

Supplemental Material for

CCND2, CTNNB1, DDX3X, GLI2, SMARCA4, MYC, MYCN, PTCH1, TP53, and MLL2 gene variants and risk of childhood medulloblastoma

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Supplementary Table 1. Genes included in the present study

Chr	Gene start ^a	Gene end ^a	Gene	Number of SNPs	
2	16080686	16087129	<i>MYCN</i>	1	Recurrently amplified in medulloblastoma tumors [1-3].
2	121493199	121750229	<i>GLI2</i>	94	Recurrently amplified in medulloblastoma tumors, particularly in the SHH subgroup [2]. Mediates SHH signaling.
3	41236328	41301587	<i>CTNNB1</i>	20	Recurrently mutated in medulloblastoma tumors, particularly in the WNT subgroup [3-6].
8	128747680	128753674	<i>MYC</i>	1	Recurrently amplified in medulloblastoma tumors [1-4].
9	98205262	98279339	<i>PTCH1</i>	22	Recurrently mutated in medulloblastoma tumors, particularly in the SHH subgroup [3-6].
12	4382938	4414516	<i>CCND2</i>	27	Recurrently amplified in medulloblastoma tumors [2]. Involved in cell cycle regulation.
12	49412758	49453557	<i>MLL2 (KMT2D)</i>	9	Recurrently mutated in medulloblastoma tumors [3-6].
17	7565097	7590856	<i>TP53</i>	9	Recurrently mutated in medulloblastoma tumors [3-6].
19	11071598	11176071	<i>SMARCA4</i>	28	Recurrently mutated in medulloblastoma tumors, particularly in the WNT subgroup [3-6].
X	41192651	41223725	<i>DDX3X</i>	10	Recurrently mutated in medulloblastoma tumors, particularly in the WNT subgroup [3, 5, 6].

^a Ensemble gene annotations (GRCh37.p8) were obtained from MartView (<http://biomart.org>)

SHH, sonic hedgehog.

Supplementary Table 2. OR and p-values for risk of medulloblastoma for all investigated SNPs

SNP (minor/major(ref) allele)	chr	bp (GRCh37/hg19)	OR _{comb} ^a	P _{comb} ^a	Gene
rs922 (A/G)	2	16086469	1.20	0.236	<i>MYCN</i>
rs67611826 (G/A)	2	121500265	1.26	0.084	<i>GLI2</i>
rs62150664 (A/G)	2	121501287	0.94	0.681	<i>GLI2</i>
rs13008945 (G/A)	2	121506802	0.70	0.033	<i>GLI2</i>
rs6757829 (G/A)	2	121509295	0.75	0.244	<i>GLI2</i>
rs776501 (A/G)	2	121509378	0.80	0.102	<i>GLI2</i>
rs776504 (A/C)	2	121513197	1.28	0.065	<i>GLI2</i>
rs11901641 (C/A)	2	121513993	0.93	0.631	<i>GLI2</i>
rs13034141 (C/A)	2	121515390	0.72	0.076	<i>GLI2</i>
rs707490 (A/G)	2	121517174	1.27	0.071	<i>GLI2</i>
rs4848627 (G/A)	2	121530262	0.75	0.078	<i>GLI2</i>
rs11677381 (A/G)	2	121530351	1.54	0.086	<i>GLI2</i>
rs2121992 (A/G)	2	121532932	0.72	0.046	<i>GLI2</i>
rs17389953 (C/A)	2	121532983	0.92	0.578	<i>GLI2</i>
rs4848628 (C/A)	2	121536569	0.68	0.039	<i>GLI2</i>
rs79503443 (A/G)	2	121537891	0.66	0.374	<i>GLI2</i>
rs2053989 (G/A)	2	121544316	1.13	0.380	<i>GLI2</i>
rs7605011 (A/G)	2	121551822	1.36	0.218	<i>GLI2</i>
rs3820723 (A/C)	2	121552558	1.32	0.258	<i>GLI2</i>
rs7569100 (G/A)	2	121554317	0.92	0.754	<i>GLI2</i>
rs36124185 (G/A)	2	121558036	0.96	0.763	<i>GLI2</i>
rs4848119 (A/G)	2	121559113	1.06	0.638	<i>GLI2</i>
rs4848632 (A/G)	2	121562595	1.06	0.649	<i>GLI2</i>
rs76201712 (A/G)	2	121563734	1.13	0.517	<i>GLI2</i>
rs55633297 (G/A)	2	121565142	1.05	0.845	<i>GLI2</i>
rs895485 (G/A)	2	121570665	1.29	0.058	<i>GLI2</i>
rs4848636 (A/G)	2	121574575	0.61	0.349	<i>GLI2</i>
rs2871627 (A/G)	2	121578394	0.61	0.354	<i>GLI2</i>
rs895491 (G/A)	2	121578827	1.25	0.093	<i>GLI2</i>
rs11122821 (G/A)	2	121579810	0.74	0.018	<i>GLI2</i>
rs11122822 (A/G)	2	121579866	1.21	0.139	<i>GLI2</i>
rs7561607 (A/G)	2	121580431	1.23	0.104	<i>GLI2</i>
rs116029714 (G/A)	2	121581093	1.24	0.755	<i>GLI2</i>
rs2310896 (A/G)	2	121581416	1.08	0.698	<i>GLI2</i>
rs58292323 (G/A)	2	121581586	0.65	0.161	<i>GLI2</i>
rs4848640 (A/G)	2	121585366	0.74	0.451	<i>GLI2</i>
rs1992900 (A/G)	2	121588532	1.32	0.049	<i>GLI2</i>
rs895479 (G/A)	2	121589440	1.15	0.443	<i>GLI2</i>
rs13393992 (A/G)	2	121591328	1.30	0.144	<i>GLI2</i>

