Heterogeneous lengths of copy number mutations in human coagulopathy revealed by genome-wide high-density SNP array

Hee-Jin Kim,[1,2] Duk-Kyung Kim,[2,3] Ki-Young Yoo,[4] Chur-Woo You,[5] Jong-Ha Yoo,[6] Ki-O Lee,[7] In-Ae Park,[7] Hae-Sun Choung,[1] Hee-Jung Kim,[1] Min-Jung Song,[1] and Sun-Hee Kim[1]

[1]Department of Laboratory Medicine & Genetics, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul; [2]Cardiac & Vascular Center, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul; [3]Department of Medicine, Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul; [4]Korea Hemophilia Foundation, Seoul; [5]Department of Pediatrics, Eulji University School of Medicine, Daejeon; [6]Department of Laboratory Medicine, National Health Insurance Corporation Ilsan Hospital, Goyang; and [7]Samsung Biomedical Research Institute, Samsung Medical Center, Seoul, Korea

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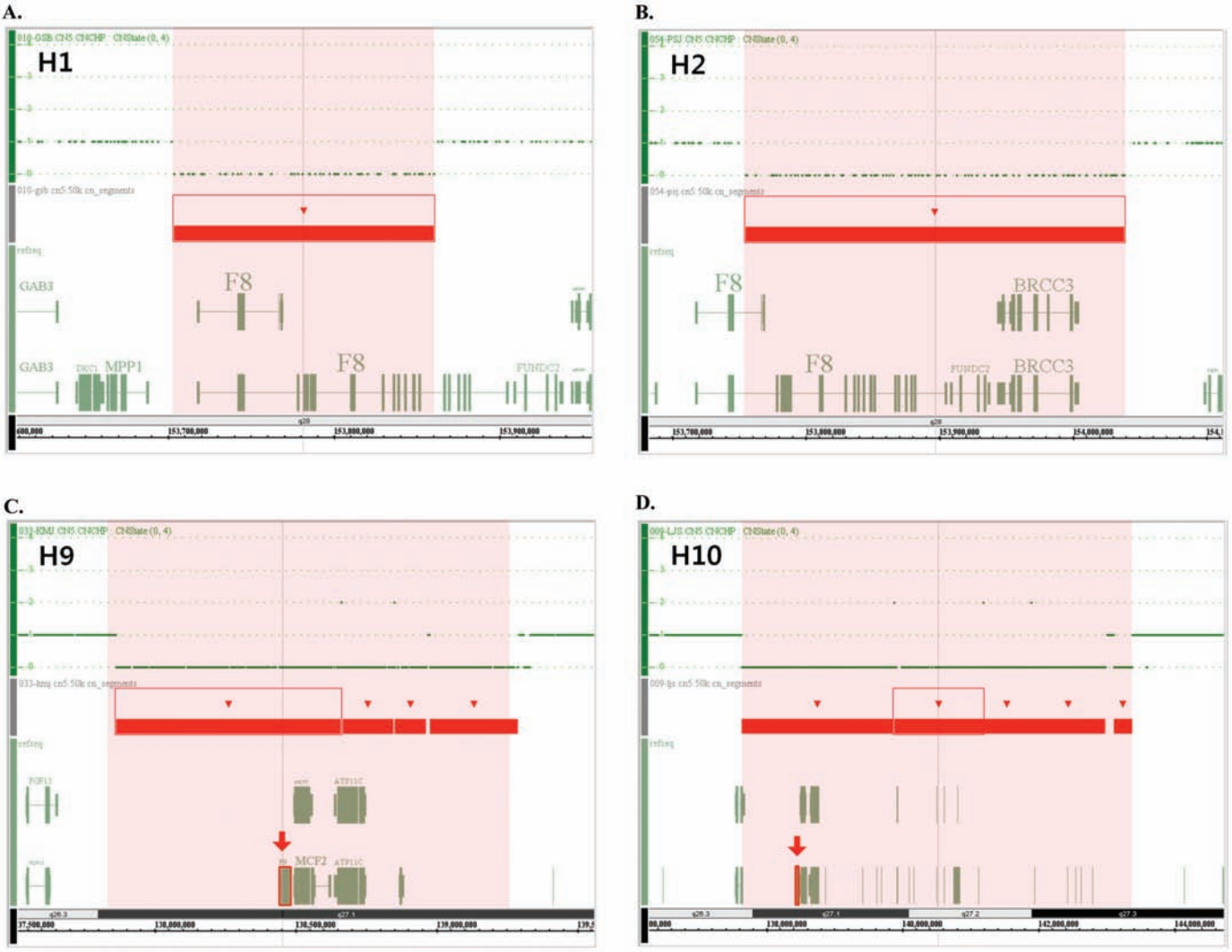
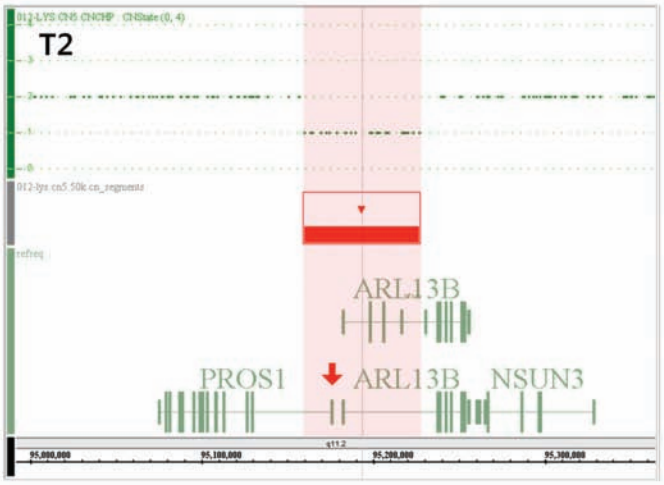
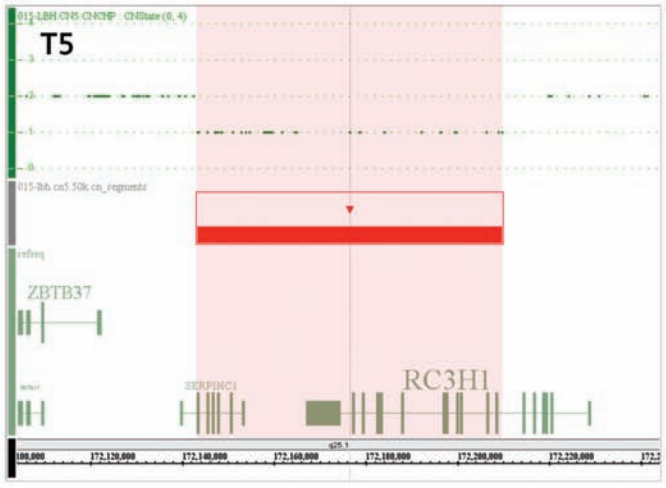


