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Supplemental Data

IRF4 Variants Have Age-Specific Effects on Nevus Count and Predispose to Melanoma

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Figure S1. Quantile-Quantile Plot for the Genome-wide Association Scan for Total Nevus Count in Queensland Adolescent Twins

The measured genotype analysis was performed as a mixed-model score test using the MERLIN package ("merlin –fastassoc"). The grey colored region represents an approximate 95% confidence envelope (that does not adjust for inter-SNP linkage disequilibrium). Note that lambda is 1.004, despite the large number of associated SNPs.

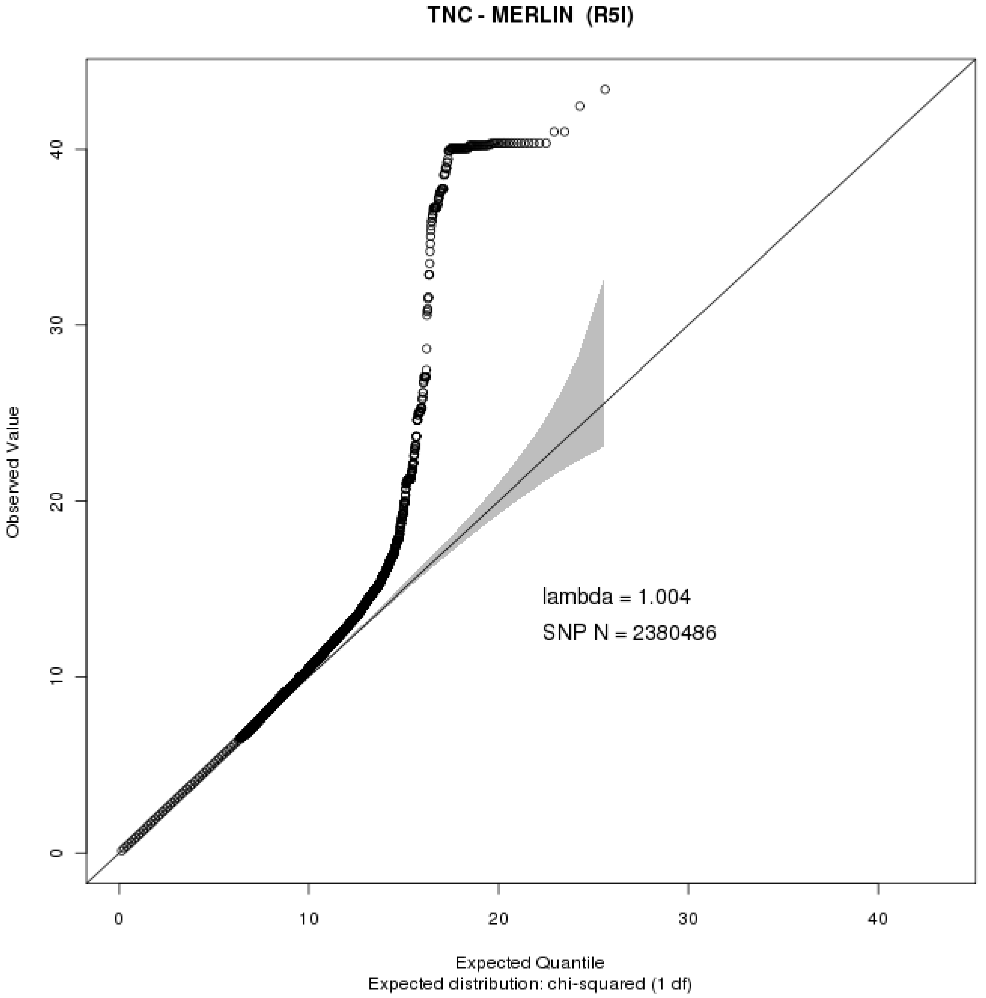

Figure S2. Nevus Counts in Queensland Adolescent Twins, UK Adolescent (Leeds) and Adult (Twins UK) Twins, UK Unrelated Controls and Melanoma Cases (Leeds) by Age Band and rs12203592 Genotype

A model including a restricted cubic spline fit for age concluded that the three UK non-melanoma-case samples did not differ significantly from one another (P=0.94), so we present data for melanoma cases in the upper row of plots, the Queensland twin data in the first two plots of the lower row, and the combined data from the UK non-cases in the remaining plots. The notches in the bars represent approximate 95% confidence intervals on the medians, and in some cases exceed the interquartile range.