rs11122826 (A/C)	2	121591950	1.20	0.474	GLI2
rs17005273 (A/G)	2	121592523	0.98	0.920	GLI2
rs11885290 (G/A)	2	121592733	0.98	0.876	GLI2
rs7559777 (A/C)	2	121595616	1.05	0.718	GLI2
rs74641446 (G/A)	2	121595667	0.98	0.943	GLI2
rs72971958 (G/A)	2	121598502	1.04	0.846	GLI2
rs7557372 (A/C)	2	121598768	0.91	0.575	GLI2
rs17005295 (G/A)	2	121600223	1.01	0.941	GLI2
rs4848641 (A/G)	2	121603584	0.97	0.830	GLI2
rs55917638 (A/G)	2	121606405	0.70	0.470	GLI2
rs67124142 (G/A)	2	121607893	1.04	0.743	GLI2
rs2166898 (A/G)	2	121612659	0.76	0.097	GLI2
rs55706422 (G/A)	2	121613509	1.03	0.807	GLI2
rs4848643 (A/G)	2	121618625	1.14	0.334	GLI2
rs4143116 (A/G)	2	121620828	1.03	0.839	GLI2
rs4848126 (A/G)	2	121627175	1.10	0.462	GLI2
rs277554 (G/A)	2	121631384	1.15	0.283	GLI2
rs277552 (A/G)	2	121632454	1.16	0.299	GLI2
rs55751863 (A/G)	2	121632865	1.02	0.928	GLI2
rs62150745 (A/G)	2	121633571	0.84	0.600	GLI2
rs277550 (A/G)	2	121634010	1.18	0.222	GLI2
rs72835595 (C/A)	2	121640679	1.24	0.625	GLI2
rs6541743 (A/G)	2	121647802	1.15	0.292	GLI2
rs12478509 (G/A)	2	121648806	0.88	0.527	GLI2
rs10199310 (A/G)	2	121649279	0.93	0.706	GLI2
rs79465951 (A/G)	2	121651189	1.19	0.409	GLI2
rs277536 (A/G)	2	121652747	1.06	0.647	GLI2
rs277539 (G/A)	2	121653647	1.05	0.703	GLI2
rs776189 (A/G)	2	121655419	1.54	0.096	GLI2
rs277541 (A/G)	2	121656429	1.23	0.530	GLI2
rs10173670 (C/A)	2	121657322	2.01	0.088	GLI2
rs11675340 (A/G)	2	121665196	0.97	0.833	GLI2
rs1187934 (A/G)	2	121667962	1.23	0.530	GLI2
rs1992902 (G/A)	2	121670602	1.04	0.862	GLI2
rs1992901 (A/G)	2	121670669	0.87	0.283	GLI2
rs277524 (A/G)	2	121670757	0.83	0.154	GLI2
rs10206938 (G/A)	2	121675238	1.03	0.934	GLI2
rs895549 (A/G)	2	121676484	1.17	0.582	GLI2
rs895547 (A/G)	2	121676655	1.18	0.573	GLI2
rs72955383 (A/G)	2	121681721	1.69	0.292	GLI2
rs114247324 (G/A)	2	121682216	0.78	0.634	GLI2
rs6750250 (G/A)	2	121685681	0.93	0.835	GLI2
rs4848648 (G/A)	2	121686439	1.25	0.578	GLI2

