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Supplemental Data

De Novo Mutations in *MLL* Cause Wiedemann-Steiner Syndrome

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Table S1. Summary Statistics for Exome Sequencing of Individuals WSS-1, WSS-2, WSS-3, and WSS-4—Mapping and Coverage

Sequenced Exomes	WSS-1	WSS-2	WSS-3	WSS-4
Uniquely mapped reads	149672116	123768103	111861056	65553565
Uniquely mapped to target +/- 150 bp reads	135685098	92434932	101443367	59376585
Uniquely mapped to target reads	122887592	83506832	89792243	54022042
Gencode bases with coverage >1x	99.6	99.47	99.32	99.08
Gencode bases with coverage >5x	98.18	97.79	97.52	96.18
Gencode bases with coverage >10x	96.65	95.75	95.54	93.11
Gencode bases with coverage >20x	93.95	92.19	92.14	87.87
Mean coverage	212.52	144.48	154.33	93.5

Total number of mapped reads and resulting coverage of the Gencode exome.

Table S2. Summary Statistics for Exome Sequencing—Variant Calling

	WSS-1		WSS-2		WSS-3		WSS-4	
	known	novel	known	novel	known	novel	known	novel
All variants	20211	135	24561	219	24472	229	21579	175
heterozygous	11944	134	15105	216	14964	225	12952	172
homozygous	8267	1	9456	3	9508	4	8627	3
Indels	284	8	449	12	442	19	316	11
heterozygous	122	8	226	9	229	18	151	10
homozygous	162	0	223	3	213	1	165	1
3n indels	148	3	252	8	252	15	165	7
heterozygous	81	3	163	6	161	14	96	6
homozygous	67	0	89	2	91	1	69	1
Non 3n indels	136	5	197	4	190	4	151	4
heterozygous	41	5	63	3	68	4	55	4
homozygous	95	0	134	1	122	0	96	0
SNVs	17660	111	21203	187	21100	175	18842	140
heterozygous	10550	111	13112	187	12917	173	11370	138
homozygous	7110	0	8091	0	8183	2	7472	2
Synonymous_SNVs	9124	39	11120	53	11009	61	9810	51
heterozygous	5490	39	6854	53	6754	61	5855	50
homozygous	3634	0	4266	0	4255	0	3955	1
Nonsynonymous_SNVs	8536	72	10083	134	10091	114	9032	89
heterozygous	5060	72	6258	134	6163	112	5515	88
homozygous	3476	0	3825	0	3928	2	3517	1
Splice site (10 bp)	2249	16	2871	20	2896	34	2399	24
heterozygous	1259	15	1743	20	1795	33	1419	24
homozygous	990	1	1128	0	1101	1	980	0
Transition:Transversion	3.08	2.51	2.99	2.24	3.04	2.63	3.05	3
heterozygous	3.09	2.48	2.97	2.24	3.15	2.66	2.99	2.94
homozygous	3.06	n/a	3	n/a	2.89	1	3.16	n/a

Numbers of variants of different classes identified by exome sequencing. SNV - single nucleotide variants, 3n - multiples of three nucleotides

Table S3. Numbers of Candidate Genes Highlighted by Exome Sequencing

Any x of 4 individuals	**1**	**2**	**3**	**4**
Genes with heterozygous NS/SS/I variants	11,978	8095	4503	1629
Genes with heterozygous NS/SS/I variants not in dbSNP, 1000G or control exomes	491	5	1	0

NS - nonsynonymous, SS - splice site, I - indels