The genetics of idiopathic intracranial hypertension (IIH): Integration of population studies and clinical data

Supplemental Figures

Fig. S1. Distribution of BMI by sex.
Fig. S2. Manhattan plot and fine mapping of LRRFIP1 according to FinnGene Fz9 for IIH.
Fig. S3. Manhattan plot and fine mapping according to FinnGene Fz9 for IIH for PAP.
Fig. S4. Gene-based network of EnricR KB

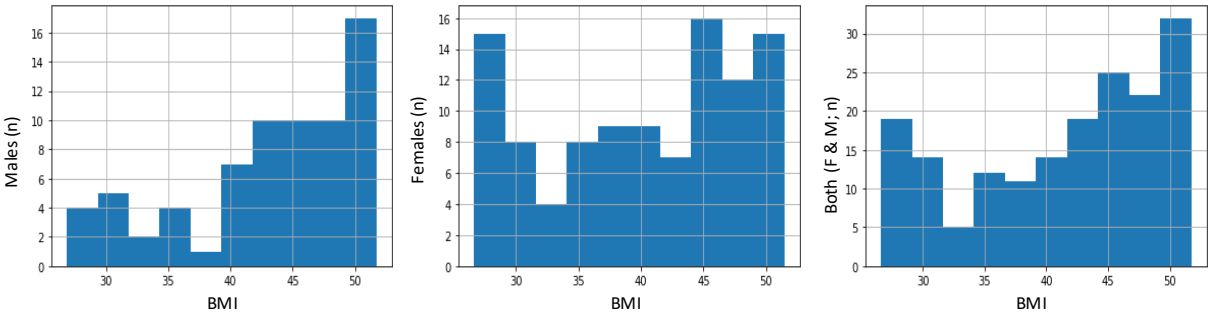

Fig. S1. BMI distribution of males, females and both. The mean and standard deviation are shown in Table 1.