rs4848649 (A/G)	2	121686533	1.02	0.970	<i>GLI2</i>
rs6541745 (A/G)	2	121687129	1.18	0.564	<i>GLI2</i>
rs10167579 (A/G)	2	121699964	1.21	0.597	<i>GLI2</i>
rs12711535 (G/A)	2	121709536	1.25	0.461	<i>GLI2</i>
rs895545 (G/A)	2	121721467	1.21	0.543	<i>GLI2</i>
rs13396540 (C/A)	2	121726151	1.03	0.930	<i>GLI2</i>
rs280199 (A/G)	2	121733476	0.89	0.419	<i>GLI2</i>
rs3768696 (G/A)	2	121733538	0.91	0.495	<i>GLI2</i>
rs7588213 (C/A)	2	121735335	0.93	0.618	<i>GLI2</i>
rs280194 (G/A)	2	121738820	0.94	0.642	<i>GLI2</i>
rs7577521 (A/G)	2	121739875	0.95	0.703	<i>GLI2</i>
rs280198 (A/G)	2	121741762	0.92	0.537	<i>GLI2</i>
rs116677865 (G/A)	2	121743169	1.16	0.777	<i>GLI2</i>
rs11564430 (A/G)	3	41239693	1.13	0.812	<i>CTNNB1</i>
rs3915129 (C/A)	3	41243742	0.94	0.612	<i>CTNNB1</i>
rs115813431 (C/A)	3	41244376	0.62	0.249	<i>CTNNB1</i>
rs12492719 (C/A)	3	41245731	1.06	0.851	<i>CTNNB1</i>
rs73081753 (G/A)	3	41245804	1.02	0.964	<i>CTNNB1</i>
rs13075993 (A/G)	3	41260017	0.93	0.562	<i>CTNNB1</i>
rs11564445 (A/G)	3	41267752	0.76	0.393	<i>CTNNB1</i>
rs11564447 (C/A)	3	41268354	0.95	0.892	<i>CTNNB1</i>
rs4135385 (G/A)	3	41279440	0.94	0.696	<i>CTNNB1</i>
rs2953 (C/A)	3	41281388	0.92	0.530	<i>CTNNB1</i>
rs113152028 (G/A)	3	41283882	1.01	0.982	<i>CTNNB1</i>
rs9883073 (A/C)	3	41284536	0.93	0.571	<i>CTNNB1</i>
rs79668667 (A/G)	3	41284947	1.02	0.975	<i>CTNNB1</i>
rs11711946 (G/A)	3	41287018	0.91	0.456	<i>CTNNB1</i>
rs6803275 (G/A)	3	41287455	1.06	0.851	<i>CTNNB1</i>
rs9850653 (A/G)	3	41292736	0.99	0.977	<i>CTNNB1</i>
rs11129896 (G/A)	3	41295767	1.01	0.909	<i>CTNNB1</i>
rs9849022 (C/A)	3	41297523	0.99	0.935	<i>CTNNB1</i>
rs73081797 (A/G)	3	41299516	1.02	0.892	<i>CTNNB1</i>
rs2293300 (C/A)	3	41301534	0.99	0.967	<i>CTNNB1</i>
rs10110283 (A/G)	8	128752178	0.75	0.434	<i>MYC</i>
rs28485160 (G/A)	9	98205645	0.80	0.332	<i>PTCH1</i>
rs16909865 (G/C)	9	98207302	0.78	0.284	<i>PTCH1</i>
rs2134721 (A/G)	9	98215306	0.94	0.692	<i>PTCH1</i>
rs28457693 (G/A)	9	98217348	0.98	0.917	<i>PTCH1</i>
rs2274693 (A/G)	9	98229297	0.96	0.811	<i>PTCH1</i>
rs16909898 (G/A)	9	98231008	0.90	0.612	<i>PTCH1</i>
rs2066836 (A/G)	9	98238358	0.98	0.882	<i>PTCH1</i>
rs2277183 (G/A)	9	98238860	1.54	0.419	<i>PTCH1</i>
rs574688 (G/C)	9	98239190	1.08	0.583	<i>PTCH1</i>