E.

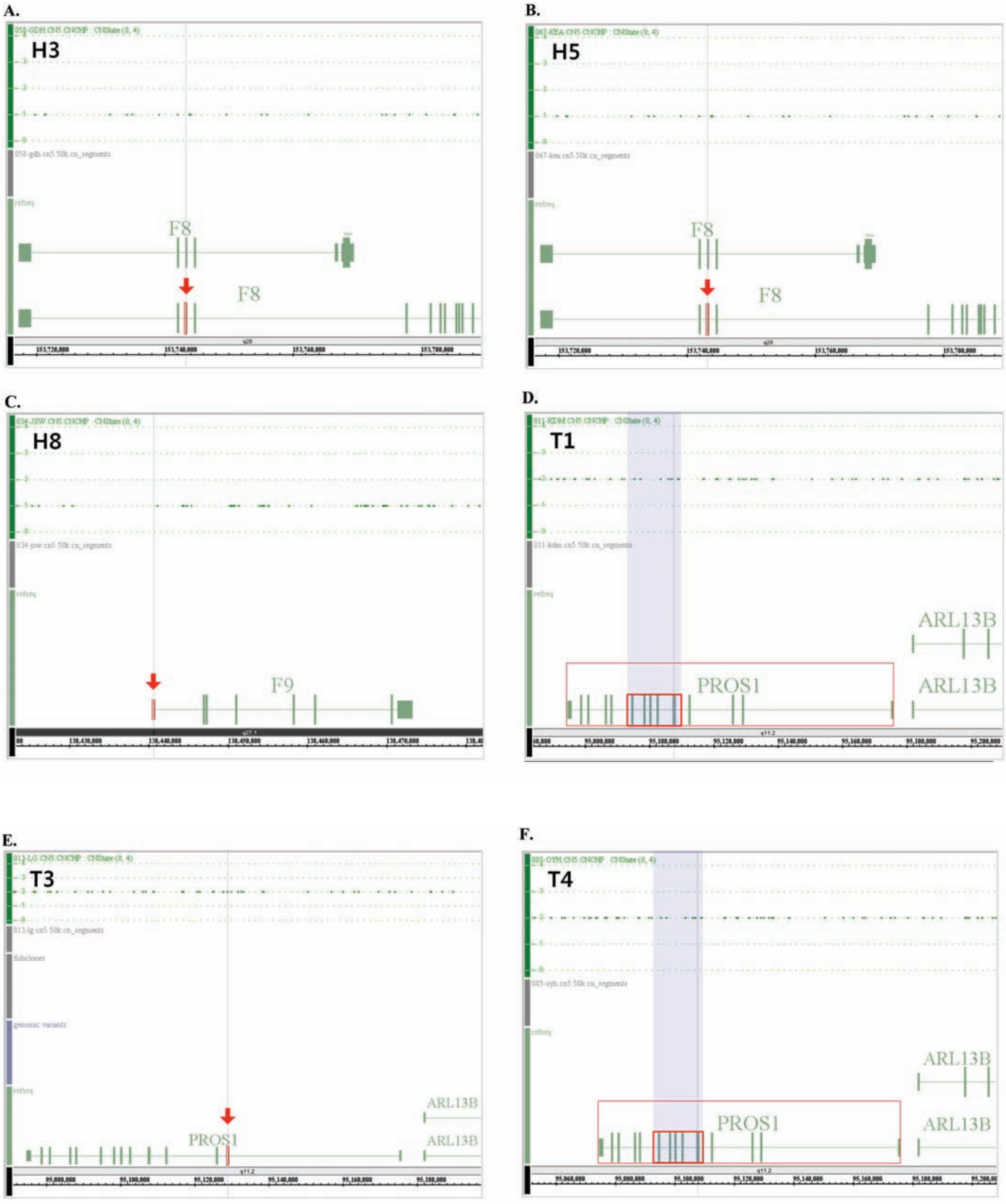
F.



