

Table S1

Male-pair sequencing-proposed structural variants of >10kb in size mapped to near or within annotated genes across the genome of the index patient (Ensembl 72)

chromosome	variant no.	5' boundary	3' boundary	read depth	size [bp]	SV	Ensembl ID	gene start	gene end	GeneName	type
chr1	1	2583845	2689336	38	105511	DEL	ENSG00000215912	2667415	2718286	TTC34	intronic
chr1	2	5256270	7261255	12	53595	DEL	ENSG00000227077	7276715	7276715	ENSG00000227207	pseudogene
chr1	3	10235140	102388152	9	17012	DEL	ENSG00000162699	102358661	102359603	DNAJA1P5	pseudogene
chr1	3	102351140	102368152	9	17012	DEL	ENSG00000118733	102268130	102426286	OLFM3	intronic
chr1	4	144891176	144908295	6	17119	DEL	ENSG00000178104	144836157	145076186	PDE4DIP	intronic
chr1	5	174792396	174805858	9	13462	DEL	ENSG00000152061	174128548	174964445	RABGAP1L	intronic
chr1	6	184810129	184825656	10	15527	DEL	ENSG00000135842	184759858	184943682	FAM129A	intronic
chr1	7	229607862	229820533	13	17151	DEL	ENSG00000213028	229824746	229825651	ENSG00000213028	pseudogene
chr1	8	247847031	247861292	11	14261	DEL	ENSG00000235749	247830951	247910384	ENSG00000235749	intronic
chr1	9	248048260	248062520	14	14260	DEL	ENSG00000238243	248031277	248060449	OR2W3	intronic
chr10	10	47018242	47064611	140	46369	INV	ENSG00000150165	47011753	47174093	ANXA8L1	intronic
chr10	11	111567051	111582236	17	15185	DEL	ENSG00000224907	111567409	111567887	ENSG00000224907	pseudogene
chr11	12	1910784	1964943	92	54159	DEL	ENSG00000184682	1910375	1912084	C11orf89	unclear
chr11	12	1910784	1964943	92	54159	DEL	ENSG00000229671	1923877	1929145	ENSG00000229671	linc/transcr.
chr11	12	1910784	1964943	92	54159	DEL	ENSG00000252187	1941339	1941488	ENSG00000252187	unclear
chr11	12	1910784	1964943	92	54159	DEL	ENSG00000130592	1872000	1913497	LSP1	unclear
chr11	12	1910784	1964943	92	54159	DEL	ENSG00000130595	1940792	1959936	TNNT3	unclear
chr11	13	6088619	6101135	13	12516	DEL	ENSG00000234895	6088103	6089016	OR52X1P	pseudogene
chr11	14	29003837	29017896	19	14059	DEL	ENSG00000249867	28724162	29085368	ENSG00000249867	intronic
chr11	15	69088711	69141936	16	53225	DEL	ENSG00000172927	69061605	69182494	MEYOV	intronic
chr11	16	9285945	9293666	14	14021	DEL	ENSG00000180773	92827341	9293130	SLC35A4	intronic
chr11	17	101562129	101579339	17	17210	DEL	ENSG00000137672	101322295	101743293	TRPC6	intronic
chr11	18	114420698	114432410	7	11712	DEL	ENSG00000095110	114392437	114430617	NXPE1	intronic
chr12	19	11190412	11254341	12	63929	DEL	ENSG00000256400	11249778	11249873	ENSG00000256400	unclear
chr12	19	11190412	11254341	12	63929	DEL	ENSG00000111215	10977559	11324212	PRR4	unclear
chr12	19	11190412	11254341	12	63929	DEL	ENSG00000212127	11090005	11324172	TAS2R14	unclear
chr12	19	11190412	11254341	12	63929	DEL	ENSG00000253774	11243886	11244912	TAS2R43	unclear
chr12	19	11190412	11254341	12	63929	DEL	ENSG00000228761	11213954	11214893	TAS2R46	unclear
chr12	19	11190412	11254341	12	63929	DEL	ENSG00000256019	11200931	11201855	TAS2R63P	unclear
chr12	19	11190412	11254341	12	63929	DEL	ENSG00000256274	11229368	11231770	TAS2R64P	unclear
chr12	20	43016464	43030165	8	13701	DEL	ENSG00000257510	43009305	43040300	ENSG00000257510	linc/transcr.
chr12	20	43016464	43030165	8	13701	DEL	ENSG00000257687	43020810	43020939	ENSG00000257687	pseudogene
chr12	21	45899907	45914382	12	14475	DEL	ENSG00000257657	45912101	46004403	ENSG00000257657	intronic
chr12	22	99792360	99808326	8	14466	DEL	ENSG00000185046	99792360	99808326	ANKS1B	intronic
chr13	23	51064709	51076629	10	11920	DEL	ENSG00000176124	50656307	51297372	LEU11	intronic
chr13	24	114323378	114427858	21	104480	DEL	ENSG00000185974	114321594	114438637	GRK1	intronic
chr15	25	29157401	29210439	13	53038	DEL	ENSG00000034053	29129629	29410518	APBA2	intronic
chr15	26	83547144	83562742	20	15698	DEL	ENSG00000103942	83509838	83654661	HOMER2	intronic
chr16	27	18828787	18843173	10	14386	DEL	ENSG00000157106	18816175	18937776	SHG1	intronic
chr16	28	75258269	75281293	35	25427	INV	ENSG00000240338	75259972	75262828	ENSG00000240338	linc/transcr.
chr16	29	83666054	83681468	15	15414	DEL	ENSG00000140945	82660408	83830204	CDH13	intronic