rs2297086 (A/G)	9	98240120	1.00	0.981	<i>PTCH1</i>
rs2297087 (G/A)	9	98242925	0.94	0.700	<i>PTCH1</i>
rs17369383 (G/A)	9	98245974	0.94	0.700	<i>PTCH1</i>
rs2282041 (G/A)	9	98248387	1.04	0.849	<i>PTCH1</i>
rs78400819 (A/G)	9	98248780	0.49	0.113	<i>PTCH1</i>
rs77224875 (G/A)	9	98254218	0.45	0.008	<i>PTCH1</i>
rs473902 (C/A)	9	98256235	0.87	0.511	<i>PTCH1</i>
rs473902 (C/A)	9	98256235	0.87	0.524	<i>PTCH1</i>
rs10512248 (C/A)	9	98259703	1.04	0.800	<i>PTCH1</i>
rs60417486 (A/G)	9	98262178	0.97	0.904	<i>PTCH1</i>
rs16909922 (G/A)	9	98265901	0.97	0.904	<i>PTCH1</i>
rs34241213 (A/G)	9	98272006	1.68	0.151	<i>PTCH1</i>
rs74499984 (C/A)	9	98272491	1.42	0.465	<i>PTCH1</i>
rs3217795 (G/A)	12	4386064	1.12	0.660	<i>CCND2</i>
rs3217805 (G/C)	12	4388084	0.67	0.003	<i>CCND2</i>
rs4372527 (G/A)	12	4388914	1.38	0.024	<i>CCND2</i>
rs11063072 (G/A)	12	4389158	0.87	0.625	<i>CCND2</i>
rs3217818 (G/A)	12	4390326	0.99	0.979	<i>CCND2</i>
rs3217830 (A/G)	12	4392530	1.29	0.072	<i>CCND2</i>
rs3217833 (A/G)	12	4393474	1.10	0.591	<i>CCND2</i>
rs3217840 (G/A)	12	4394877	1.29	0.067	<i>CCND2</i>
rs3217863 (A/G)	12	4399496	0.66	0.055	<i>CCND2</i>
rs3217867 (G/A)	12	4399917	0.91	0.529	<i>CCND2</i>
rs3217871 (C/A)	12	4400569	0.66	0.290	<i>CCND2</i>
rs3217874 (A/G)	12	4400808	1.05	0.723	<i>CCND2</i>
rs3217875 (G/A)	12	4401201	0.81	0.550	<i>CCND2</i>
rs3217882 (A/G)	12	4402817	0.97	0.840	<i>CCND2</i>
rs3217889 (A/G)	12	4403771	0.91	0.850	<i>CCND2</i>
rs3217898 (G/A)	12	4404376	1.23	0.132	<i>CCND2</i>
rs3217901 (G/A)	12	4405389	1.04	0.775	<i>CCND2</i>
rs3217905 (G/A)	12	4405686	0.93	0.590	<i>CCND2</i>
rs3217906 (G/A)	12	4405804	1.00	0.996	<i>CCND2</i>
rs10849028 (C/A)	12	4405850	0.82	0.603	<i>CCND2</i>
rs12299509 (G/A)	12	4406281	0.89	0.376	<i>CCND2</i>
rs3217907 (A/C)	12	4406836	0.78	0.055	<i>CCND2</i>
rs3217916 (G/A)	12	4408673	1.16	0.319	<i>CCND2</i>
rs3217925 (A/G)	12	4411639	1.07	0.659	<i>CCND2</i>
rs3217926 (G/A)	12	4411683	1.22	0.115	<i>CCND2</i>
rs1049612 (G/A)	12	4412762	1.23	0.107	<i>CCND2</i>
rs3217933 (G/A)	12	4413000	0.77	0.075	<i>CCND2</i>
rs7975791 (A/G)	12	49413486	1.02	0.953	<i>MLL2 (KMT2D)</i>
rs10875912 (G/A)	12	49416944	1.13	0.380	<i>MLL2 (KMT2D)</i>
rs55776396 (C/A)	12	49418435	1.15	0.688	<i>MLL2 (KMT2D)</i>

