

Recessive mosaicism in *ABCA12* causes a unique phenotype of segmental congenital ichthyosiform erythroderma mimicking erythrokeratoderma variabilis

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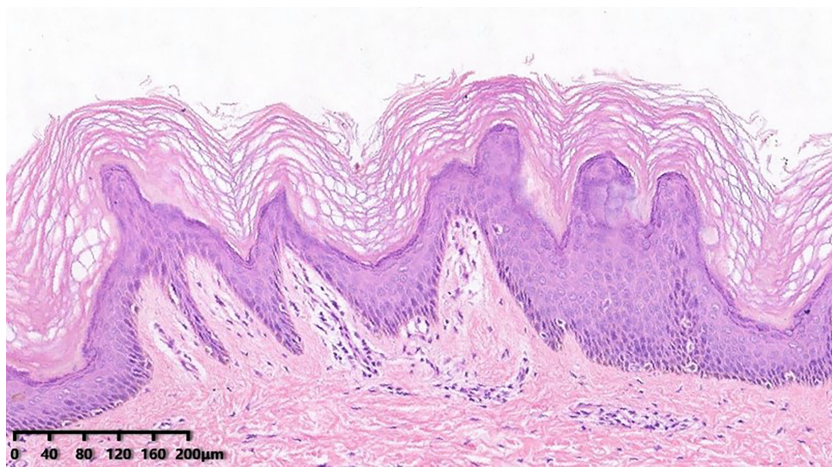
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SUPPLEMENTARY MATERIAL

Supplementary Figure 1. Comparison before and after treatment. Keratotic plaques thinned, and erythematous patches appeared after moisturizing treatment.



Supplementary Figure 2. Histological findings of the case. Hematoxylin-eosin staining of the skin (bar, 200µm).



Supplementary Figure 3. Sanger sequencing shows the presence of both mutations in the blood samples. **A)** *De novo* mosaic deletion mutation c.6861_6869del (p.Leu2288_Gly2290del); **B)** heterozygous missense mutation c.4724C>T (p.Thr1575Met) in the *ABCA12* gene.

A) ABCA12:NM_173076.3:exon46:c.6861_6869del;p.L2288_G2290del

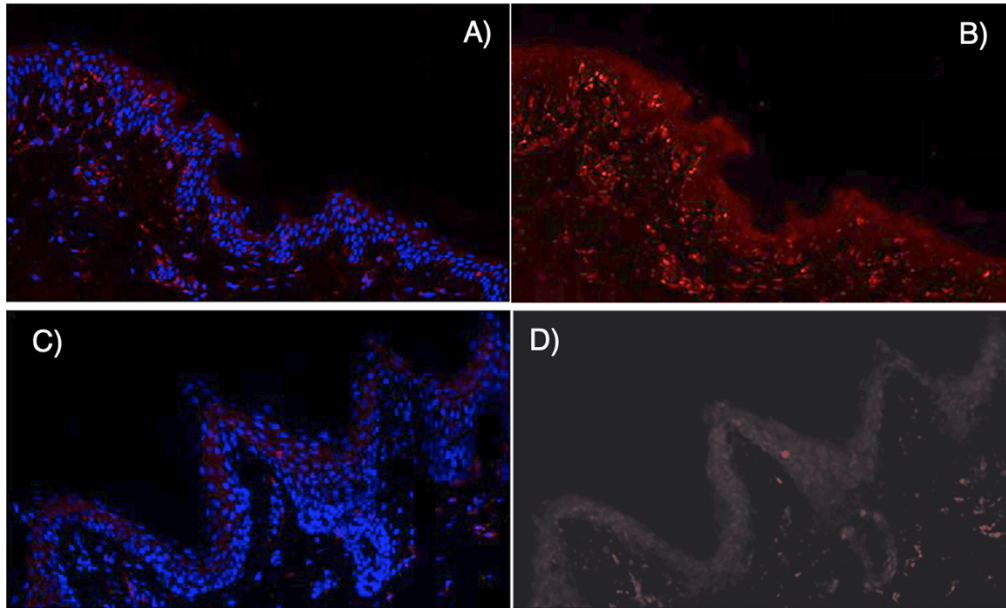


B)

ABCA12:NM_173076.3:exon31:c.4724C>T;p.T1575M



Supplementary Figure 4. Immunofluorescence staining showed that ABCA12 was prominently localized in the granular layer of the epidermis in control skin (**A**, **B**). In contrast, patient skin displayed only faint ABCA12 staining (**C**, **D**). Cryosections were incubated with a rabbit polyclonal anti-human ABCA12 antibody (1:200 dilution; GeneTex). Blue staining (DAPI) indicates nuclei. Scale bar=50 μ m.



Supplementary Table 1. List of genes included in the panel.