chr17	30	21208079	21225633	20	17554	DEL	ENSG00000034152	21187984	21218552	MAD2K3	intronic
chr17	31	39199154	39216570	9	17416	DEL	ENSG00000212725	39202793	39203568	KRTAP2-1	intergenic
chr17	31	39199154	39216570	9	17416	DEL	ENSG00000214518	39210750	39211482	KRTAP2-2	intergenic
chr17	31	39199154	39216570	9	17416	DEL	ENSG00000212724	39215495	39216344	KRTAP2-3	intergenic
chr17	32	41136326	41408077	35	27452	DEL	ENSG00000238266	41369826	41389898	ENSG00000238266	intronic
chr17	33	41400914	41466407	19	65493	INV	ENSG00000188825	41447213	41468567	ENSG00000188825	linc/transcr.
chr19	34	19829848	19843750	9	13902	DEL	ENSG00000105708	19821280	19843906	ZNF14	intronic
chr19	35	35847100	35867440	15	20340	DEL	ENSG00000185897	35849362	35851387	FFAR3	intergenic
chr19	35	35847100	35867440	15	20340	DEL	ENSG00000126251	35861831	35863855	GPRA2	intergenic
chr19	36	39259961	39284385	11	24424	INV	ENSG00000182472	39220827	39260544	CAPN12	intronic
chr19	37	52129340	52152491	28	24951	DEL	ENSG00000243469	52152115	52152863	RPLP5	pseudogene
chr19	37	52129340	52152491	28	24951	DEL	ENSG00000254415	52145806	52150054	SIGLEC14	intronic
chr19	37	52129340	52152491	28	24951	DEL	ENSG00000105501	52114781	52150151	SIGLEC5	intronic
chr19	38	54796983	54810383	9	13400	DEL	ENSG00000240197	54797902	54798219	ENSG00000240197	pseudogene
chr19	38	54796983	54810383	9	13400	DEL	ENSG00000251431	54808714	54809423	ENSG00000251431	pseudogene
chr19	38	54796983	54810383	9	13400	DEL	ENSG00000170866	54799854	54809952	LILRA3	coding hit
chr2	39	22346880	16219530	21	139849150	INV	ENSG00000231200	22156208	22753977	ENSG00000231200	coding hit
chr2	39	22346880	16219530	21	139849150	INV	ENSG00000115233	16219530	16228328	ENSG00000115233	coding hit
chr2	40	48847148	48862880	11	15732	DEL	ENSG00000239995	48859995	48860492	ENSG00000239995	pseudogene
chr2	40	48847148	48862880	11	15732	DEL	ENSG00000242441	48844937	48860287	TGF2A1L	intronic
chr2	40	48847148	48862880	11	15732	DEL	ENSG00000138039	48859428	48862880	LHCR	intronic
chr2	40	48847148	48862880	11	15732	DEL	ENSG00000068781	48757064	49003654	STON1-GTF2A1L	intronic
chr2	41	110105277	110255199	30	150242	DEL	ENSG00000172985	109748004	110262207	SH3RF3	intronic
chr2	41	14961623	14972424	24	10776	DEL	ENSG00000168289	14962819	149683273	KIF5C	intronic
chr2	43	185675948	188166401	24	2490453	INV	ENSG00000224063	188786797	188419390	ENSG00000224063	linc/transcr.
chr2	43	185675948	188166401	24	2490453	INV	ENSG00000170396	185463093	185804219	ZNF804A	coding hit
chr2	44	242849529	243040821	13	191292	DEL	ENSG00000220804	243030784	243102304	ENSG00000220804	linc/transcr.
chr2	44	242849529	243040821	13	191292	DEL	ENSG00000226423	242967334	242968327	ENSG00000226423	intronic
chr2	44	242849529	243040821	13	191292	DEL	ENSG00000232002	242989846	243026289	ENSG00000232002	linc/transcr.
chr2	44	242849529	243040821	13	191292	DEL	ENSG00000233964	24282814	243029373	ENSG00000233964	linc/transcr.
chr2	44	242849529	243040821	13	191292	DEL	ENSG00000237940	242912834	242919427	ENSG00000237940	intronic
chr2	44	242849529	243040821	13	191292	DEL	ENSG00000261186	243029505	243030608	ENSG00000261186	linc/transcr.
chr21	45	42952285	43009493	14	57208	DEL	ENSG00000223400	42948062	42953426	ENSG00000223400	intergenic
chr22	46	24270522	24314151	17	43629	DEL	ENSG00000206090	24282185	24284985	ENSG00000206090	pseudogene
chr22	46	24270522	24314151	17	43629	DEL	ENSG00000225282	24269087	24271761	ENSG00000225282	pseudogene
chr22	46	24270522	24314151	17	43629	DEL	ENSG00000228039	24305942	24306944	ENSG00000228039	pseudogene
chr22	46	24270522	24314151	17	43629	DEL	ENSG00000231271	242921025	24292229	ENSG00000231271	pseudogene
chr22	46	24270522	24314151	17	43629	DEL	ENSG00000099977	24313554	24322660	DDT	intergenic
chr22	46	24270522	24314151	17	43629	DEL	ENSG00000099974	24309089	24314721	DDTL	intergenic
chr22	46	24270522	24314151	17	43629	DEL	ENSG00000133433	24299601	24303373	GSTT2B	intronic
chr3	47	82155221	8228706	10	13485	DEL	ENSG00000227110	7994492	8653610	LMCD1-AS1	intronic
chr3	48	20308321	20322485	10	14164	DEL					