SUPPLEMENTAL MATERIAL

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Supplemental Data Description

Supplemental Table 1

Summary data for significant associations between inherited variants and CTDs among the CHOP CTD case-control analysis ($p \leq 5x10^{-8}$)

Supplemental Table 2 (separate Excel file)

Summary data for suggestive associations between maternal variants and CTDs+LVOTDs from the meta-analysis ($p \leq 10^{-5}$)

Supplemental Table 3 (separate Excel file)

Summary data for suggestive associations between maternal variants and CTDs from the meta-analysis ($p \leq 10^{-5}$)

Supplemental Table 4 (separate Excel file)

Summary data for suggestive associations between maternal variants and LVOTDs from the meta-analysis ($p \leq 10^{-5}$)

Supplemental Table 5 (separate Excel file)

Summary data for suggestive associations between inherited variants and CTDs+LVOTDs from the meta-analysis ($p \leq 10^{-5}$)

Supplemental Table 6 (separate Excel file)

Summary data for suggestive associations between inherited variants and CTDs from the meta-analysis ($p \leq 10^{-5}$)

Supplemental Table 7 (separate Excel file)

Summary data for suggestive associations between inherited variants and LVOTDs from the meta-analysis ($p \leq 10^{-5}$)

Supplemental Table 8 (separate Excel file)

Our meta-analysis results for variants reported in previous GWAS of heart defects

Supplemental Figure 1

Quantile-quantile plots for the meta-analysis of: A) inherited variants and CTDs+LVOTDs, B) inherited variants and any CTDs, C) inherited variants and LVOTDs, D) maternal variants and CTDs+LVOTDs, E) maternal variants and any CTDs, F) maternal variants and LVOTDs

Supplemental Figure 2

Manhattan plot for the meta-analysis of: A) inherited variants and CTDs+LVOTDs, B) inherited variants and any CTDs, C) inherited variants and LVOTDs, D) maternal variants and CTDs+LVOTDs, E) maternal variants and any CTDs, F) maternal variants and LVOTDs

Supplemental Table 1. Results for inherited variants associated with heart defects ($p<5x10^{-8}$) in the CHOP CTD case-control analysis[A]