rs117856947 (C/A)	12	49422033	0.66	0.287	<i>MLL2 (KMT2D)</i>
rs10875914 (G/A)	12	49422094	1.11	0.421	<i>MLL2 (KMT2D)</i>
rs80132640 (G/A)	12	49426460	1.02	0.954	<i>MLL2 (KMT2D)</i>
rs10875915 (A/G)	12	49430094	1.15	0.309	<i>MLL2 (KMT2D)</i>
rs2304275 (G/A)	12	49442813	1.14	0.372	<i>MLL2 (KMT2D)</i>
rs833836 (A/G)	12	49451459	0.96	0.794	<i>MLL2 (KMT2D)</i>
rs118051719 (A/G)	17	7566428	1.52	0.460	<i>TP53</i>
rs9893249 (G/A)	17	7570189	0.86	0.261	<i>TP53</i>
rs9914052 (G/C)	17	7570823	0.81	0.546	<i>TP53</i>
rs17881556 (G/A)	17	7570869	0.97	0.926	<i>TP53</i>
rs858528 (A/G)	17	7574936	1.37	0.112	<i>TP53</i>
rs1625895 (A/G)	17	7578115	1.29	0.204	<i>TP53</i>
rs2909430 (G/A)	17	7578645	1.34	0.135	<i>TP53</i>
rs11652704 (G/A)	17	7584400	0.87	0.536	<i>TP53</i>
rs17881035 (G/A)	17	7587899	0.73	0.262	<i>TP53</i>
rs11879293 (A/G)	19	11072610	1.00	0.995	<i>SMARCA4</i>
rs2163800 (G/A)	19	11075706	1.04	0.786	<i>SMARCA4</i>
rs73011369 (A/G)	19	11077662	1.08	0.773	<i>SMARCA4</i>
rs12610374 (A/G)	19	11080921	0.94	0.681	<i>SMARCA4</i>
rs4804553 (G/A)	19	11081405	0.94	0.681	<i>SMARCA4</i>
rs4804554 (A/C)	19	11083210	0.98	0.910	<i>SMARCA4</i>
rs17001070 (G/A)	19	11086746	1.08	0.826	<i>SMARCA4</i>
rs12459603 (A/G)	19	11089846	1.36	0.371	<i>SMARCA4</i>
rs11672595 (A/G)	19	11094494	0.72	0.382	<i>SMARCA4</i>
rs76614166 (G/A)	19	11104243	0.59	0.251	<i>SMARCA4</i>
rs17001075 (G/A)	19	11105593	1.13	0.713	<i>SMARCA4</i>
rs7935 (G/A)	19	11105608	0.98	0.891	<i>SMARCA4</i>
rs57900086 (G/A)	19	11109468	0.97	0.921	<i>SMARCA4</i>
rs17001076 (A/G)	19	11113520	0.98	0.933	<i>SMARCA4</i>
rs17001078 (G/A)	19	11113651	1.00	0.985	<i>SMARCA4</i>
rs12983316 (G/A)	19	11114352	1.05	0.778	<i>SMARCA4</i>
rs13345127 (A/G)	19	11116266	0.95	0.788	<i>SMARCA4</i>
rs73013166 (G/A)	19	11126160	1.06	0.840	<i>SMARCA4</i>
rs73013169 (A/C)	19	11127069	0.98	0.901	<i>SMARCA4</i>
rs17001095 (G/A)	19	11133272	1.00	0.995	<i>SMARCA4</i>
rs56167249 (G/A)	19	11157423	1.05	0.808	<i>SMARCA4</i>
rs77287598 (A/G)	19	11159449	0.69	0.234	<i>SMARCA4</i>
rs6511719 (G/A)	19	11164790	1.06	0.838	<i>SMARCA4</i>
rs3786724 (G/A)	19	11166791	1.00	0.998	<i>SMARCA4</i>
rs3786725 (A/G)	19	11166827	1.00	0.991	<i>SMARCA4</i>
rs7258189 (G/A)	19	11169947	0.94	0.646	<i>SMARCA4</i>
rs8099996 (G/A)	19	11174625	0.94	0.653	<i>SMARCA4</i>
rs73013198 (A/G)	19	11174742	1.01	0.954	<i>SMARCA4</i>