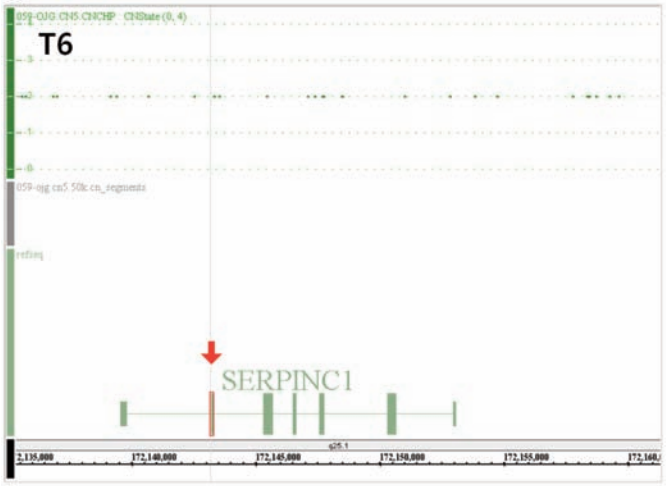
G.



Online Supplementary Figure S1. The screenshots of the Genotyping Console representing the copy number (CN) mutations causing hereditary coagulation disorders revealed by SNP-array experiments obtained by using the Genotyping Console. (A) Patient H1 with severe hemophilia A (HA) with ~157 Kb deletion (CN 0) involving exons 7-26 of *F8*. (B) Patient H2 with severe HA with ~283 Kb deletion (CN 0) involving exons 1-22 of *F8*. Note that the adjacent genes, *FUNDC2* and *BRCC3*, are also included in the deletion segment. (C) Patient H9 with severe hemophilia B (HB) with ~797 Kb deletion (CN 0) involving the whole *F9* gene (small red box in the bottom indicated by a red arrow), along with adjacent genes, *MCF2*, *ATP11C*, and *CXorf66*. (D) Patient H10 with severe HB with ~5,739 Kb deletion (CN 0) involving the whole *F9* gene (small red box in the bottom indicated by a red arrow), along with ~45 adjacent genes. (E) Patient H11 with severe HB with ~2,822 Kb deletion (CN 0) involving the whole *F9* gene (small red box in the bottom indicated by a red arrow), along with ~28 adjacent genes. (F) Patient T2 with protein S deficiency with ~67 Kb deletion (CN 1) involving the exon 1 of *PROS1* (red arrow), along with part of the adjacent *ARL13B* gene. (G) Patient T5 with antithrombin deficiency with ~66 Kb deletion (CN 1) involving exons 1-6, along with part of the adjacent *RC3H1* gene.


A.
H3
F8
F8
B.
H5
F8
F8
C.
H8
F9
D.
T1
ARL13B
PROS1
ARL13B
E.
T3
ARL13B
PROS1
ARL13B
F.
T4
ARL13B
PROS1
ARL13B

G.



Online Supplementary Figure S2. The screenshots of the Genotyping Console representing the SNP-array data showing neutral copy number (CN) status on the genomic area of CN mutations causing hereditary coagulation disorders revealed by multiplex ligation-dependent probe amplification. (A) Patient H3 with severe hemophilia A (HA) from a single exon (exon 24) duplication of *F8* (red arrow). (B) Patient H5 with severe HA from a single exon (exon 24) deletion of *F8* (red arrow). (C) Patient H8 with severe hemophilia B from a single exon (exon 1) deletion of *F9* (red arrow). (D) Patient T1 with protein S (PS) deficiency from duplication of exons 5-10 *PROS1* (small red box). Note that the SNP markers covering this area represent neutral CN status (CN 2). (E) Patient T3 with PS deficiency from deletion of a single exon (exon 2) of *PROS1* (red arrow). (F) Patient T4 with PS deficiency from duplication of exons 5-10 of *PROS1* (small red box). Note that the SNP markers covering this area represent neutral CN status (CN 2). (G) Patient T6 with antithrombin deficiency from deletion of a single exon (exon 6) of *SERPINC1* (red arrow).

Online Supplementary Table S1.

Patient H1

Region Displayed: 153,704K-153,860K bp

Number of Genes in Region: 5 (3*)

start	stop	Symbol	O	Cyto	Description
153704817	153717173	LOC100132963	-	Xq28	similar to hCG1808463
153717260	153904192	F8	-	Xq28	coagulation factor VIII, procoagulant component (hemophilia A)
153766511	153767027	H2AFB1	+	Xq28	H2A histone family, member B1
153767829	153769530	F8A1	+	Xq28	coagulation factor VIII-associated (intronic transcript) 1
153790293	153800681	LOC553820	-	Xq28	eukaryotic translation elongation factor 1 alpha 1 pseudogene

*Only including unique functional genes approved by the HUGO Gene Nomenclature Committee (HGNC).

Patient H2

Region Displayed: 153,755K-154,038K bp

Number of Genes in Region: 7 (6*)

Start	Stop	Symbol	O	Cyto	Description
153717260	153904192	F8	-	Xq28	coagulation factor VIII, procoagulant component (hemophilia A)
153766511	153767027	H2AFB1	+	Xq28	H2A histone family, member B1
153767829	153769530	F8A1	+	Xq28	coagulation factor VIII-associated (intronic transcript) 1
153790293	153800681	LOC553820	-	Xq28	eukaryotic translation elongation factor 1 alpha 1 pseudogene
153908258	153938385	FUNDC2	+	Xq28	FUN14 domain containing 2
153943080	153952830	MTCP1	-	Xq28	mature T-cell proliferation 1
153952904	154004543	BRCC3	+	Xq28	BRCA1/BRCA2-containing complex, subunit 3

*Only including unique functional genes approved by the HUGO Gene Nomenclature Committee (HGNC).