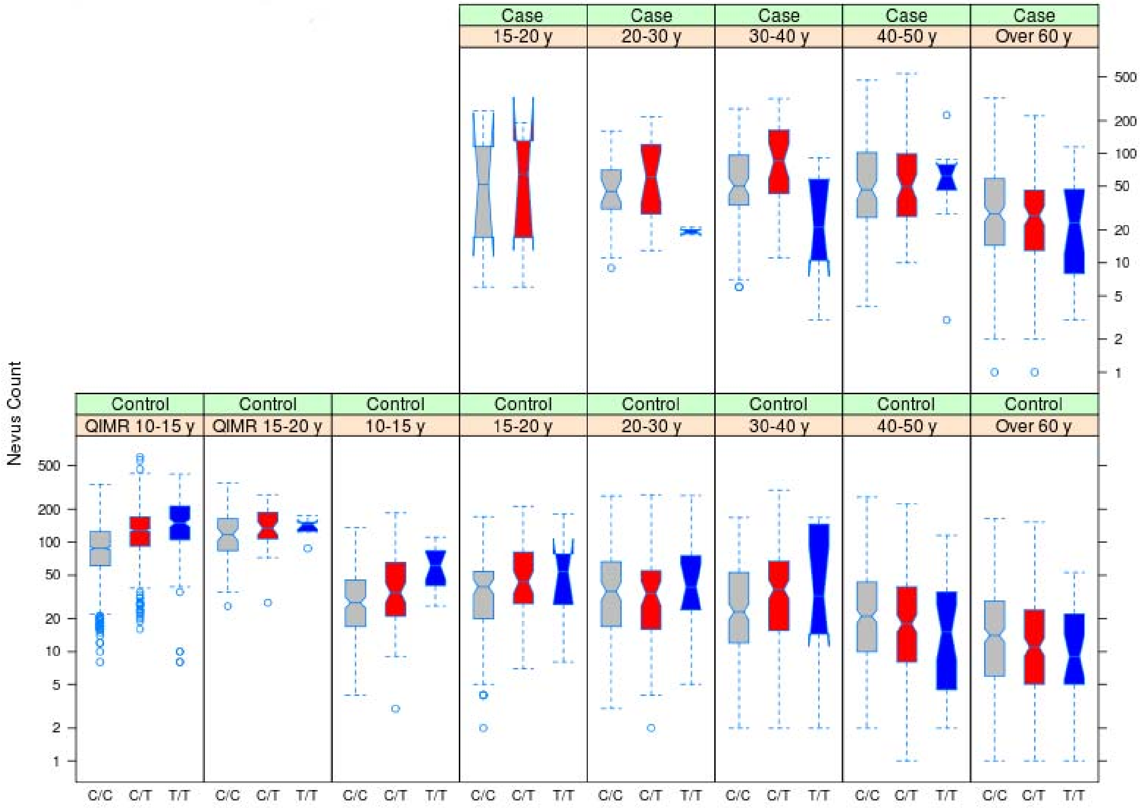

Figure S3. Analysis of Melanoma on the Trunk for Case-Control Data from Sweden, Leeds and Brisbane for rs12203592

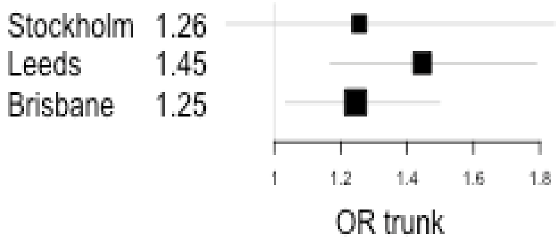

Table S1. Risk of Melanoma on Different Body Sites versus rs12203592 Genotype in Leeds, QIMR, and Stockholm Case-Control Comparisons

Study	Tumour Site*	N	OR (C allele)
Leeds	Head	151	1.05 (0.77- 1.43)
	Limbs	543	1.20 (0.97-1.48)
	Trunk	404	1.53 (1.21-1.94)
	Other	94	1.04 (0.72-1.51)
QIMR	Head	169	1.12 (0.85-1.46)
	Limbs	394	1.05 (0.87-1.26)
	Trunk	486	1.22 (1.03-1.48)
	Other	356	1.11 (0.91-1.35)
Stockholm	Head	88	0.63 (0.38-1.05)
	Limbs	330	0.95 (0.66-1.36)
	Trunk	349	1.26 (0.86-1.85)
	Other	8	0.57 (0.13-2.45)
Overall**	Head	408	1.00 (0.83-1.21)
	Limbs	1267	1.09 (0.96-1.23)
	Trunk	1239	1.33 (1.16-1.52)
	Other	458	1.08 (0.91-1.28)

* Head (site codes C440-C444), trunk (C445), limbs (C446-C447), Other (C449, C809, unspecified).

** Multinomial regression stratifying on ethnicity/study.