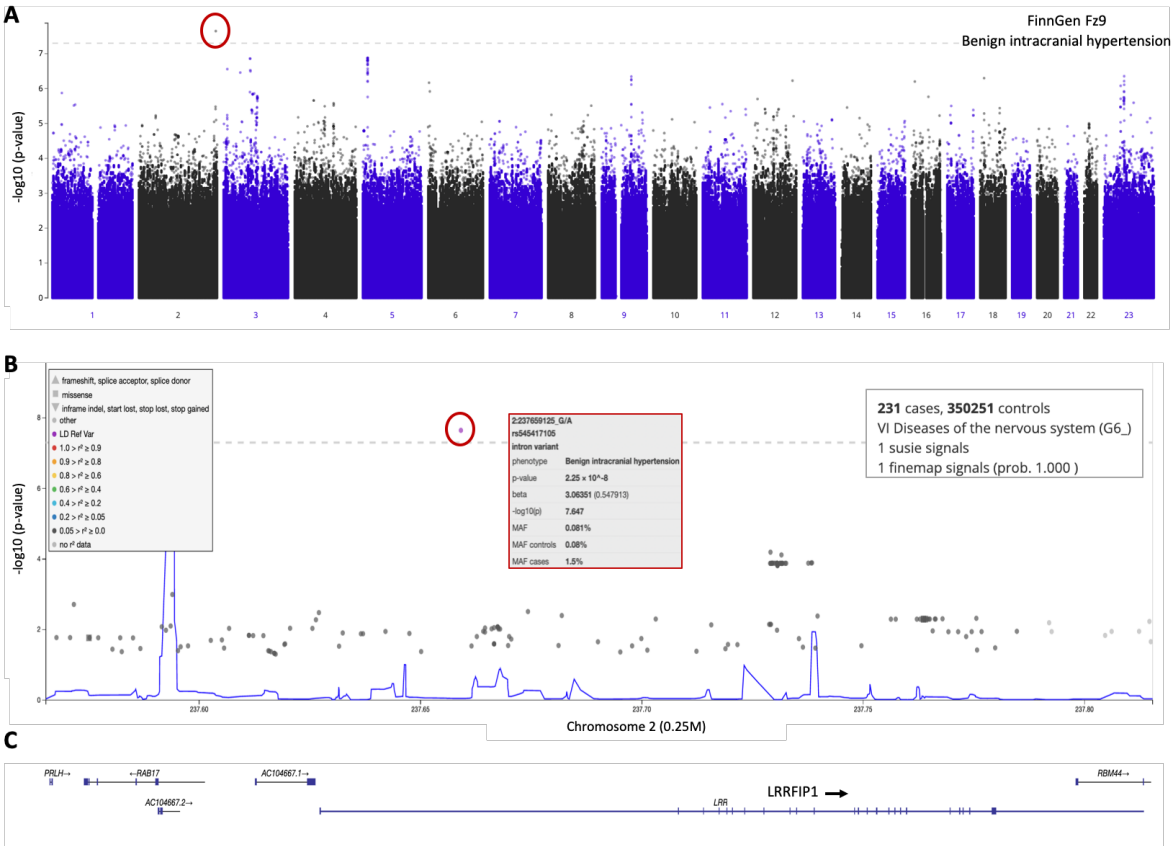

Fig. S2. FinnGen results for IIH associations. **(A)** Manhattan plot of the results from FinnGen Fz9 for Benign intracranial hypertension (named G6_BENINTRAHYP). The variant 2:237659125:G:A (rs545417105) reached significance with a p-value of 2.25e-08 (circled red). The analysis was performed on 231 cases and 350,251 controls**.** The dashed horizontal line marks the p-value threshold of 5.0e-08 for GWAS. **(B)** Fine map of variant 2:237659125:G:A (rs545417105) for a segment of 250k in Chr 2. The candidate SNP is associated with the statistical measures (red circles). Each imputed SNP is marked if the pair correlation r^2 is <0.05. The blue plot is an estimate for the recombination rate from the LD blocks. **(C)** Chromosomal segment of Chr2 from position 237.57 M to 237.82 M (total 250k). Gene lengths are shown by a horizontal line with the exons marked by the vertical lines. The arrow of LRRFIP1 denotes its transcription directionality.