Patient H6

Region Displayed: 153,891K-153,943K bp

Number of Genes in Region: 2 (2*)

Start	Stop	Symbol	O	Cyto	Description
153717260	153904192	F8	-	Xq28	coagulation factor VIII, procoagulant component (hemophilia A)
153908258	153938385	FUNDC2	+	Xq28	FUN14 domain containing 2

*Only including unique functional genes approved by the HUGO Gene Nomenclature Committee (HGNC).

Patient H7

Region Displayed: 148,599K-154,887K bp

Number of Genes in Region: 161 (126*)

Start	Stop	Symbol	O	Cyto	Description
148481974	148663581	LOC100130086	-	Xq28	hypothetical protein LOC100130086
148575477	148604507	MAGEA11	-	Xq28	melanoma antigen family A, 11
148658294	148662686	LOC100132969	+	Xq28	hypothetical protein LOC100132969
148663576	148666336	HSFX1	+	Xq28	heat shock transcription factor family, X linked 1
148671402	148677206	MAGEA9	+	Xq28	melanoma antigen family A, 9
148697998	148698275	MAGEA7	+	Xq28	melanoma antigen family A, 7, pseudogene
148770654	148775266	MAGEA8	+	Xq28	melanoma antigen family A, 8
148845967	148847100	LOC642980	-	Xq28	hypothetical LOC642980
148847832	148850682	LOC100132460	+	Xq28	similar to LOC100126053 protein
148851073	148857374	CXorf40B	-	Xq28	chromosome X open reading frame 40B
148864251	148865366	LOC100129660	+	Xq28	similar to hCG2000002
149033269	149035710	LOC643015	+	Xq28	similar to nucleolar protein 11
149149697	149151799	LOC389901	-	Xq28	
149364378	149433104	MAMLD1	+	Xq28	mastermind-like domain containing 1
149487727	149592272	MTM1	+	Xq28	myotubularin 1
149612527	149684233	MTMR1	+	Xq28	myotubularin related protein 1
149660273	149660722	LOC100129373	+	Xq28	hypothetical protein LOC100129373
149685467	149817837	CD99L2	-	Xq28	CD99 molecule-like 2
149902421	149909906	HMGB3	+	Xq28	high-mobility group box 3
149912632	149913222	LOC392557	+	Xq28	similar to ribosomal protein L19
150094373	150099736	LOC100128688	-	Xq28	hypothetical protein LOC100128688
150095717	150100588	GPR50	+	Xq28	G protein-coupled receptor 50
150144832	150146845	LOC286456	-	Xq28	similar to NGFI-A binding protein 1
150316363	150328494	LOC203547	+	Xq28	hypothetical protein LOC203547
150428124	150435390	LOC100129236	+	Xq28	similar to hCG1653500
150482663	150595867	PASD1	+	Xq28	PAS domain containing 1
150617435	150620669	PRRG3	+	Xq28	proline rich Gla (G-carboxyglutamic acid) 3 (transmembrane)

