Supplementary Online Content

Atzmony L, Khan HM, Lim YH, et al. Association of Second-Hit, Postzygotic *PMVK* and *MVD* Mutations With Linear Porokeratosis. *JAMA Dermatol.* Published online April 3, 2019. doi: 10.1001/jamadermatol.2019.0016

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

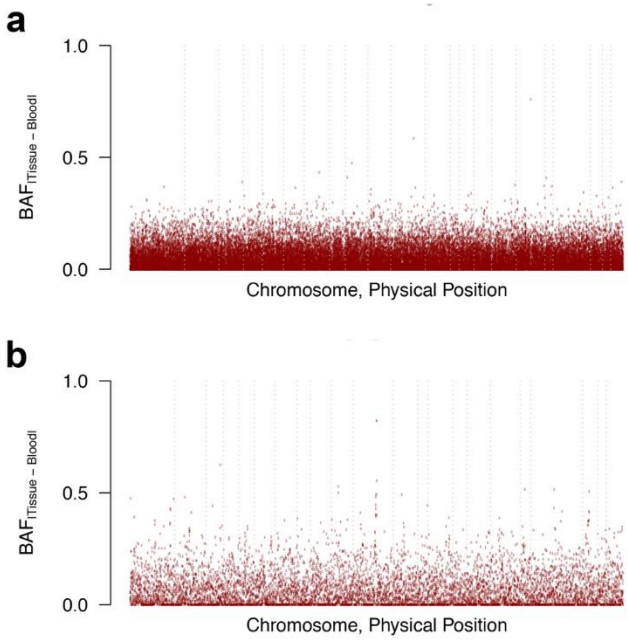
eTable 1. Primers list.

Name	Sequence (5'-3')	Use
PMVK exon 1-F	ATTCCGCGCGGTGTCC	PCR and Sanger sequencing
PMVK exon1-R	GTGACACCCAAAAGCACTCCTC	PCR and Sanger sequencing
PMVK exon4-F	GTGAGTCTAGCTGCTGACATGC	PCR and Sanger sequencing
PMVK exon4-R	ATCAGGAGGCCTCAGATTCTCC	PCR and Sanger sequencing
MVD intron1-F	GCGGACCTACGTCAGCAACC	PCR and Sanger sequencing
MVD intron1-R	GAGCTTGTCACGCGAAGGAG	PCR and Sanger sequencing
MVD exon7-F	CGCCAAGACCACGTGCAG	PCR
MVD exon7-R	ACAGCAGGGTGGAGCTTTCAGACAC	PCR
PMVK-F	TGTGGTTTGTCTACCGGGC	qPCR
PMVK-R	AGGAGGCCTCAGATTCTCCTT	qPCR
ACTB-F	CTCATGGCCTTGTCACACGA	qPCR
ACTB-R	TGAGCTCTTTTTCTGGTGTTTGTC	qPCR
MVDc exon1-5-F	TTGTACAGCGCCGGTCAAC	Selective cDNA amplification and Sanger sequencing
MVDc exon1-5-R	GGTGTAGGCTAGGCAGGCATAG	Selective cDNA amplification and Sanger sequencing
MVDc-F	AACATCGCGGTCATCAAGTA	qRT-PCR
MVDc-R	AACTGGTCCTGGTGCAGAGT	qRT-PCR
ACTBc-F	GATCATTGCTCCTCCTGAGC	qRT-PCR
ACTBc-R	ACATCTGCTGGAAGGTGGAC	qRT-PCR

eTable 2. Exome sequencing statistics.

