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Supplemental Data

X-Linked Congenital Hypertrichosis Syndrome

Is Associated with Interchromosomal Insertions

Mediated by a Human-Specific Palindrome near *SOX3*

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Table S1. PCR Primers Used in the Study

For microsatellite markers

Marker	Forward Primer (5'-3')	Reverse Primer (5'-3')
ZLS1	GGGCAACAGAGAGTGAGAC	GGCCCACCCTAATCCAATAT
ZLS2	TTAGCCAGACCTCACAATTG	GCAGTATTGGTAGGATATCT
ZLS3	GCCCAAAGCTAGTCAACAG	AGGCTGCAATGAGCTGTAA
ZLS4	TCTTCAACCAGAGTACCCATTG	GTTCTCTTTACAGACAGTTCATC
ZLS5	GCAGAAATTACCTATATCTTC	GAATGGATGGAGAGATGTAT
ZLS6	GGAAGGAGGAAGGAAGGAAAC	GCACTCTTGGGCACTTATC
ZLS7	AGCAGTTTCGTGGTTTGTTC	AAGCCTGGGTAGTCGTAGTT
DXS1211	GGCAGAACTTGATAGGTAGAT	TCTTTTCTAGACCCTTCTGGC
ZLS8	TGTCCTGTCATGTTGGTAAAT	CATGCTCATGATTTAACACTG
DXS1227	AGAGGTCCGAGTCTTCCAC	ATAAGGGTTTACTCCCCCAA
ZLS9	GAGGAACAGTATCTTCTTGGTC	TGAGATTGTGTCCAGGTCTT
ZLS10	TGAAATCTGTTAGCCTTCTTG	GCATATTTGCATAATGAGGAT
ZLS11	CTACATATCTACACCATCCTC	ATGAGGCATCAGTGGAGGTC
DXS8106	AGTTCACATCCGTATCTAC	AGCTGTAGAGTTGAGGAATG

For qPCR assays

Target	Forward Primer (5'-3')	Reverse Primer (5'-3')
5q35.3	TCCTCTCAACAATTAGATGCCTGTAG	ACCAAGTTTCACACCAGTTTCCAT
4q31.2	CCAGCCTTGTCATCATCAGATTC	AGTTGGCCGATTCCTCAGAAG

For long-range PCR to amplify insertion junctions

Target	Forward Primer (5'-3')	Reverse Primer (5'-3')
D-JBc	GGTAGCATCTCTCAAATCAACCAGGTAT	AATGCTTTGTAGTGGCTTTGTTTCC
P-JBc	GTTAGCATCTTCAAATCCTGGAAC	CTCAAGGCCTCCATCCTCAATATC
D-JBm	AATGCTTTGTAGTGGCTTTGTTTCC	GAATCGTAAGTTCTGGTGGCATTGTGGG
P-JBm	GTTAGCATCTTCAAATCCTGGAAC	AAGCTCTGCGCAGCTTGCCAGCAC

D-JBc, distal JBPcn; P-JBc, proximal JBPcn; D-JBm, distal JBPmx; P-JBm, proximal JBPmx.

For long-range PCR to screen palindrome-mediated insertions in male controls

Target	Forward Primer (5'-3')	Reverse Primer (5'-3')
Palindrome	TGGCATTACAAGAGTTAGCTTCTGA	AATGCTTTGTAGTGGCTTTGTTTCC

For RT-PCR analysis

Target	Forward Primer (5'-3')	Reverse Primer (5'-3')
SOX3	GTTGGGACGCCTTGTTTACG	TAGCGGAAGAAATATCAAACAG
GAPDH	TGAAGGTCGGAGTCAACGGATTTGGT	CATGTGGGCCATGAGGTCCACCAC

Table S2. Two-Point LOD Scores for Markers on Chromosome Xq26.3-q27.2

Marker	Repeat	$\theta=$				
		0.0	0.1	0.2	0.3	0.4
ZLS1	AGAA	$-\infty$	0.53	0.53	0.37	0.16
ZLS2	AGAT	$-\infty$	2.32	1.95	1.38	0.67
ZLS3	TATT	$-\infty$	1.05	1.01	0.78	0.45
ZLS4	CA	2.37	2.33	2.19	2.01	1.61
ZLS5	TCTA	3.61	3.02	2.35	1.60	0.76
ZLS6	AAGG	3.91	3.27	2.56	1.75	0.84
ZLS7	ATCT	3.91	3.27	2.56	1.75	0.84
DXS1211	AC	3.91	3.27	2.56	1.75	0.84
ZLS8	ATCT	3.91	3.27	2.56	1.75	0.84
DXS1227	CA	2.37	2.01	1.61	1.15	0.62
ZLS9	GT	2.37	2.01	1.61	1.15	0.62
ZLS10	CA	$-\infty$	2.32	1.96	1.39	0.71
ZLS11	CA	$-\infty$	1.06	1.05	0.72	0.25
DXS8106	CA	$-\infty$	-1.72	-1.02	-0.72	-0.41