SNP	Chr[B]	Position[C] (bp)	Gene	Function	P-value	Odds ratio (95% CI)[D]	CADD[E]	GWAVA-TSS score
rs2765283	1	118536418	*SPAG17*	Intron	8.25E-09	0.53 (0.42-0.66)	5.45	0.34
rs11816696	10	25362890	Intergenic	-	5.44E-09	1.55 (1.33-1.80)	3.67	0.32
rs1689340	12	86574519	*MGAT4C*	Intron	2.02E-10	1.78 (1.49-2.14)	3.09	0.28
rs1689357	12	86618937	*MGAT4C*	Intron	4.54E-09	1.70 (1.42-2.04)	2.04	0.30
rs1698787	12	86618008	*MGAT4C*	Intron	5.38E-09	1.69 (1.41-2.03)	1.95	0.32
rs2452814	12	86586255	*MGAT4C*	Intron	1.04E-10	1.79 (1.50-2.15)	0.57	0.23
rs6538022	12	86584173	*MGAT4C*	Intron	1.98E-10	1.78 (1.48-2.14)	0.73	0.36
rs6538023	12	86585133	*MGAT4C*	Intron	6.57E-11	2.00 (1.61-2.48)	0.60	0.25
rs6538024	12	86585136	*MGAT4C*	Intron	3.21E-11	1.95 (1.59-2.38)	0.57	0.21
rs6538025	12	86600196	*MGAT4C*	Intron	1.45E-09	1.72 (1.44-2.06)	1.44	0.35
rs6538026	12	86600321	*MGAT4C*	Intron	2.94E-10	1.77 (1.47-2.12)	1.90	0.26
rs709812	12	86608050	*MGAT4C*	Intron	2.03E-09	1.72 (1.43-2.06)	1.79	0.31
rs7956812	12	86592670	*MGAT4C*	Intron	5.19E-14	1.94 (1.62-2.31)	2.22	0.30
rs839101	12	86574359	*MGAT4C*	Intron	2.38E-10	1.78 (1.48-2.14)	0.99	0.24
rs839102	12	86578110	*MGAT4C*	Intron	1.4E-10	1.79 (1.49-2.15)	2.76	0.35
rs839103	12	86582700	*MGAT4C*	Intron	3.5E-10	1.77 (1.47-2.13)	1.27	0.24
rs839104	12	86608721	*MGAT4C*	Intron	3.25E-09	1.71 (1.43-2.05)	4.08	0.24
rs839105	12	86609200	*MGAT4C*	Intron	1.36E-08	1.67 (1.39-2.00)	4.95	0.24
rs839108	12	86569704	*MGAT4C*	Intron	1.79E-08	1.65 (1.38-1.97)	0.02	0.29
rs839148	12	86598998	*MGAT4C*	Intron	1.65E-11	1.82 (1.52-2.18)	2.54	0.23
rs839149	12	86598656	*MGAT4C*	Intron	1.38E-12	1.88 (1.57-2.26)	1.96	0.31
rs839150	12	86598519	*MGAT4C*	Intron	4.55E-12	1.85 (1.55-2.21)	0.57	0.29
rs839151	12	86597014	*MGAT4C*	Intron	2.89E-14	1.95 (1.63-2.33)	1.49	0.27
rs839152	12	86597000	*MGAT4C*	Intron	1.02E-15	2.15 (1.77-2.61)	0.54	0.28
rs839153	12	86596980	*MGAT4C*	Intron	2.89E-14	1.95 (1.63-2.33)	2.46	0.31
rs839154	12	86596330	*MGAT4C*	Intron	2.89E-14	1.95 (1.63-2.33)	5.99	0.28
rs839155	12	86595986	*MGAT4C*	Intron	2.55E-13	1.90 (1.59-2.27)	0.88	0.21
rs839156	12	86595690	*MGAT4C*	Intron	1.67E-15	2.14 (1.76-2.60)	0.21	0.26
rs839157	12	86595546	*MGAT4C*	Intron	5.47E-14	1.93 (1.62-2.31)	0.63	0.23
rs839158	12	86595472	*MGAT4C*	Intron	5.47E-14	1.93 (1.62-2.31)	1.16	0.22
rs839159	12	86593708	*MGAT4C*	Intron	5.26E-14	1.94 (1.62-2.31)	4.74	0.24
rs839160	12	86593387	*MGAT4C*	Intron	5.26E-14	1.94 (1.62-2.31)	3.42	0.21
rs839162	12	86590307	*MGAT4C*	Intron	2.84E-12	2.05 (1.66-2.52)	1.33	0.29
rs839163	12	86588732	*MGAT4C*	Intron	3.61E-14	1.96 (1.63-2.34)	11.54	0.37
rs839164	12	86587630	*MGAT4C*	Intron	2.42E-13	1.92 (1.60-2.30)	5.32	0.32
rs839165	12	86586654	*MGAT4C*	Intron	1.04E-10	1.79 (1.50-2.15)	9.61	0.30

rs839166	12	86586394	*MGAT4C*	Intron	4E-11	1.94 (1.59-2.37)	7.67	0.25
rs839168	12	86603948	*MGAT4C*	Intron	2.36E-09	1.72 (1.43-2.06)	0.22	0.36
rs839170	12	86600854	*MGAT4C*	Intron	3.05E-10	1.76 (1.47-2.12)	3.33	0.26
rs839171	12	86600654	*MGAT4C*	Intron	4.08E-10	1.76 (1.47-2.12)	1.41	0.22
rs839172	12	86599496	*MGAT4C*	Intron	2.28E-11	1.82 (1.52-2.18)	0.75	0.24
rs844432	12	86595009	*MGAT4C*	Intron	5.52E-14	1.93 (1.62-2.31)	0.80	0.29
rs844434	12	86601871	*MGAT4C*	Intron	3.9E-10	1.86 (1.52-2.27)	0.23	0.40
rs844435	12	86599670	*MGAT4C*	Intron	2.28E-11	1.82 (1.52-2.18)	0.72	0.23
rs844436	12	86599573	*MGAT4C*	Intron	2.28E-11	1.82 (1.52-2.18)	1.23	0.24
rs860813	12	86599579	*MGAT4C*	Intron	2.28E-11	1.82 (1.52-2.18)	0.76	0.26
rs863392	12	86580946	*MGAT4C*	Intron	1.99E-10	1.78 (1.48-2.14)	12.10	0.33
rs863394	12	86594783	*MGAT4C*	Intron	1.71E-15	2.14 (1.76-2.60)	4.80	0.19
rs863395	12	86593101	*MGAT4C*	Intron	5.26E-14	1.94 (1.62-2.31)	4.83	0.25
rs865138	12	86583292	*MGAT4C*	Intron	1.85E-10	1.78 (1.49-2.14)	0.16	0.42
rs865721	12	86589761	*MGAT4C*	Intron	1.67E-15	2.16 (1.77-2.63)	0.48	0.32
rs2406176	21	19719426	*PRSS7*	Intron	6.99E-09	1.60 (1.36-1.88)	6.49	0.32

