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Supplemental Data

Hereditary Leukonychia, or Porcelain Nails,

Resulting from Mutations in *PLCD1*

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a**Ala574Thr****Cys209Arg**

	3' ←	5' →		3' ←	5' →
Human	SSDTRWG	A	PYIRSLHGV	ELSDTQSHDC	E
Chimp	SSDTRWG	A	PYIRSLHGV	ELSDTQSHDC	E
Gorilla	SSDTRWG	A	PYIRSLHGV	ELSDTQSHDC	E
Rhesus	SSDTRWG	A	PYIRSLHGV	ELSDTQSRDC	E
Baboon	SSDTRWG	A	PYIRSLHGV	ELSDTQSRDC	E
Mouse	SSDTRWG	A	PYIRSLHGV	ELSDTQSHDC	E
Rat	SSDTRWG	A	PYIRSLCSV	ELSDTQSHDC	E
Guinea-Pig	SSDTRWG	A	PYIRSLHGV	ELSDTQSHDC	E
Cow	SSDTRWG	A	PYIRSLH—V	ELSDTQSHDC	E
Horse	SSDTRWG	A	PYIRSLHGV	ELSDTQSHDC	E
Dog	SSDTRWG	A	PYIRSLH—V	ELSDTQSHDC	E
Elephant	SSDTRWG	A	PYIRSLHGV	ELSDTQSRDC	E
Chicken	SSDTRWG	A	PYIRSLHRI	ELAETKSKDC	Y
X-Tropicalis	SSDTRFG	D	PYIRTLQRT	ELTGSQSKDC	H
Zebrafish	SSDTRSG	A	PYIRSLKDI	ELTGS—STDCK	

b

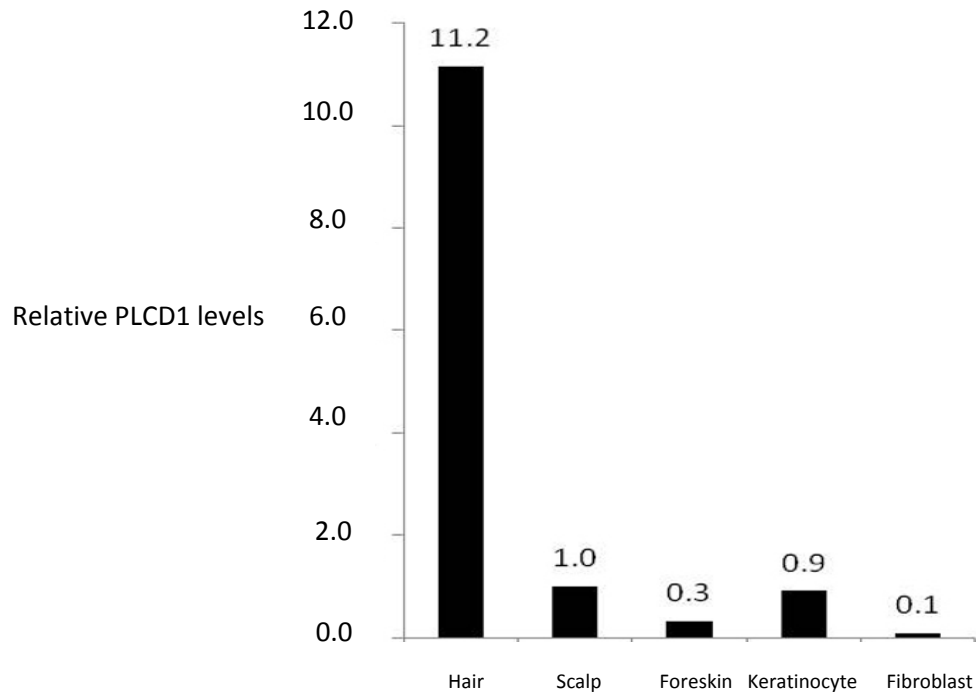


Figure S1: (a) The amino acid Ala574 is highly conserved across diverse vertebrate species and lies in PI-PLC Y-box . This region is highly conserved among the different isoforms of phospholipases and is important for the enzymatic catalytic activity. The amino acid Cys209 is highly conserved in many organisms and lies in the EF-hand domain which is present in many calcium-binding proteins.(b) PLCD1 levels as measured from several body sites, and in keratinocytes and fibroblasts.

Table S1. Primers used in the study. Primers to amplify additional microsatellite markers

Primer name	Forward primer	Reverse primer	Product size (bp)
CTDSPL	GTCGATATAAGAAACAACCCTAAC C	CAGCACAAAAGCTGCATATACT G	102
DLEC	GGGCAAAATGGTGAAATCTGATC	CATGCCACCACACTCAGCTA	112
Primers to amplify PLCD1 from genomic DNA			
PLCD1 -1	CTAGCTCCTCCCTCCCATTC	TAAAGGCTCCAAGGCAAATG	363
PLCD1 -2	ATGAGGCTGGTGAAAAGTGG	CCCATCTGTGGTTTTGATCC	379
PLCD1 -3	GGGACTGACCTCACAGCATT	TTATGGCAGCCCAAAGTACC	394
PLCD1 -4-5	GGGACCTTGGTAGGTTCCAG	TCTTCTGGCTGCAGTCTCC	598
PLCD1 -6-7	CTGGAAGACTGGCTCAGGAC	CACCCCTAGCATCCCACTC	599
PLCD1 -8-9	AGGTGAGGAGTGGGATGCTA	GCAGCCACAGAGAAGTGA	553
PLCD1 -10-11	CCTACCCTACCCCAACCACT	CTTGGGACTGAAACCCTCCT	494
PLCD1 -12-13	GCCCTGCTAACTCACCATGT	CCATATCAGCAGCATGGACA	567
PLCD1 -14	GTCCCCCTAGGTAGGTGCTG	GCAGGAGGGAACACAGAGAG	371
PLCD1 -15	GCCGAATGACCAGAACTCTC	GGAATGAAGGACAGCTCCAG	334