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150635164	150642322	FATE1	+	Xq28	fetal and adult testis expressed 1
150653874	150664692	CNGA2	+	Xq27	cyclic nucleotide gated channel alpha 2
150823559	150834002	LOC100128125	-	Xq28	similar to hCG1653500
150832017	150844298	MAGEA4	+	Xq28	melanoma antigen family A, 4
150872252	150893807	GABRE	-	Xq28	gamma-aminobutyric acid (GABA) A receptor, epsilon
150877705	150877785	MIRN224	-	Xq28	microRNA 224
150878755	150878839	MIRN452	-	Xq28	microRNA 452
151033182	151037100	MAGEA5	-	Xq28	melanoma antigen family A, 5
151053547	151057681	MAGEA10	-	Xq28	melanoma antigen family A, 10
151086290	151370487	GABRA3	-	Xq28	gamma-aminobutyric acid (GABA) A receptor, alpha 3
151311346	151311426	MIRN105-1	-	Xq28	microRNA 105-1
151312548	151312656	MIRN767	-	Xq28	microRNA 767
151313539	151313619	MIRN105-2	-	Xq28	microRNA 105-2
151369753	151400152	KRT8P8	+	Xq28	keratin 8 pseudogene 8
151557293	151572481	GABRQ	+	Xq28	gamma-aminobutyric acid (GABA) receptor, theta
151617901	151621470	MAGEA6	+	Xq28	melanoma antigen family A, 6
151627399	151628403	CSAG2	-	Xq28	CSAG family, member 2
151634487	151637752	MAGEA2B	+	Xq28	melanoma antigen family A, 2B
151646936	151647478	LOC100130935	-	Xq28	similar to CSAG family, member 2
151649949	151653828	MAGEA12	-	Xq28	melanoma antigen family A, 12
151653884	151660174	CSAG1	+	Xq28	chondrosarcoma associated gene 1
151669044	151673020	MAGEA2	-	Xq28	melanoma antigen family A, 2
151678543	151679391	CSAG3B	+	Xq28	CSAG family, member 3B
151685308	151688896	MAGEA3	-	Xq28	melanoma antigen family A, 3
151703603	151708516	psMAGEA	-	Xq28	melanoma antigen pseudogene, family A
151746527	151749957	CETN2	-	Xq28	centrin, EF-hand protein, 2
151750167	151788563	NSDHL	+	Xq28	NAD(P) dependent steroid dehydrogenase-like
151833653	151892678	ZNF185	+	Xq28	zinc finger protein 185 (LIM domain)
151908024	151911417	PNMA5	-	Xq28	paraneoplastic antigen like 5
151947786	151948925	LOC100128960	+	Xq28	similar to hCG1645335
151970965	151975594	LOC100129956	-	Xq28	hypothetical LOC100129956
151975422	151979483	PNMA3	+	Xq28	paraneoplastic antigen MA3
151991521	151994057	PNMA6A	+	Xq28	paraneoplastic antigen like 6A
151994652	151996650	PNMA6B	-	Xq28	paraneoplastic antigen like 6B
152041130	152046934	LOC728307	-	Xq28	similar to melanoma associated antigen (mutated) 1-like 1
152134716	152139310	MAGEA1	-	Xq28	melanoma antigen family A, 1 (directs expression of antigen MZ2-E)
152190272	152196454	LOC728317	+	Xq28	similar to melanoma associated antigen (mutated) 1-like 1
152237415	152253805	LOC649201	-	Xq28	similar to paraneoplastic antigen MA3
152262060	152270249	ZNF275	+	Xq28	zinc finger protein 275
152315381	152317501	LOC649238	-	Xq28	similar to paraneoplastic antigen MA3
152329775	152340280	ZFP92	+	Xq28	zinc finger protein 92 homolog (mouse)
152363372	152365139	TREX2	-	Xq28	three prime repair exonuclease 2
152366318	152389283	UCHL5IP	-	Xq28	UCHL5 interacting protein
152392294	152405740	LOC389904	+	Xq28	similar to extracellular matrix protein 2
152413605	152428198	BGN	+	Xq28	biglycan
152454774	152501581	ATP2B3	+	Xq28	ATPase, Ca++ transporting, plasma membrane 3
152506577	152517775	FAM58A	-	Xq28	family with sequence similarity 58, member A
152521870	152523799	KRT18P48	-	Xq28	keratin 18 pseudogene 48
152525166	152529360	LOC100131652	+	Xq28	hypothetical LOC100131652
152561182	152569971	DUSP9	+	Xq28	dual specificity phosphatase 9
152580778	152581390	LOC347544	+	Xq28	ribosomal protein L18a pseudogene
152588417	152592974	PNCK	-	Xq28	pregnancy upregulated non-ubiquitously expressed CaM kinase
152606586	152615234	SLC6A8	+	Xq28	solute carrier family 6 (neurotransmitter transporter, creatine), member 8
152619146	152643081	BCAP31	-	Xq28	B-cell receptor-associated protein 31
152643530	152663375	ABCD1	+	Xq28	ATP-binding cassette, sub-family D (ALD), member 1
152682905	152697989	PLXNB3	+	Xq28	plexin B3
152699704	152704381	SRPK3	+	Xq28	SFRS protein kinase 3
152704415	152713161	IDH3G	-	Xq28	isocitrate dehydrogenase 3 (NAD+) gamma
152713288	152717148	SSR4	+	Xq28	signal sequence receptor, delta (translocon-associated protein delta)
152720817	152749197	PDZD4	-	Xq28	PDZ domain containing 4
152759900	152760217	CYCSP45	+	Xq28	cytochrome c, somatic pseudogene 45
152780581	152794505	L1CAM	-	Xq28	L1 cell adhesion molecule
152799321	152807638	LCAP	+	Xq28	lung carcinoma-associated protein
152823564	152825834	AVPR2	+	Xq28	arginine vasopressin receptor 2 (nephrogenic diabetes insipidus)