Table S3. Palindrome-Mediated Deletions in Normal Control Individuals

Ethnic Group	Case	Size (bp) & Position (GRCh37/hg19)	Insert (bp)	Reference
Chinese	114	6,374 (chrX: 139,496,584-139,502,957)		This study
Chinese	4	7,918 (chrX: 139,495,035-139,502,951)	102	This study
Mexican	195CA	1,565 (chrX: 139,501,394-139,502,958)	60	This study
Mexican	179CA	173 (chrX: 139,502,954-139,503,126)	2	This study
Indian	GIP024	7991 (chrX: 139,494,993-139,502,983)		This study
Indian	GIP124	7991 (chrX: 139,494,993-139,502,983)		This study
Indian	GIP163	7991 (chrX: 139,494,993-139,502,983)		This study
Indian	GIP044	9104 (chrX: 139,493,885-139,502,988)	19	This study
Indian	PAT473	7963 (chrX: 139,494,995-139,502,957)		This study
Yoruban		209 (chrX: 139,502,954-139,503,162)		Bentley, et al. 2008

Insert refers to concomitant insertion. Breakpoints within the palindrome are in red.

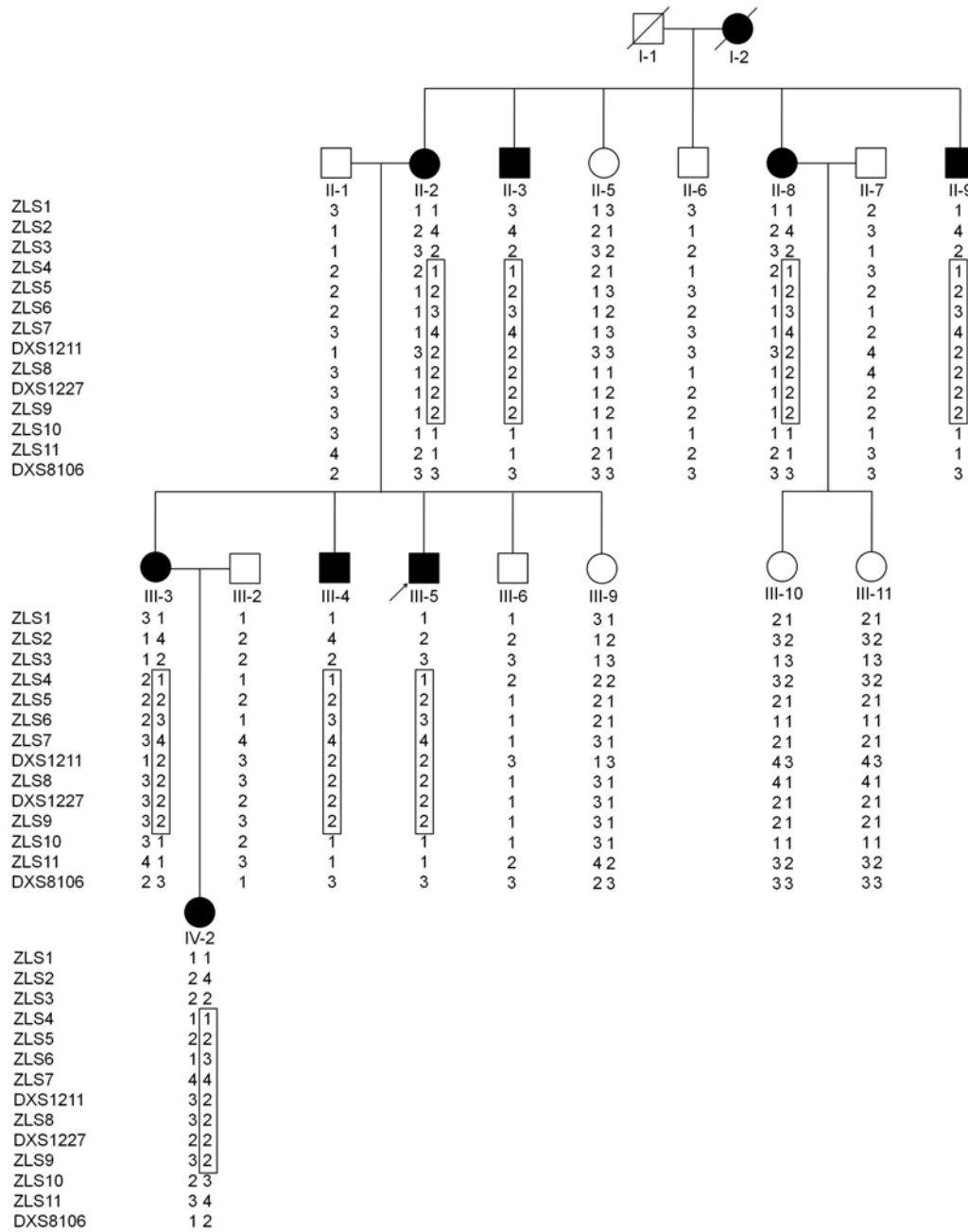


Figure S1. Haplotype Analysis of the Chinese Family with a Distinct X-Linked Hypertrichosis Syndrome

The linked haplotype is boxed.