[A] None of the eight other GWAS of maternal or inherited SNPs identified GWAS-significant associations ($p<5x10^{-8}$) with heart defects

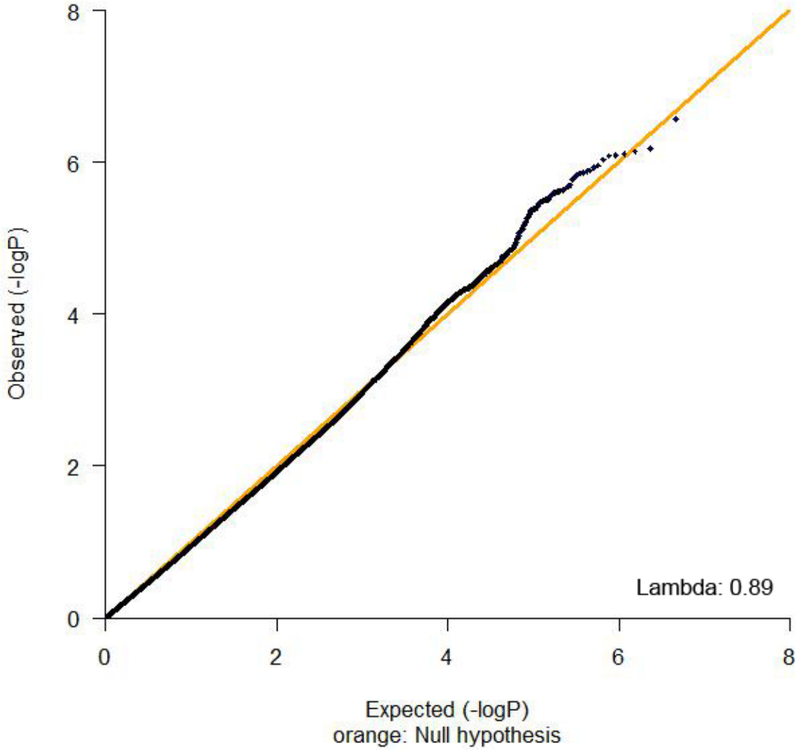
[B] Chromosome

[C] Hg19/NCBI build 37

[D] Estimate for carrying one copy of the high-risk allele compared to no copies, and corresponding 95% confidence interval

[E] Scaled CADD score

A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Expected (-logP)
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Lambda: 0.89