ATM	ANAPC1	SUMF1	WNT10A	ANTXR2	XRCC3	DST	GTPBP2	STAT1
PTEN	CREBBP	CHKB	SMARCAD1	INSR	POT1	CD151	MDM2	TRAF3IP2
BRAF	RBBP8	SGPL1	LEMD3	COX7B	CDH3	EXPH5	ZMPSTE2	IL17RA
SASH1	MTOR	PSAT1	FGFR2	HCCS	XPA	FKBP10	ATP7A	IL17RC
TMC6	TSC1	SHOC2	ATP2A2	FGFR3	XPC	PAX6	KRT86	SERPINB8
FANCC	IFNG	PHYH	TERC	PIGN	DDB2	LAMA3	SMAD4	FLG2
TWIST2	SOX10	PEX7	PKP1	C1R	ERCC5	ADA	BANF1	CYLD
TP63	PRKAR1A	FITM2	GJB6	C1S	IGF2	GLUL	ODC1	FLCN
NF1	TTC37	ALDH3A2	LAMC2	PPOX	SUZ12	NECTIN1	SMARCA2	FBLN5
AKT1	APC	STIM1	LOR	LBR	RASA1	IFT122	PNPLA6	LTBP4
SMARCAL	SKIV2L	GTF2E2	SERPINB7	RECQL4	PTCH2	WDR35	OFD1	ELN
KRAS	RET	PHGDH	CTSC	NSD1	HPS3	IFT43	POLR3A	EFEMP2
FANCD2	RIPK4	TGM5	PLEC	JAK3	HPS4	WDR19	PSMB8	ATP6V1E1
MAP2K1	IGF1R	NOD2	PEPD	GMPPA	HPS5	ATP6V1B2	PQBP1	ATP6V1A
ABCB6	EOGT	MBTPS2	AAGAB	TNFRSF1A	HPS6	KRT85	POC1A	ATP6AP1
TMC8	FLG	PIGL	AAAS	ALAS2	DTNBP1	HOXC13	WNT10B	SLC2A10
FANCA	KRT1	PSAP	TAT	POLH	BLOC1S3	IKBKG	TRPS1	GORAB
MAP2K2	ABHD5	LMNA	NOP10	TBX1	BLOC1S6	EVC2	TBX3	TALDO1
KLLN	EBP	KRT5	HPGD	AIRE	SOS1	ORAI1	EZH2	MGP
PIK3CA	NSDHL	SLC29A3	KRT17	GALNT3	RIT1	MSX1	DCAF17	ATP6AP2
SEC23B	KRT2	SLC27A4	DKC1	SAMD9	RRAS2	GRHL2	KRT25	GGCX
CIB1	TGM1	PEX5	SLCO2A1	ANTXR1	COL3A1	KDF1	ATP6V0A2	COL5A1

IL7	PNPLA1	PEX6	NHP2	MLPH	KRT4	PRKD1	ARHGAP3	NAA10
FANCB	ST14	FGFR1	WRAP53	POMC	KRT13	KREMEN1	GSN	PMVK
FANCE	ALOX12B	ANOS1	TERT	OCRL	BAP1	TSPEAR	AHSG	CLPX
FANCF	ALOXE3	KANSL1	TINF2	SNAI2	ASXL1	CST6	CBS	PLCD1
FANCI	ABCA12	NLRP3	RTEL1	BTK	KLHL7	RHOA	GATA1	RSPO4
BRIP1	NIPAL4	NEK9	PARN	TYR	STK11	ITPR2	CCBE1	OSMR
PALB2	CERS3	CARD14	ABCC9	SLC45A2	TEK	COL11A1	PIK3R1	IL31RA
RAD51C	CYP4F22	MPLKIP	PSEN1	TYRP1	TRIM37	DPH1	DOCK6	IDS
SLX4	LIPN	GNPAT	RHBDF2	IRF4	HRAS	GREM2	RBPJ	HGSNAT
FANCG	STS	AGPS	NECTIN 4	MC1R	CYP26C1	HR	DLL4	GNS
FANCL	POMP	RNF113A	FERMT1	SLC24A4	MED25	CDH1	XYLT1	GUSB
ERCC4	SPINK5	ASPRV1	USB1	GPR143	PCGF2	FOXL2	TRAIP	FUCA1
MAD2L2	ERCC2	LSS	CDSN	CTC1	SLC24A5	TFAP2A	GNRH1	IL36RN
RAD51	ERCC3	ARSE	MITF	EDNRA	B4GALT7	RMRP	LRP1	AP1S3
XRCC2	GTF2H5	SULT2B1	PAX3	OCA2	KRT74	FIG4	UROS	LTA
BRCA1	TARS1	SREBF1	EDN3	FOXD3	RBM28	ARID1B	APOE	IFIH1
MSH2	KRT10	DSC2	EDNRB	PLCG2	NBAS	ARID1A	FAM111B	IL1RN
MSH6	VPS33B	ENPP1	GNA11	NBN	NOTCH1	SMARCA4	EGFR	CTLA4
PMS2	VIPAS39	NLRP1	CYBA	DSTYK	SLC25A24	DPF2	KCTD1	ZNF750
GNAS	DSP	KLHL24	NCF2	DCLRE1C	JAK1	SMARCC2	BRF1	PNPLA2
PTPN11	SNAP29	DSG1	CYBB	CXCR4	KIT	SEC23A	COL5A2	IRF2BP2
RAD50	SRD5A3	GJB3	NCF4	ADAR	LAMTOR2	ALDH18A	PLOD1	LIG4
SMARCB1	DOLK	GJB4	POR	PTPN22	LRMDA	PYCR1	COL1A1	FZD6
RECQL3	CSTA	GJA1	USP8	TRAC	AP3D1	PORCN	CHST14	STAT3