rs4358953 (A/G)	X	41196417	0.88	0.374	<i>DDX3X</i>
rs11542970 (G/A)	X	41197200	1.00	0.997	<i>DDX3X</i>
rs953114 (C/A)	X	41197542	0.87	0.339	<i>DDX3X</i>
rs2275943 (G/A)	X	41198456	0.94	0.753	<i>DDX3X</i>
rs6610545 (G/A)	X	41198715	0.88	0.379	<i>DDX3X</i>
rs12851342 (C/A)	X	41199626	1.02	0.936	<i>DDX3X</i>
rs4827268 (A/G)	X	41199812	0.66	0.204	<i>DDX3X</i>
rs10521420 (A/G)	X	41202159	0.65	0.198	<i>DDX3X</i>
rs72626410 (A/G)	X	41210256	0.66	0.204	<i>DDX3X</i>
rs7877664 (A/G)	X	41210942	1.02	0.910	<i>DDX3X</i>

^a Fixed-effect model meta-analysis combining the results from the Swedish and the Danish datasets

Supplementary Table 3. OR and p-values for eight genetic variants (in *GLI2*, *PTCH1* and *CCND2*), calculated in the Swedish and Danish datasets separately.

SNP (minor/major(ref) allele)	Gene	OR_{Sv}^a (95% CI)	P_{Sv}^a	OR_{Dk}^b (95% CI)	P_{Dk}^b
rs13008945 (G/A)	<i>GLI2</i>	0.57 (0.35-0.93)	0.024	0.83 (0.53-1.30)	0.411
rs2121992 (A/G)	<i>GLI2</i>	0.51 (0.31-0.83)	0.007	0.94 (0.61-1.45)	0.778
rs4848628 (C/A)	<i>GLI2</i>	0.48 (0.27-0.84)	0.011	0.87 (0.55-1.40)	0.577
rs11122821 (G/A)	<i>GLI2</i>	0.68 (0.48-0.97)	0.032	0.80 (0.56-1.15)	0.232
rs1992900 (A/G)	<i>GLI2</i>	1.39 (0.93-2.07)	0.104	1.26 (0.85-1.86)	0.247
rs77224875 (G/A)	<i>PTCH1</i>	0.37 (0.16-0.87)	0.023	0.54 (0.25-1.19)	0.128
rs3217805 (G/C)	<i>CCND2</i>	0.69 (0.47-1.01)	0.058	0.65 (0.45-0.93)	0.018
rs4372527 (G/A)	<i>CCND2</i>	1.17 (0.80-1.73)	0.421	1.64 (1.10-2.45)	0.016

^a OR, 95% CI, and p-value calculated using unconditional logistic regression in the Swedish dataset.

^b OR, 95% CI, and p-value calculated using unconditional logistic regression in the Danish dataset.

Supplementary References

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