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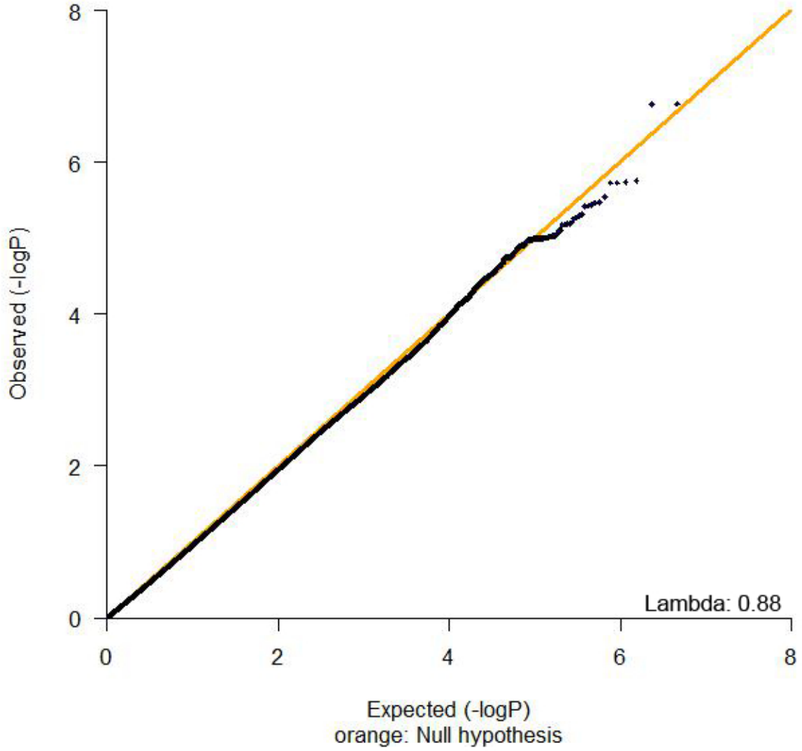
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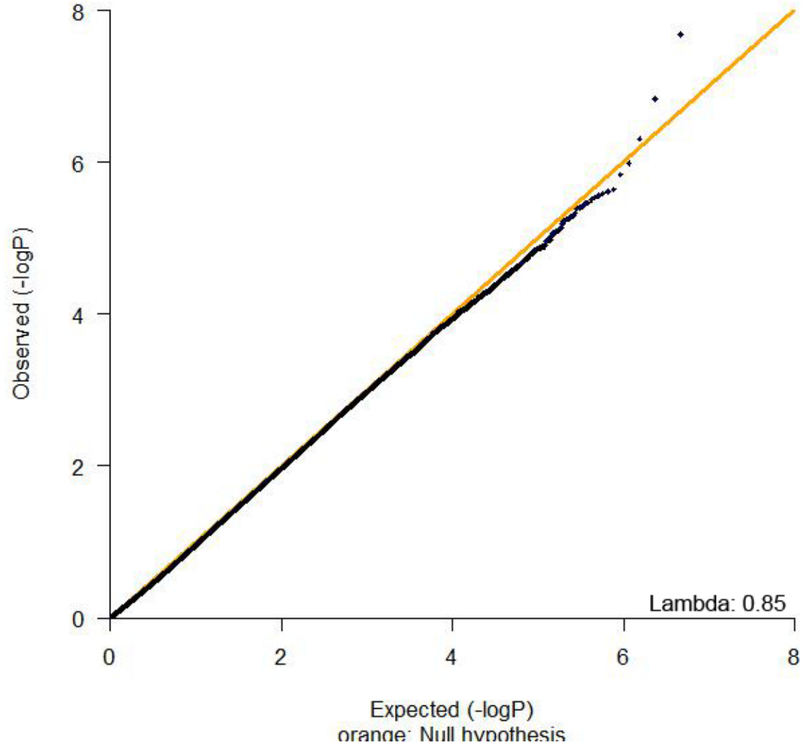
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