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152826025	152844892	ARHGAP4	-	Xq28	Rho GTPase activating protein 4
152848571	152853662	ARD1A	-	Xq28	ARD1 homolog A, N-acetyltransferase (S. cerevisiae)
152853917	152863426	RENBP	-	Xq28	renin binding protein
152866202	152890013	HCFC1	-	Xq28	host cell factor C1 (VP16-accessory protein)
152891185	152901840	TMEM187	+	Xq28	transmembrane protein 187
152929151	152938536	IRAK1	-	Xq28	interleukin-1 receptor-associated kinase 1
152940458	153016323	MECP2	-	Xq28	methyl CpG binding protein 2 (Rett syndrome)
153013619	153016356	LOC100128952	+	Xq28	hypothetical protein LOC100128952
153062939	153077701	OPN1LW	+	Xq28	opsin 1 (cone pigments), long-wave-sensitive (color blindness, protan)
153077878	153098828	TEX28P2	-	Xq28	testis expressed 28 pseudogene 2
153101361	153114725	OPN1MW	+	Xq28	opsin 1 (cone pigments), medium-wave-sensitive (color blindness, deutan)
153114908	153136049	TEX28P1	-	Xq28	testis expressed 28 pseudogene 1
153138461	153151953	OPN1MW2	+	Xq28	opsin 1 (cone pigments), medium-wave-sensitive 2
153152126	153176632	TEX28	-	Xq28	testis expressed 28
153177345	153211894	TKTL1	+	Xq28	transketolase-like 1
153230091	153252845	FLNA	-	Xq28	filamin A, alpha (actin binding protein 280)
153254916	153255347	LOC100131857	-	Xq28	hypothetical protein LOC100131857
153260981	153263075	EMD	+	Xq28	emerin (Emery-Dreifuss muscular dystrophy)
153279912	153283874	RPL10	+	Xq28	ribosomal protein L10
153281816	153281950	SNORA70	+	Xq28	small nucleolar RNA, H/ACA box 70
153282773	153293621	DNASE1L1	-	Xq28	deoxyribonuclease I-like 1
153293071	153303259	TAZ	+	Xq28	tafazzin (cardiomyopathy, dilated 3A (X-linked); endocardial fibroelastosis 2; Barth syndrome)
153310172	153318056	ATP6AP1	+	Xq28	ATPase, H+ transporting, lysosomal accessory protein 1
153318715	153325008	GDI1	+	Xq28	GDP dissociation inhibitor 1
153325702	153332190	FAM50A	+	Xq28	family with sequence similarity 50, member A
153339817	153355179	PLXNA3	+	Xq28	plexin A3
153358435	153360790	LAGE3	-	Xq28	L antigen family, member 3
153365250	153368126	UBL4A	-	Xq28	ubiquitin-like 4A
153368842	153372189	SLC10A3	-	Xq28	solute carrier family 10 (sodium/bile acid cotransporter family), member 3
153387700	153397567	FAM3A	-	Xq28	family with sequence similarity 3, member A
153412800	153428981	G6PD	-	Xq28	glucose-6-phosphate dehydrogenase
153423653	153446455	IKBKG	+	Xq28	inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase gamma
153449493	153463455	LOC643894	+	Xq28	[illegible]
153452673	153453385	LOC100132967	+	Xq28	similar to hCG2042244
153462002	153463637	LOC340600	+	Xq28	cyclic-AMP-dependent transcription factor ATF-4 pseudogene
153466610	153468269	CTAG1A	+	Xq28	cancer/testis antigen 1A
153499059	153500714	CTAG1B	-	Xq28	cancer/testis antigen 1B
153513930	153514642	CXorf52	-	Xq28	chromosome X open reading frame 52
153520760	153529845	IKBKGP	-	Xq28	inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase gamma pseudogene
153533445	153535036	CTAG2	-	Xq28	cancer/testis antigen 2
153540101	153541229	OR3B1P	-	Xq28	olfactory receptor, family 3, subfamily B, member 1 pseudogene
153556720	153632542	GAB3	-	Xq28	GRB2-associated binding protein 3
153644344	153659154	DKC1	+	Xq28	dyskeratosis congenita 1, dyskerin
153649997	153650128	SNORA36A	+	Xq28	small nucleolar RNA, H/ACA box 36A
153656467	153656595	SNORA56	+	Xq28	small nucleolar RNA, H/ACA box 56
153660162	153686957	MPP1	-	Xq28	membrane protein, palmitoylated 1, 55kDa
153697668	153699090	LOC728776	-	Xq28	high-mobility group nucleosome binding domain 1 pseudogene
153704817	153717173	LOC100132963	-	Xq28	similar to hCG1808463
153717260	153904192	F8	-	Xq28	coagulation factor VIII, procoagulant component (hemophilia A)
153766511	153767027	H2AFB1	+	Xq28	H2A histone family, member B1
153767829	153769530	F8A1	+	Xq28	coagulation factor VIII-associated (intronic transcript) 1
153790293	153800681	LOC553820	-	Xq28	eukaryotic translation elongation factor 1 alpha 1 pseudogene
153908258	153938385	FUNDC2	+	Xq28	FUN14 domain containing 2
153943080	153952830	MTCP1	-	Xq28	mature T-cell proliferation 1
153952904	154004543	BRCC3	+	Xq28	BRCA1/BRCA2-containing complex, subunit 3
154097744	154121292	VBP1	+	Xq28	von Hippel-Lindau binding protein 1
154140720	154147046	RAB39B	-	Xq28	RAB39B, member RAS oncogene family
154158694	154217180	CLIC2	-	Xq28	chloride intracellular channel 2
154174358	154176025	PHF10P1	+	Xq28	PHD finger protein 10 pseudogene 1
154231158	154233743	LOC553939	+	Xq28	trimethyllysine hydroxylase, epsilon pseudogene
154263630	154263977	H2AFB2	+	Xq28	H2A histone family, member B2
154264958	154266073	F8A2	+	Xq28	coagulation factor VIII-associated (intronic transcript) 2
154340340	154341455	F8A3	-	Xq28	coagulation factor VIII-associated (intronic transcript) 3
154342274	154342790	H2AFB3	-	Xq28	H2A histone family, member B3

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154372967	154495791	TMLHE	-	Xq28	trimethyllysine hydroxylase, epsilon
154650645	154665311	SPRY3	+	q28 and Yq1	sprouty homolog 3 (Drosophila)
154710950	154712518	AMDP1	-	Xq22-q28	S-adenosylmethionine decarboxylase pseudogene 1
154764207	154826614	VAMP7	+	q28 and Yq1	vesicle-associated membrane protein 7
154868104	154869205	TRPC6P	-	Xq28;Yq12	transient receptor potential cation channel, subfamily C, member 6 pseudogene
154880440	154893676	IL9R	+	q28 and Yq1	interleukin 9 receptor

*Only including unique functional genes approved by the HUGO Gene Nomenclature Committee (HGNC).