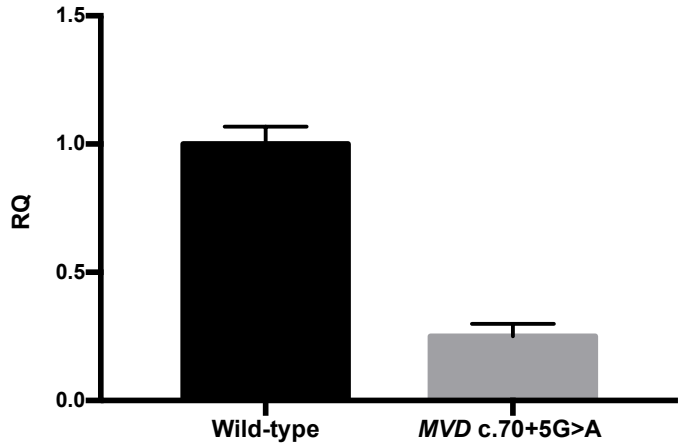
Sample	Mean coverage	Bases covered >15x	Mean read length
Participant 1-blood	45.5	94.3%	99 bases
Participant 1-tissue	133.6	98.8%	99 bases
Participant 2-blood	38.6	89.8%	99 bases
Participant 2-tissue	139.9	98.6%	99 bases
Participant 3-blood	37.7	90.7%	99 bases
Participant 3-tissue	82.7	92.7%	99 bases
Participant 3's mother- blood	29.39	81.9%	99 bases

eFigure 1. There is no evidence for somatic LOH in participants 2 and 3.



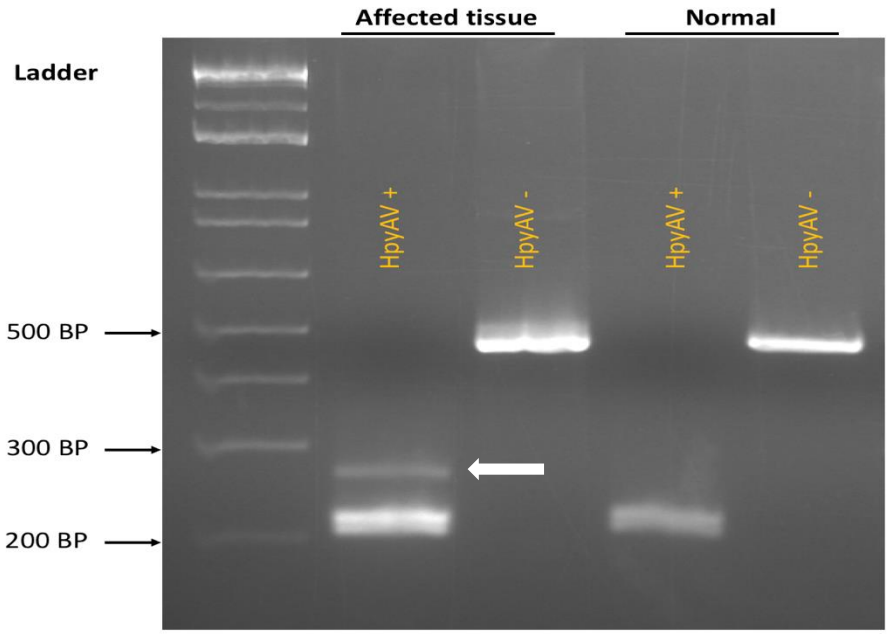
B- allele frequency differences between affected tissue and blood are plotted across the genome for participant 2 (a) and participant 3 (b) by chromosome and physical position**.** Dashed vertical lines separate individual chromosomes.

eFigure 2: Expression of wild type *MVD* transcript is significantly reduced in human keratinocytes with heterozygous *MVD* c.70+5G>A mutation suggesting that *MVD* c.70+5G>A transcript undergos degradation.



Comparative qRT-PCR was employed to assess the expression of wild-type *MVD* transcript in heterozygous *MVD* c.70+5G>A mutated human keratinocytes. Wild-type human keratinocytes were used as a reference sample. The fold change in the expression level of wild-type MVD transcript from mutated keratinocytes is caluculated using relative quantification (RQ). RQ= 0.25, $p<0.001$.

eFigure 3: Restriction fragment length polymorphism (RFLP) analysis confirms *MVD* c.811_815del p.F271fs in affected skin of participant 3.



Agarose gel electrophoresis of digested and undigested PCR amplicons of affected tissue (473 bp) and normal (478 bp) of participant 3. Digestion with HpyAV of mutated amplicon showing specific band of 254 bp length that is absent in normal tissue.