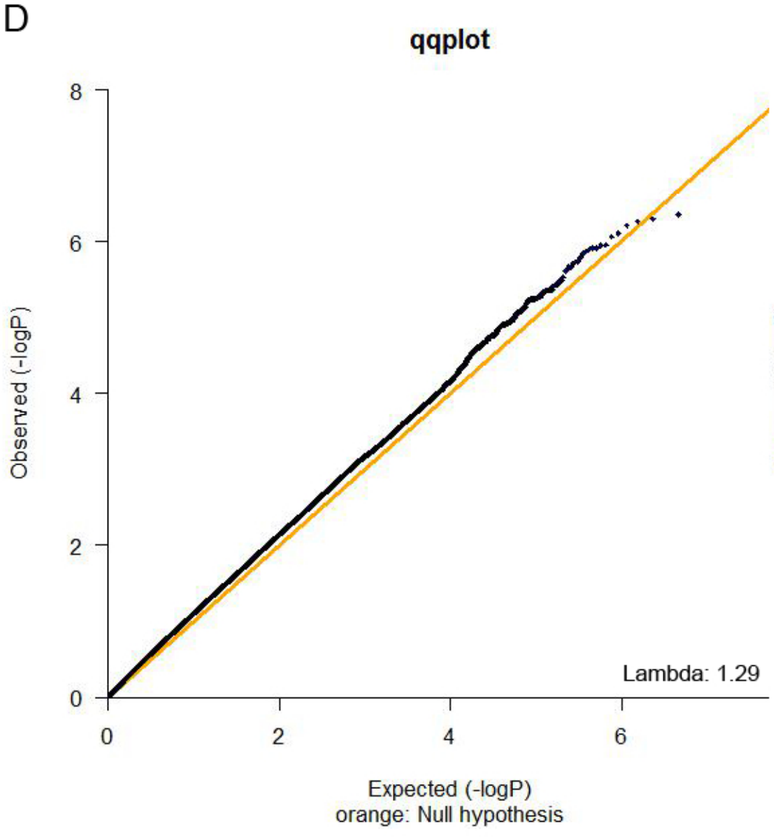
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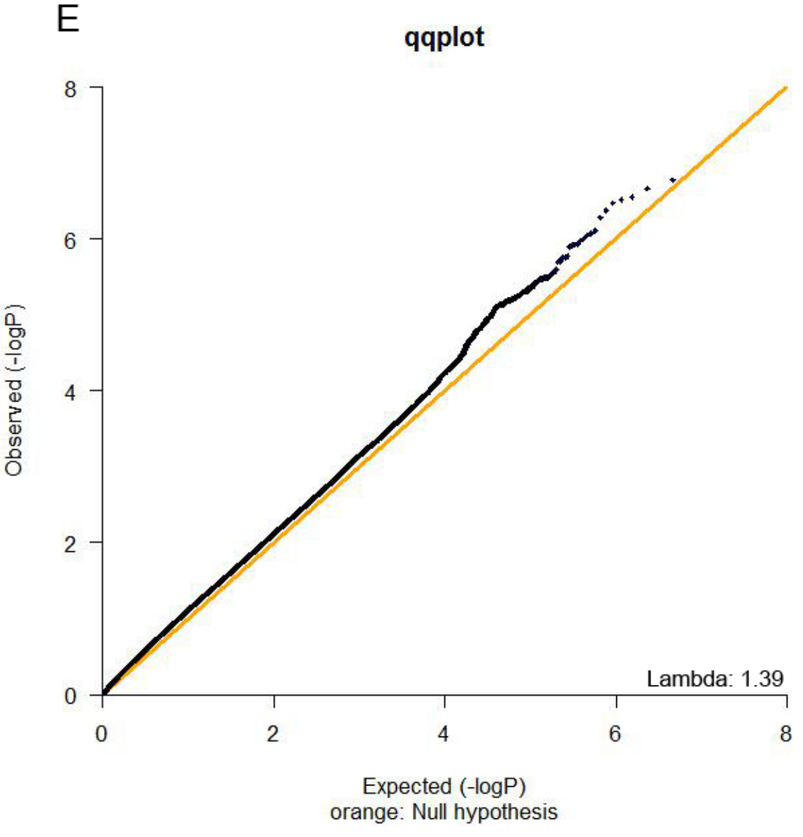
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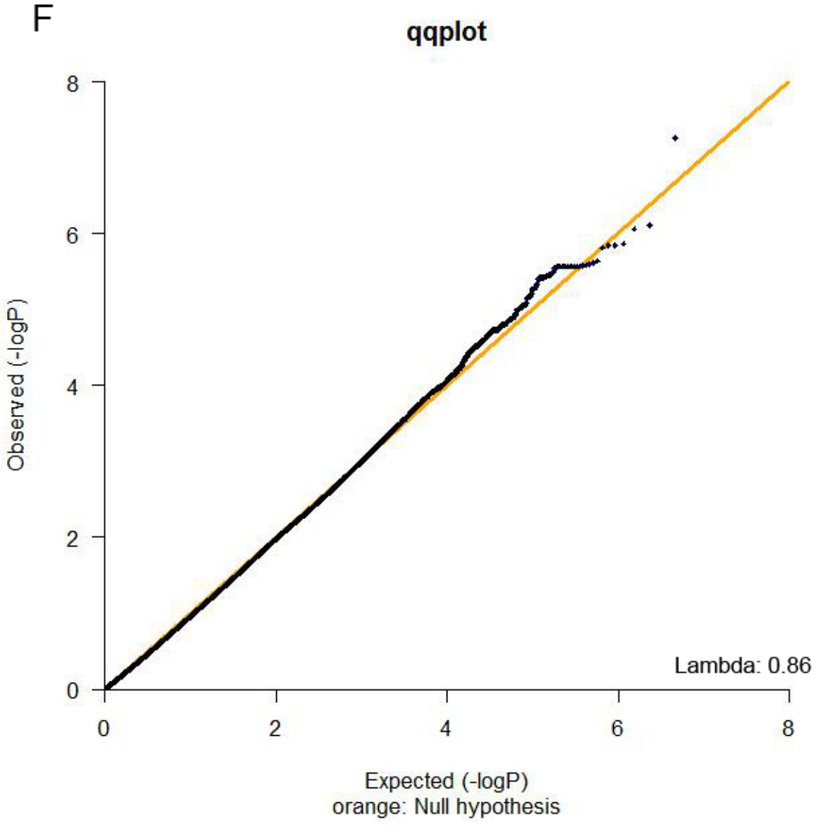