Patient H9

Region Displayed: 137,864K-139,329K bp

Number of Genes in Region: 13 (7*)

Start	Stop	Symbol	O	Cyto	Description
138271765	138272307	CXorf19	+	Xq27	chromosome X open reading frame 19
138356541	138358898	SRD5AP1	+	Xq24-qter	steroid-5-alpha-reductase, alpha polypeptide pseudogene 1 (3-oxo-5 alpha-steroid delta 4-dehydrogenase alpha pseudogene)
138440561	138473283	F9	+	Xq27.1-q27.2	coagulation factor IX (plasma thromboplastic component, Christmas disease, hemophilia B)
138491596	138618047	MCF2	-	Xq27	MCF.2 cell line derived transforming sequence
138630404	138630607	BCYRN1P1	+	Xq27.1	brain cytoplasmic RNA 1, pseudogene 1
138636171	138742113	ATP11C	-	Xq27.1	ATPase, class VI, type 11C
138833972	138834055	MIRN505	-	Xq27.1	microRNA 505
138865550	138875345	LOC347487	-	Xq27.1	hypothetical LOC347487
138927087	138927864	LOC728660	+	Xq27.1	hypothetical protein LOC728660
138942029	138943795	LOC643689	+	Xq27.1	heterogeneous nuclear ribonucleoprotein A3 pseudogene
139001492	139002736	LOC389895	+	Xq27.1	similar to CG4768-PA
139125851	139126622	LOC729078	-	Xq27.1	hypothetical protein LOC729078
139306993	139307824	LOC266694	+	Xq27.1	embryonic ectoderm development pseudogene

*Only including unique functional genes approved by the HUGO Gene Nomenclature Committee (HGNC).

Patient H10

Region Displayed: 137,633K-143,373K bp

Number of Genes in Region: 42 (28*)

Start	Stop	Symbol	O	Cyto	Description
137541401	137649181	FGF13	-	Xq26.3	fibroblast growth factor 13
137716202	137716994	LOC100130620	+	Xq26.3	hypothetical LOC100130620
138271765	138272307	CXorf19	+	Xq27	chromosome X open reading frame 19
138356541	138358898	SRD5AP1	+	Xq24-qter	steroid-5-alpha-reductase, alpha polypeptide pseudogene 1 (3-oxo-5 alpha-steroid delta 4-dehydrogenase alpha pseudogene)
138440561	138473283	F9	+	Xq27.1-q27.2	coagulation factor IX (plasma thromboplastic component, Christmas disease, hemophilia B)
138491596	138618047	MCF2	-	Xq27	MCF.2 cell line derived transforming sequence
138630404	138630607	BCYRN1P1	+	Xq27.1	brain cytoplasmic RNA 1, pseudogene 1
138636171	138742113	ATP11C	-	Xq27.1	ATPase, class VI, type 11C
138833972	138834055	MIRN505	-	Xq27.1	microRNA 505
138865550	138875345	LOC347487	-	Xq27.1	hypothetical LOC347487
138927087	138927864	LOC728660	+	Xq27.1	hypothetical protein LOC728660
138942029	138943795	LOC643689	+	Xq27.1	heterogeneous nuclear ribonucleoprotein A3 pseudogene
139001492	139002736	LOC389895	+	Xq27.1	similar to CG4768-PA
139125851	139126622	LOC729078	-	Xq27.1	hypothetical protein LOC729078
139306993	139307824	LOC266694	+	Xq27.1	embryonic ectoderm development pseudogene
139412818	139414891	SOX3	-	Xq27.1	SRY (sex determining region Y)-box 3
139421081	139624662	LOC286411	+	Xq27.1	hypothetical protein LOC286411
139693091	139694389	CDR1	-	Xq27.1-q27.2	cerebellar degeneration-related protein 1, 34kDa
139912422	139913537	LOC100133171	+	Xq27.1	hypothetical protein LOC100133171
139924427	139925545	SPANXB1	+	Xq27.1	SPANX family, member B1
140060218	140061190	dJ507I15.1	+	Xq27.1	ribosomal protein L36a pseudogene
140097596	140098976	LDOC1	-	Xq27	leucine zipper, down-regulated in cancer 1
140163262	140164312	SPANXC	-	Xq27.1	SPANX family, member C
140185124	140186001	LOC100132808	-	Xq27.2	hypothetical LOC100132808
140302507	140303107	NDUFB3P5	+	Xq26.3-q27.3	NADH dehydrogenase (ubiquinone) 1 beta subcomplex, 3, 12kDa pseudogene 5
140418515	140565735	CXorf18	+	Xq27.2	chromosome X open reading frame 18
140499472	140500502	SPANXA1	-	Xq27.1	sperm protein associated with the nucleus, X-linked, family member A1
140505498	140506567	SPANXA2	+	Xq27.1	SPANX family, member A2
140541651	140554453	LOC645188	+	Xq27.2	hypothetical LOC645188
140613234	140614264	SPANXD	-	Xq27.1	SPANX family, member D
140753768	140813284	MAGEC3	+	Xq27.2	melanoma antigen family C, 3
140819346	140824849	MAGEC1	+	Xq26	melanoma antigen family C, 1
141063478	141091530	LOC392555	-	Xq27.2	similar to Melanoma-associated antigen C2 (MAGE-C2 antigen) (MAGE-E1 antigen) (Hepatocellular carcinoma-associated antigen 587) (Cancer-testis antigen 187) (CT18)

