

Supplemental Data

Copy-Number Mutations on Chromosome 17q24.2-q24.3

in Congenital Generalized Hypertrichosis Terminalis

with or without Gingival Hyperplasia

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Table S1. Primers for two-point linkage analysis

Amplicon	Primer Sequence (5'-3')		Amplicon Position	
	Forward	Reverse	Start*	Stop*
D17S789	CTGCATACGAAGGGTAGGAC	ACTCCAAATCAAGTTTGTACTGAGA	64140160	64140315
D17S795	TGAGTTGTTAGCAATACTCGTAGG	TTTTCATGCGCTATCTCCAG	64189799	64189903
D17S2182	CAGGGGAGAGCAAAGCTGTATT	TCTGGACAGGTTATTATACTGGG	64651512	64651725
D17S1786	GAGACTCTGTATGGCACCC	AAACTGGATAACGCCCTG	65006332	65006492
D17S1295	TCTAGCCGTTGTGTGGTA	TTCTCAGACTCTCAGTCTTGG	65525564	65525782
D17S1534	TTTGGAGTGAAGTTAAAGA	ATTTTCAAGTACAGATAAAACTGCC	67409223	67409451
D17S1351	CCAAAGTCCTAAAGGTGGG	TTTCAGGCAAATAAGGCAG	68183747	68183919

* Genomic position of PCR primers corresponding to the March 2006 human genome

reference sequence NCBI Build 36.1

Table S2. Primers for qPCR assays

Amplicon	Primer Sequence (5'-3')		Amplicon Position	
	Forward	Reverse	Start*	Stop*
1	GAGCATTGGGAGAGCATCAGG	TCAAGGGCATAAGCTCAGCG	64258990	64259090
2 (<i>ABCA8</i>)	CCTGCCTGCATGGATTTGA	CCAGATGGAAGCCTCAGGAC	64414596	64414696
3 (<i>ABCA9</i>)	TGCAGAAGGGTAGAGGCCTG	CCCAAACCTTCAGCTCTTCAGAAC	64515924	64516024
4 (<i>ABCA6</i>)	CCATGATGCTCTGAAACAATAACAC	AGATTCAGCATTCTGGTGAGTGAC	64641291	64641391
5 (<i>ABCA10</i>)	AGCAATTTGACCACAAGCTGTCTT	TGTTTTCGACTTTGCTTTGCC	64735434	64735529
6 (<i>ABCA5</i>)	TTAAGCTGACCCTATGTGGGCA	GTCTCTTTGGCTGTCTATCTTGATC	64808805	64808911
7 (<i>MAP2K6</i>)	GGTTGTCTGGTCACTGAGGTCAC	CAGGGAACCTGAGACAGGCTAC	65042119	65042219
8	TGAAACAAAAGTGTGTGTTGGCA	GGAACAAAAGCTTCTCACCATCTG	65272254	65272345
9 (<i>KCNJ16</i>)	GCAAAAGATGCATTCAGTAAGCTG	GGTCCAAGTGGCAAACCTAGTG	65641524	65641614
10 (<i>KCNJ2</i>)	AGGGAAGATGCCACACCTGG	TCCCTGCTTTGGCATTTC	65685299	65685399
11	GAAGCAGCCGTGTAATCCTCC	CAGGGTGTCTGAGATTCCCATG	65747075	65747175

* Genomic position of PCR primers corresponding to the March 2006 human genome reference sequence NCBI Build 36.1

Table S3. Primers for analysis of breakpoint junctions

Family	Primer Sequence (5'-3')		Position	
	Forward	Reverse	Start*	Stop*
SY	GAGTGAGACCATCAGGGTGTGAAAC TGG	GTTGGCTTCCCAATGCGGTAGGATG	64340824	65305425
GD	CAAGGGGAATGGTCCATCTTTTGGC	CTGCATAGGTAATGTGGTGTCTTCCA GG	64583839	65139513
BJ	CTCCTGTCTCCACACTCTGAGCCTC	AGACAGGCTGTGCCTTAATCCTCTC	64491005	65169893

* Genomic position of PCR primers corresponding to the March 2006 human genome reference sequence NCBI Build 36.1

Table S4. Primers for RT-PCR analysis

Gene	Primer Sequence (5'-3')		Amplicon Position	
	Forward	Reverse	Start*	Stop*
<i>ABCA5</i>	GCAGCGTTGGCTTGTATTGC	CTCCTCACAACTGGCAACC	chr17: 64768976	64763913
<i>ABCA6</i>	GGTCTATGCCCGTAGATGGTTC	TACTGCCCTCAAGAGAACGTGC	chr17: 64592019	64592876
<i>ABCA10</i>	GTCGGTCTACTTTATGCTCCTTCAG	AGGACAGATCACTGCAATACTTGG	chr17: 64700894	64702209
<i>MAP2K6</i>	GGTTGCAAACCATACATGGCC	GATGCCACATCTGTTCTTTGG	chr17: 65031357	65049461
<i>KRT15</i>	TCGTGGTTCTTCTTCAGGTAGGC	GGGTTTTGGTGGTGGCTTTG	chr17: 36926590	36928345
<i>ACTB</i>	GTAGTTTCGTGGATGCCACAGG	TACAATGAGCTGCGTGTGGCT	chr7: 5534312	5535410

* Genomic position of PCR primers corresponding to the March 2006 human genome reference sequence NCBI Build 36.1

Table S5. Two-point LOD scores for markers on chromosome 17q24.2-q24.3

Marker	$\theta =$						
	0.00	0.01	0.05	0.10	0.20	0.30	0.40
D17S789	$-\infty$	0.08	0.67	0.83	0.83	0.65	0.38
D17S795	$-\infty$	1.56	2.04	2.06	1.75	1.23	0.59
D17S2182	3.91	3.85	3.60	3.27	2.56	1.75	0.84
D17S1786	3.91	3.85	3.60	3.27	2.56	1.75	0.84
D17S1295	2.41	2.37	2.21	2.00	1.54	1.02	0.45
D17S1534	3.15	3.10	2.86	2.56	1.91	1.19	0.47
D17S1351	$-\infty$	-0.14	1.04	1.36	1.35	1.01	0.49

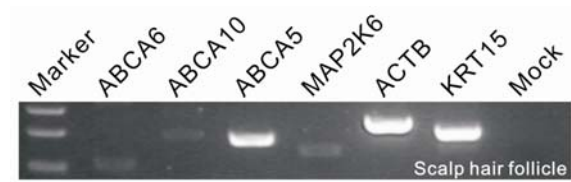


Figure S1. Expression of mRNA for *ABCA5*, *ABCA6*, *ABCA10*, *MAP2K6* and control genes in human scalp hair follicles.

Marker: DL2,000 DNA marker (TaKaRa); *ACTB*: the β -actin house keeping gene; *KRT15*: the keratin 15 gene expressed in the hair follicle stem cells. Mock: negative control.