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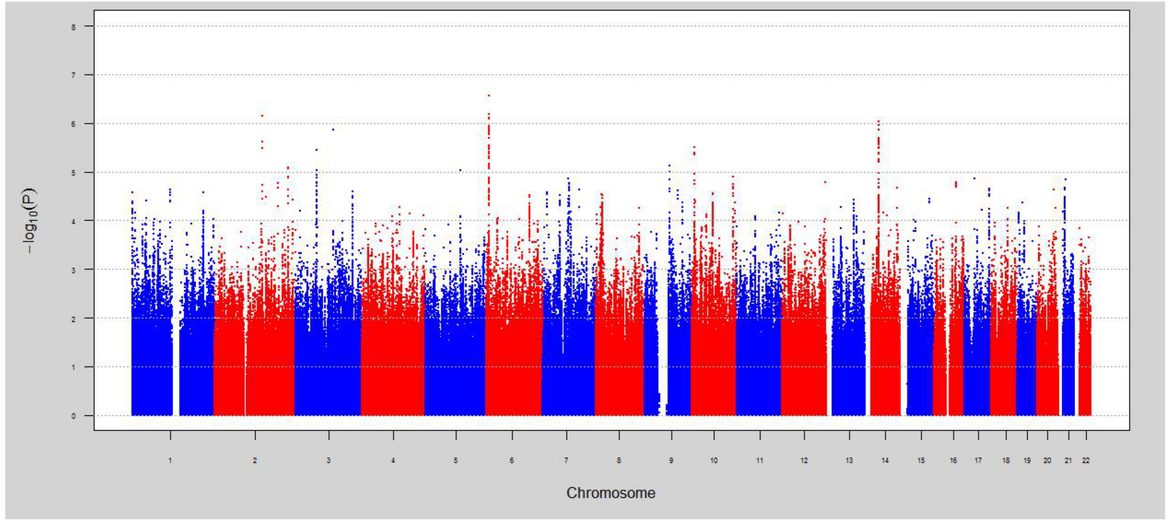
A)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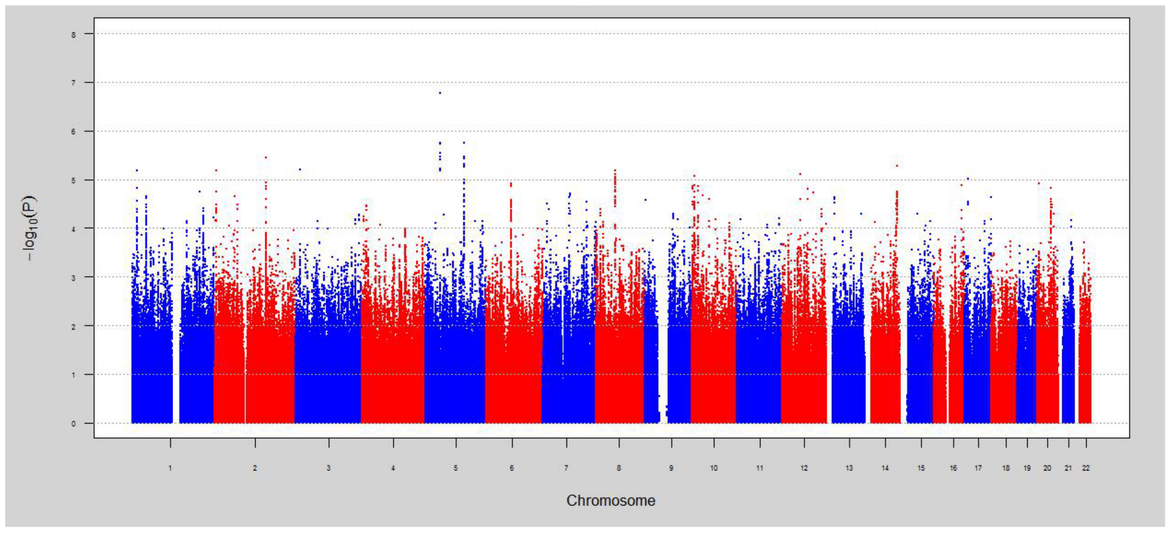
Chromosome