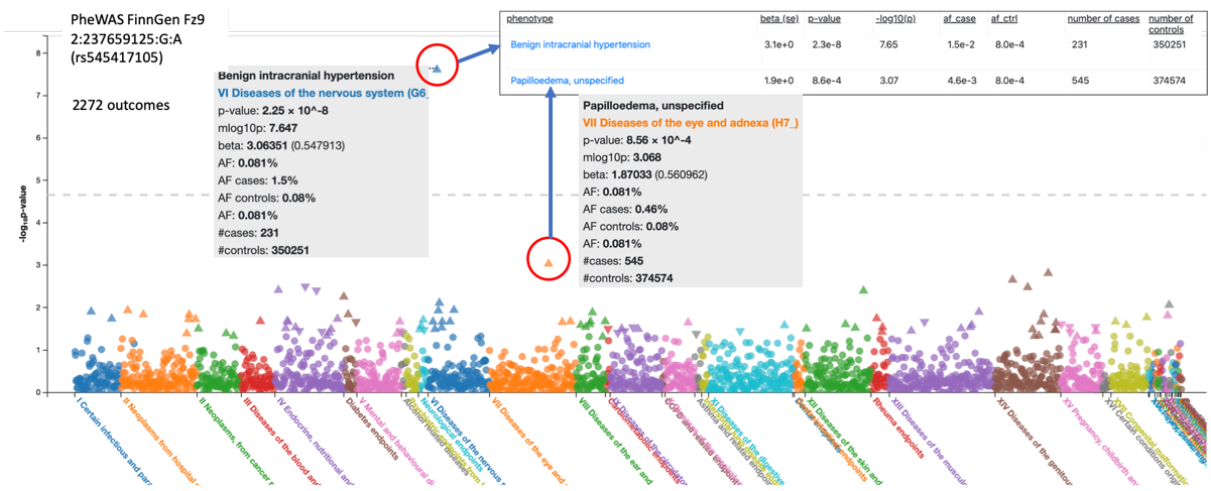

Fig. S3. PheWAS results for variant 2:237659125:G:A (rs545417105). Phenotypes (2272 codes) are clustered by their medical relevance (color coded). The triangle pointing up and down indicate an increase or decrease in the risk for the relevant disease, respectively. The most significant phenotypes were associated with an increased risk for IIH and PAP. The AF ratio of the alternative allele in cases and controls is 18.75 and 5.75 for IIH and PAP, respectively. The statistical evidence is shown in the inset.