BRCA2	AP1B1	KRT14	GNAQ	NFKB2	EPG5	ALX4	ADAMTS2	IL6R
KITLG	GJB2	JUP	UBE2T	LYST	CLCN7	LHB	TNXB	ZNF341
UBR1	CLDN1	PERP	SLC6A19	MYO5A	DNAJC21	GNRHR	SLC39A13	CHUK
KMT2D	ELOVL4	KRT6A	HAMP	RAB27A	LIFR	APCDD1	FKBP14	KRT83
SPRED1	GINS1	KRT6B	SLC40A1	AP3B1	B3GALT6	SNRPE	COL1A2	TRPM4
RAF1	CARMIL2	RSPO1	ACD	HPS1	IDUA	RPL21	FLNA	LMBRD1
MEN1	IL2RB	KANK2	EDAR	SOX18	PROC	KRT71	DSE	MVK
NF2	RIN2	AQP5	EDA	ERCC6	PROS1	DSG4	AEBP1	MVD
NRAS	SLURP1	TRPV3	NFKBIA	ERCC8	COL7A1	LIPH	TGFBR1	SLC17A9
PPP1CB	AP1S1	KRT16	EDARADD	UVSSA	MTX2	LPAR6	ENG	FDPS
CBL	MSMO1	KRT6C	PDGFRB	CDKN2A	ITGA6	DSC3	BMS1	PSENNEN

ESCO2	PIGA	CAST	NOTCH3
APOB	HMGCS1	ATR	PTPRC
PCSK9	HMGCS2	BCS1L	RAG1
CCDC115	HTRA1	BLM	RAG2
RBP4	HYAL1	BLNK	RNASEH2 A
RAI1	IDI1	BTD	RNASEH2 B
ABCA1	IDI2	C1QA	RNASEH2 C
APOC2	IGHM	C1QB	RNF135
PSMB4	IPLL1	C1QC	SAMHD1
POFUT1	IL10	CAPN12	SART3
POGLUT1	IL17F	CARD11	SCN9A
ADAM10	IL2RG	CARD9	SDHB

NCSTN	IL6ST	CASP14	SDHD
SMAD3	IL7R	CD3D	SDR9C7
TGFBR2	IRF6	CD79A	SF3B1
WAS	KDSR	CD79B	SHARPIN
FOXP3	KL	CFTR	SLC30A2
ARPC1B	KRIT1	CHI3L1	SLC39A4
TUBB	KRT26	CHST8	SMO
PADI3	KRT27	COG6	SMPD1
TSC2	KRT28	COX4I2	SPINT2
KRT9	KRT7	CPOX	SSH1
HFE	KRT72	CRLF3	ST3GAL5
HERC2	KRT73	CXCR1	STAT5B
FCGR2A	LAMC1	CYSLTR2	SUZ12P1
PTCH1	LCA5	DLX3	TAOK2
CTNNB1	LDHA	DOCK8	TBC1D24
MLH1	LDLR	ECM1	TBK1
ITGB4	LMX1B	EGF	TCF4
KRT81	LPIN2	EIF1AX	TFE3
LAMB3	LRRC8A	EIF2AK3	TGFB2
PSMB9	LYNX1	ELOVL1	TLR9
FCGR2B	LYZ	EPHB4	TMEM165
SUFU	MANBA	EPS8L3	STING1
MMP1	MBTPS1	EVC	TMEM232
COL17A1	MEFV	F12	TNFAIP3

TREX1	MMACHC	FANCM	TRIM32
DNASE1	MMP2	FBN1	TYK2
ABCB9	MPDU1	FECH	UBE2A
ABCC11	MRE11	FH	UROD
ABCC6	MUTYH	FOXC2	UTP6
ABCG8	NAGA	FOXE1	VDR
ACTA1	NEU1	FOXN1	VEGFA
ACTA2	NHEJ1	FREM1	WIPF1
ACVRL1	NPC1	GAN	WNT7A
ADAM17	NPC2	GATA2	WRN
ADAMTS1	OTULIN	GBA	XYLT2
ADAP2	PAH	GGPS1	ZNF592
AGA	PEX1	GJC2	ZPR1
AGGF1	PIGO	GLA	NECTIN4
AKT2	PIGV	GLB1	CDK4
AKT3	PIK3R2	GLI3	ITGA3
ALAD	PLG	GLMN	NTRK1
ALAS1	PLOD3	GNPTG	CLEC7A
ASL	POLA1	GPNMB	LDLRAP1
ASXL3	PPP1R13L	HARS	ATP2C1
ATAD5	PSMA3	HLCS	PSTPIP1
ATIC	PSPH	HMBS	HMGCR