B)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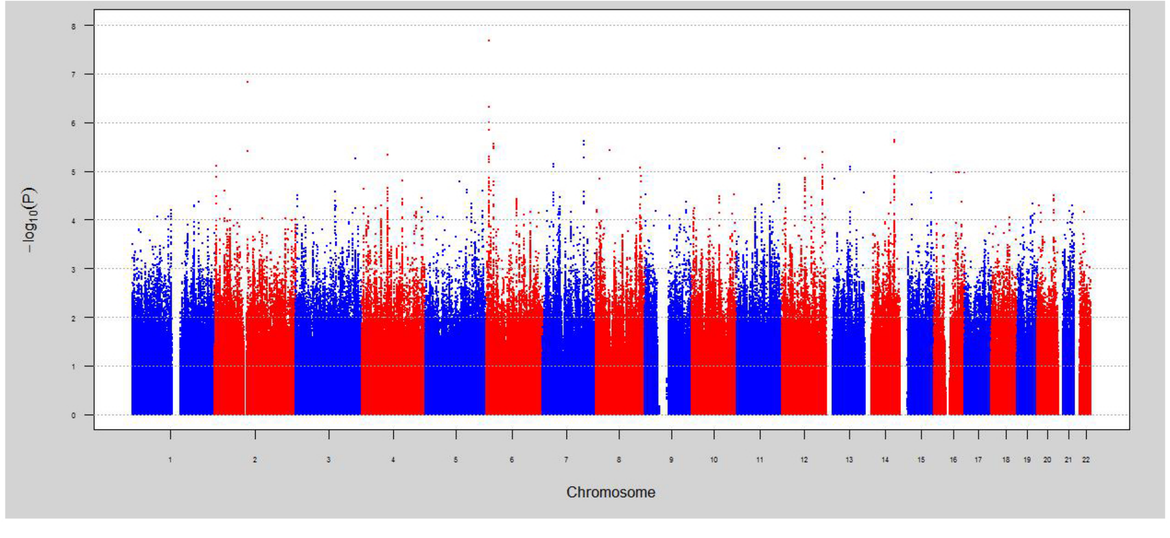
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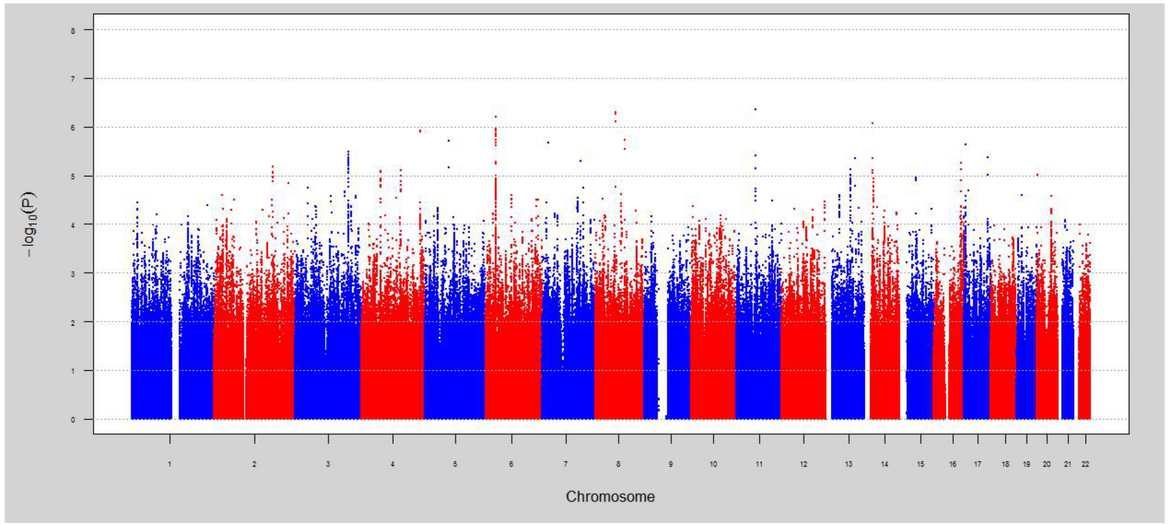