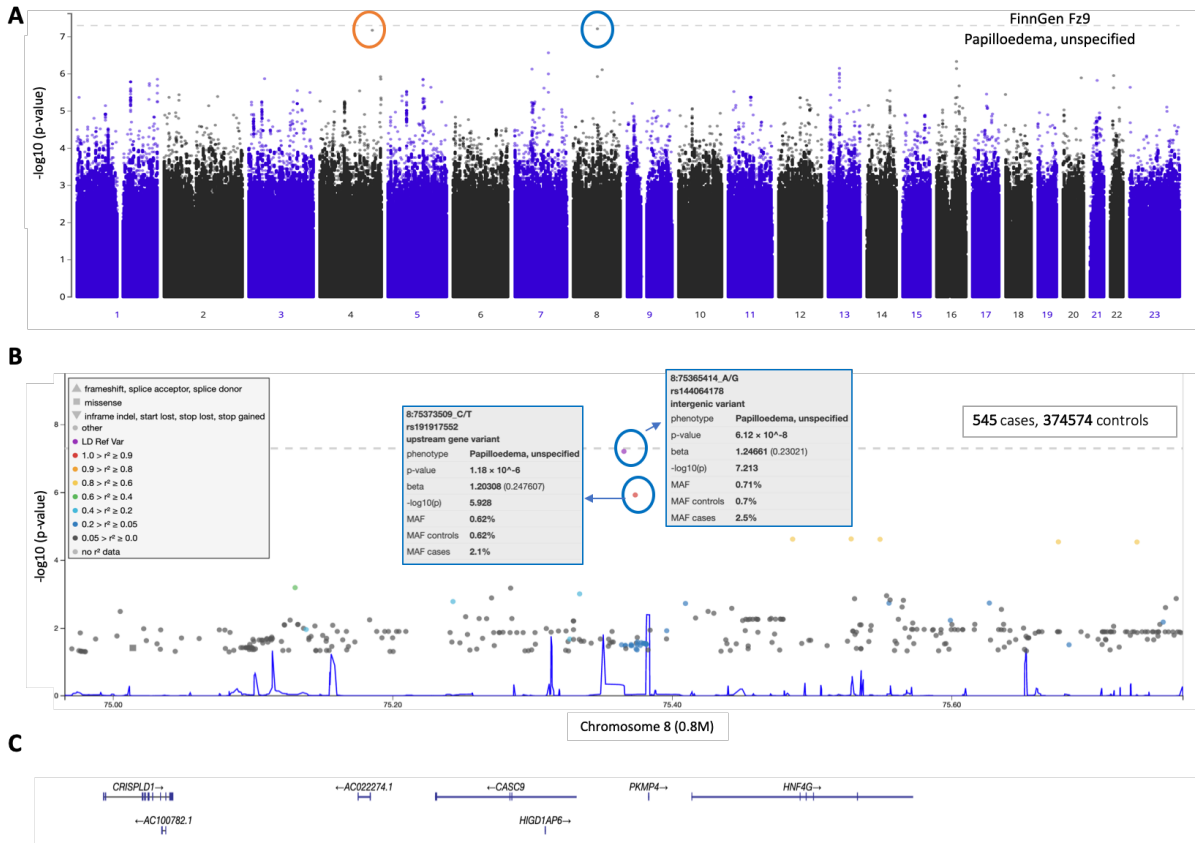

Fig. S4. FinnGen results for PAP associations. **(A)** Manhattan plot of the results from FinnGen Fz9 for papilloedema, unspecified. The variants 4:162403766:T:C (rs1235196153) and 8:75365414:A:G (rs144064178) are circled in orange and blue, respectively. Both variants are slightly below the significance threshold dashed horizontal line). **(B)** The fine mapping analysis for a segment of 1.6M in Chr 8. The candidate SNPs are associated with the statistical measures (blue circles). All imputed SNP are shown if the pair correlation (r^2) is <0.05. The blue plot is an estimate for the recombination rate (see Methods). **(C)** Chromosomal segment of Chr8 from position 75.0 M to 75.8 M (total 0.8M). Gene lengths are shown by horizontal lines with exons marked by the vertical lines. The arrows indicate transcription directionality. The nearest gene of the leading variant in Chr8 is HNF4G.

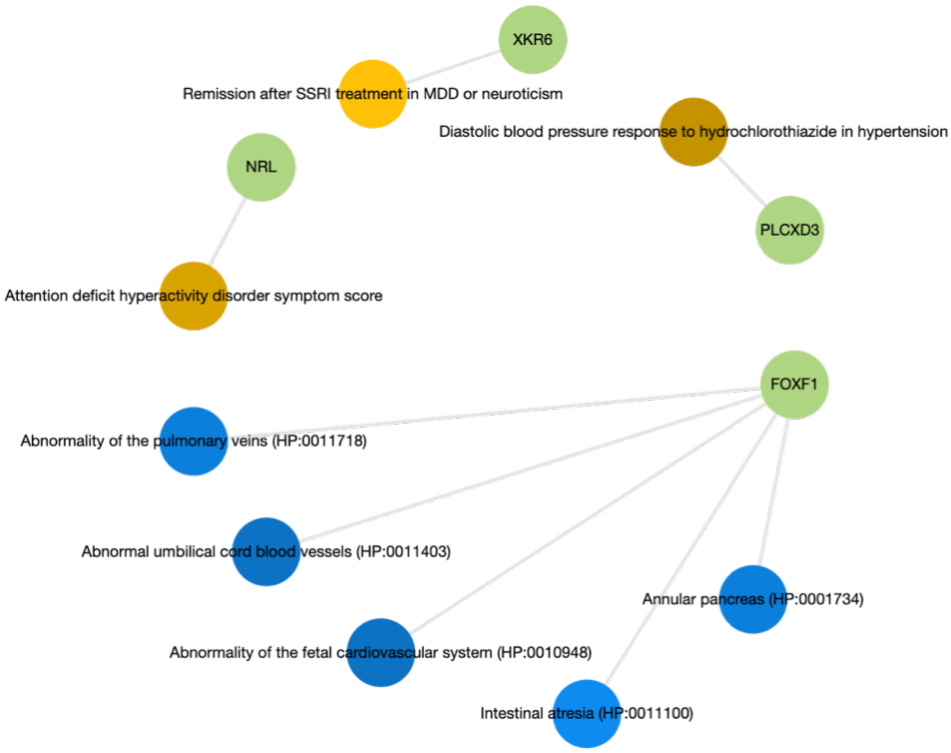

Fig. S5. A network graph based on SKAT top results of UKB IIH/PAP. Connectivity by evidence from GWAS catalog and HPO are shown. Only genes that are have a statistically significant are shown. The lighter color indicates a more significant q-value (data derived from the GWAS catalog and HPO with blue and orange, respectively). Connecting genes are colored green.