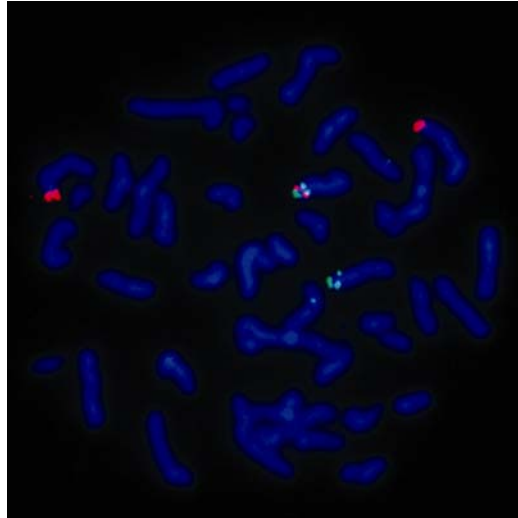


Figure S2. Original Metaphase FISH from which Figure 2D was Prepared

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chr5      177957135+  GGTCCACACATCTTTTGAGATTCTTCCCATCAAAGGCAGAGTGTGGCCAGGTGTGGTGGCTCAA 177957200
Reads with Difference:
*
GB50E9E01AYYGV      150-  GGTCCACACATCTTTTGAGATTCTTCCCATCA 119
GB50E9E02GCDUB      304+  GGTCCACACATCTTTTGAGATTCTTCCCATCA 335
GB50E9E02JL9V0      202-  GGTCCACACATCTTTTGAGATTCTTCCCATCA 171
GB50E9E01BMFP4      255-  GGTCCACACATCTTTTGAGATTCTTCCCATCA 224
GB50E9E02HCIWV      387-  GGTCCACACATCTTTTGAGATTCTTCCCATCA 356
GB50E9E01AWY1N      306-  GGTCCACACATCTTTTGAGATTCTTCCCATCA 275
GB50E9E01CMLRN      117+  GGTCCACACATCTTTTGAGATTCTTCCCATCA 149
GB50E9E02IU8XF      110+  GGTCCACACATCTTTTGAGATTCTTCCCATCA 141
GB50E9E02IMG9B      291-  GGTCCACACATCTTTTGAGATTCTTCCCATCA 260
GB50E9E02JHJMP      382-  GGTCCACACATCTTTTGAGATTCTTCCCATCA 351
GB50E9E01B00PS      331-  GGTCCACACATCTTTTGAGATTCTTCCCATCA 300
*
chrX      139502921+  TTCAGTGCCTTTTAAAAAGCAATATAAGCTATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 139502986
Reads with Difference:
*
GB50E9E01AYYGV      118-  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 84
GB50E9E02GCDUB      336+  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 370
GB50E9E02JL9V0      170-  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 136
GB50E9E01BMFP4      223-  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 189
GB50E9E02HCIWV      355-  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 321
GB50E9E01AWY1N      274-  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 240
GB50E9E01CMLRN      150+  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 184
GB50E9E02IU8XF      142+  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 176
GB50E9E02IMG9B      259-  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 225
GB50E9E02JHJMP      350-  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 316
GB50E9E01B00PS      299-  ATATCTAGCTTATATATTGCTTTTTAAAAAGCCTG 265
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Figure S3. Alignment of 11 MPS Reads to the Human Genome Reference Revealing a Junction between the 5q35.3 and Xq27.1 Sequences

All genomic positions correspond to February 2009 human reference sequence (GRCh37).

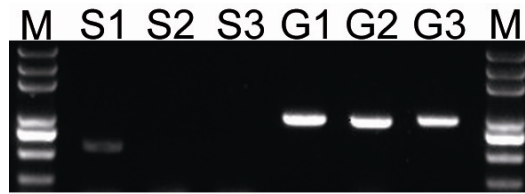


Figure S4. Expression of mRNA for *SOX3* and *GAPDH* in Human Hair Follicles

M: DL2,000 DNA marker (TaKaRa); S1-S3: RT-PCR of *SOX3*; G1-G3: RT-PCR of *GAPDH*; S1 and G1: PCR using fetal brain cDNA control (Clontech); S2 and G2: RT-PCR in scalp hair follicles from an individual with a normal hair growth; S3 and G3: RT-PCR in hair follicles from the proband of the Chinese family.