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141117796	141120742	MAGEC2	-	Xq27	melanoma antigen family C, 2
141941370	141949732	SPANXN4	+	Xq27.3	SPANX family, member N4
142424230	142432973	SPANXN3	-	Xq27.3	SPANX family, member N3
142510323	142511589	MYCL3	-	Xq27.2	v-myc myelocytomatosis viral oncogene homolog 3 (avian) (pseudogene)
142543584	142550685	SLITRK4	-	Xq27.3	SLIT and NTRK-like family, member 4
142587891	142589106	LOC100129368	-	Xq27.3	hypothetical LOC100129368
142622721	142632182	SPANXN2	-	Xq27.3	SPANX family, member N2
142794839	142796028	UBE2NL	+	Xq27.3	ubiquitin-conjugating enzyme E2N-like
143219843	143220608	RRM2P4	+	Xq27	ribonucleotide reductase M2 polypeptide pseudogene 4

*Only including unique functional genes approved by the HUGO Gene Nomenclature Committee (HGNC).

Patient H11

Region Displayed: 137,702K-140,524K bp

Number of Genes in Region: 27 (16*)

Start	Stop	Symbol	O	Cyto	Description
137716202	137716994	LOC100130620	+	Xq26.3	hypothetical LOC100130620
138271765	138272307	CXorf19	+	Xq27	chromosome X open reading frame 19
138356541	138358898	SRD5AP1	+	Xq24-qter	(steroid-5-alpha-reductase, alpha polypeptide pseudogene 1 (3-oxo-5 alpha-steroid delta 4-dehydrogenase alpha pseudogene)
138440561	138473283	F9	+	Xq27.1-q27.2	coagulation factor IX (plasma thromboplastic component, Christmas disease, hemophilia B)
138491596	138618047	MCF2	-	Xq27	MCF.2 cell line derived transforming sequence
138630404	138630607	BCYRN1P1	+	Xq27.1	brain cytoplasmic RNA 1, pseudogene 1
138636171	138742113	ATP11C	-	Xq27.1	ATPase, class VI, type 11C
138833972	138834055	MIRN505	-	Xq27.1	microRNA 505
138865550	138875345	LOC347487	-	Xq27.1	hypothetical LOC347487
138927087	138927864	LOC728660	+	Xq27.1	hypothetical protein LOC728660
138942029	138943795	LOC643689	+	Xq27.1	heterogeneous nuclear ribonucleoprotein A3 pseudogene
139001492	139002736	LOC389895	+	Xq27.1	similar to CG4768-PA
139125851	139126622	LOC729078	-	Xq27.1	hypothetical protein LOC729078
139306993	139307824	LOC266694	+	Xq27.1	embryonic ectoderm development pseudogene
139412818	139414891	SOX3	-	Xq27.1	SRY (sex determining region Y)-box 3
139421081	139624662	LOC286411	+	Xq27.1	hypothetical protein LOC286411
139693091	139694389	CDR1	-	Xq27.1-q27.2	cerebellar degeneration-related protein 1, 34kDa
139912422	139913537	LOC100133171	+	Xq27.1	hypothetical protein LOC100133171
139924427	139925545	SPANXB1	+	Xq27.1	SPANX family, member B1
140060218	140061190	dJ507I15.1	+	Xq27.1	ribosomal protein L36a pseudogene
140097596	140098976	LDOC1	-	Xq27	leucine zipper, down-regulated in cancer 1
140163262	140164312	SPANXC	-	Xq27.1	SPANX family, member C
140185124	140186001	LOC100132808	-	Xq27.2	hypothetical LOC100132808
140302507	140303107	NDUFB3P5	+	Xq26.3-q27.3	NADH dehydrogenase (ubiquinone) 1 beta subcomplex, 3, 12kDa pseudogene 5
140418515	140565735	CXorf18	+	Xq27.2	chromosome X open reading frame 18
140499472	140500502	SPANXA1	-	Xq27.1	sperm protein associated with the nucleus, X-linked, family member A1
140505498	140506567	SPANXA2	+	Xq27.1	SPANX family, member A2

*Only including unique functional genes approved by the HUGO Gene Nomenclature Committee (HGNC).

Patient T2

Region Displayed: 95,160K-95,227K bp

Number of Genes in Region: 4 (3*)

Start	Stop	Symbol	O	Cyto	Description
95074647	95175395	PROS1	-	3q11.2	protein S (alpha)
95181672	95256813	ARL13B	+	3q11.2	ADP-ribosylation factor-like 13B
95190166	95190956	LOC100129555	-	3q11.2	similar to High-mobility group nucleosome binding domain 1
95215903	95230144	STX19	-	3q11	syntaxin 19

*Only including unique functional genes approved by the HUGO Gene Nomenclature Committee (HGNC).

Patient T5

Region Displayed: 172,143K-172,210K bp

Number of Genes in Region: 2 (2*)

Start	Stop	Symbol	O	Cyto	Description
172139565	172153096	SERPINC1	-	1q23-q25.1	serpin peptidase inhibitor, clade C (antithrombin), member 1
172166975	172228833	RC3H1	-	1q25.1	ring finger and CCCH-type zinc finger domains 1

*Only including unique functional genes approved by the HUGO Gene Nomenclature Committee (HGNC).

***Above information was obtained form the UCSC Genome Browser Build hg18/NCBI Map Viewer Build 36.3.*