D)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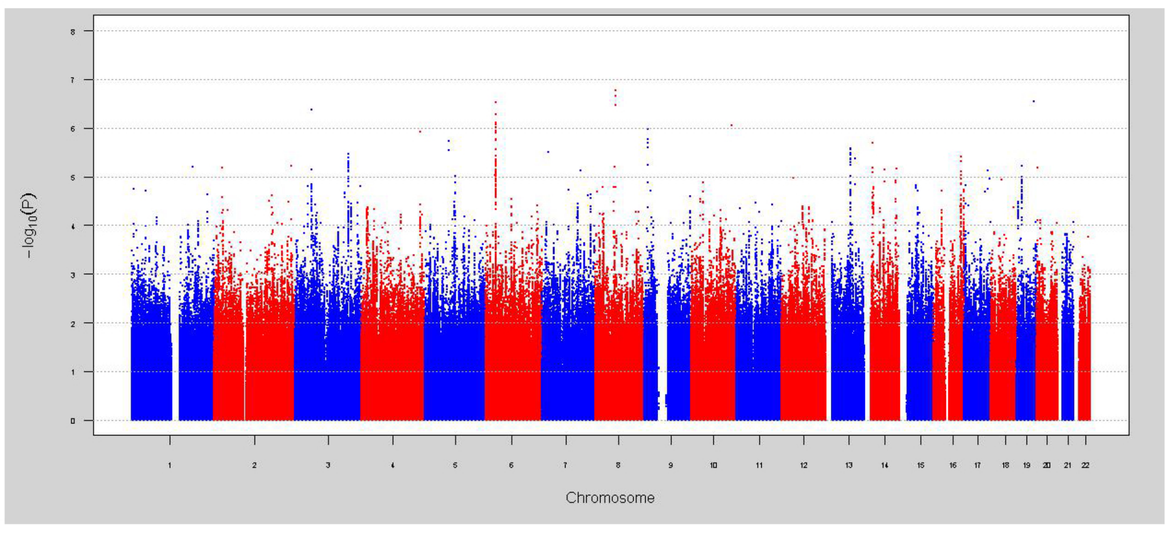
E)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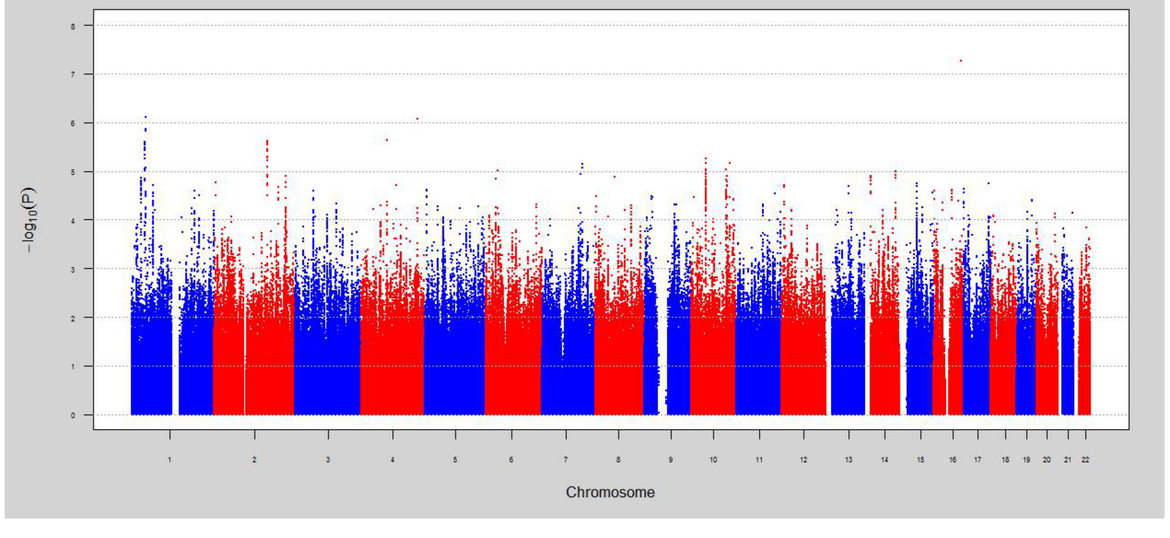
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