 mutation carriers*

Cancer type*	N		Age at diagnosis†
Non-melanoma skin cancer	12		
	8	Squamous cell carcinomas	73.5
	4	Basal cell carcinomas	68
Female-related cancer	12		
	9	Breast cancers	50
	1	Phyllodes tumor	45
	1	Borderline ovarian tumor	41
	1	Uterine cervix cancer	77
Gastro-intestinal cancer	5		
	2	Pancreatic cancers	60
	1	Stomach cancer	-
	1	Colon cancer	70
	1	Rectal cancer	43
Other cancers	4		
	2	Lung cancers	69
	1	Prostate cancer	70
	1	Malignant lymphoma	73

* The table is based on the 105 subjects with melanoma and/or verified *CDK4* mutation (including obligate carriers) where information on other cancers had been specified on the data collection forms. Altogether, 33 non-melanoma cancers were recorded in 25 subjects.

† Median age when more than one case.