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Supplementary information

Mono- and biallelic variant effects on disease at biobank scale

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Supplementary Information

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Supplementary Tables

Glossary column names in Supplementary Tables

phenotype, disease endpoint name
variant, chr:pos:ref:alt
chr, chromosome,
pos, position in GRCH38
ref, reference allele
alt, alternate allele
rsid, rsid
INFO, overall genotype imputation INFO score
AF, allele frequency in FinnGen data
AN, total allele number in FinnGen data
AC_Hom, allele count in individuals homozygous for the alternate allele in FinnGen
enrichment_nfsee, AF in Finns/ AF in NFSEE (Non-Finnish-non-Swedish-non-Estonian)
grch37_locus, position in GRCH37
cs, included in credible set after fine-mapping (additive model, method: SuSiE)
pip, posterior inclusion probability after fine-mapping (method: SuSiE)
add_pval, SAIGE p-value (additive model)
rec_pval, SAIGE p-value (recessive model)
beta, SAIGE beta (log OR scale)
sebeta, SAIGE beta standard error
top_associated_phenotype_add, most significantly associated phenotype (additive model)
top_associated_phenotype_rec, most significantly associated phenotype (recessive model)
top_add_pval, p-value of most significant association (additive model)
top_rec_pval, p-value of most significant association (recessive model)
variant_category, gnomAD variant category
gene, gene from VEP (Variant Effect Predictor) annotation to which most severe consequence applies
R6, FinnGen data release 6 (n=234,553)
R4, FinnGen data release 4 (n=176,899))
ClinVar: Category, category grouped according to 'ClinVar_ReviewStatus'

ClinVar: Review, 'ClinVar_ReviewStatus' as given originally in ClinVar
Mode of inheritance (OMIM), Mode of inheritance annotated from OMIM for this gene ('both': both dominant and recessive inheritance modes annotated)

Supplementary Table S1. Any coding variant association that has a p-value < 10^{-7} in the additive model or a p-value < 10^{-7} in the recessive model (all available disease endpoints).

Supplementary Table S2. (A more detailed version of Table 1.) Recessive associations. Only the most significant variant is shown per locus. mis, missense; pLoF, probable loss-of-function; hom, homozygotes in FinnGen; R6, FinnGen data release 6 (n=234,553); UKBB, UK biobank (n=420,531).

Supplementary Table S3. Summary statistics of coding variants with any recessive association, i.e. recessive p-value < 10^{-7} and recessive p-value two orders of magnitude below additive p-value (all available disease endpoints, associations with p-value < 0.01).

Supplementary Table S4. Simulation results of genotype and phenotype data to assess the output of our recessive and additive model in the presence of a strictly recessive or additive association.

Supplementary Table S5. Variants with recessive associations in FinnGen that overlap known GWAS hits. Adjacent variants with r^2>0.25 associated with the same trait or same parent trait are considered as one locus and only the most significant variant shown per locus. The first five columns show gene, rsid, associated phenotype, recessive p-value and beta in FinnGen. Columns six to eight give the known GWAS phenotypes, SNPs and p-values. LD (r2>=0.1) of the known GWAS SNPs to respective SNPs in FinnGen is given in the last column. FinnGen, FG.

The six large homozygous effects in Supplementary Table 3 that we thus better captured with a recessive GWAS model have been previously mentioned for two out of the six variants: Homozygosity of rs601338 in *FUT2* has been shown to protect from intestinal infection[1]. Homozygosity of rs689 in *INS* had a significantly stronger effect on Type 1 Diabetes than in heterozygotes[2] and was previously associated with age-at-diagnosis of Type 1 Diabetes (P-value = 0.001), with homozygosity for the protective T allele delaying the onset of Type 1 Diabetes by approximately 2 years in these families[3]. (In our results, the direction of effect is the same, however the reference and alternative allele are swapped, see also Table 2 in the manuscript.)

Gene	rsid	Lead Phenotype FG	p-value FG	beta FG	known GWAS … phenotype in LD	... SNP	… r2	… p-value
UGT1A6	rs1105879	Cholelithiasis	4.10E-11	0.09	Gallstone disease; Serum bilirubin levels in metabolic syndrome	rs2070959; rs2070959	0.8; 0.8	$9x10^{-11}$; $2x10^{-14}$
IGHG3	rs74093865	Immunodeficiency w. predominantly antibody defects	4.40E-08	0.5	Composite immunoglobulin trait (IgG/IgM)	rs74093831	0.72	$8x10^{-9}$
FUT2	rs601338	Intestinal infectious diseases	4.00E-09	-0.07	Number of common colds;Vitamin B12 levels	rs601338; rs492602	1; 0.99	$4x10^{-7}$; $5x10^{-17}$
TMEM214	rs1124649	Pain (limb, back, neck, head abdominally)	3.80E-11	-0.06	CRP level; polymyalgia rheumatica	rs11681145	0.78;1	$9x10^{-12}$; 0.004
INS	rs3842753	Type 1 diabetes	1.20E-41	0.24	Type 1 diabetes	rs689	0.99	$5x10^{-196}$
SLCO1A2	rs10841795	Cardiac murmurs and other cardiac sounds	1.40E-08	1	Blood metabolite levels	rs12829704	0.6	$9x10^{-21}$

Supplementary Table S6 Variants with recessive associations in FinnGen that could be tested for replication in the UK biobank[4]. The last three rows colored in brown/grey show recessive associations of different coding variants in the UK biobank than the coding variant with a genomewide significant hit in FinnGen. UK biobank, UKB; FinnGen, FG; MAF, minor allele frequency.

Gene	rsid	Phenotype FinnGen	Phenotype UKBB	MAF FG	MAF NFE gnomAD	p-value FG	beta FG	p-value UKB	beta UKB
APPL2	rs138491961	noninflammatory disorders of uterus, except cervix	N85 noninflammatory disorders of uterus, except cervix	0.0032	0.0015	3.69E-08	5.54	NA	NA
CEP250	rs56259282	Varicose veins	I83 Varicose veins of lower extremities	0.0046	0.012	1.27E-08	4.33	0.027	1.97
C10orf90	rs139123090	Conductive and sensorineural hearing loss	H90 Conductive and sensorineural hearing loss	0.0085	0.0045	2.25E-12	5.96	0.00012	56.7
PCK1	rs201186470	Glucose regulation and pancreatic secretion	E16 Other disorders of pancreatic internal secretion	0.011	0.00045	4.65E-08	9.77	NA	NA
GJB2	rs398123814	Sensorineural hearing loss	H90 Conductive and sensorineural hearing loss	0.012	0.0092	2.86E-28	6.49	3.11E-08	82.0
C15orf40	rs115868366	Demyelenating diseases of the central nervous system	Multiple sclerosis	0.013	0.018	3.94E-08	11.3	0.058	2.53
CASP7	rs141266925	Other cataract	H26 Other cataract	0.015	0.0035	2.47E-16	4.77	NA	NA
EBAG9	rs1422	Female infertility	N97 Female infertility	0.056	0.041	1.60E-11	2.0	0.0018	3.90
SERPINA1	rs28929474	Emphysema	J43 Emphysema	0.02	0.018	2.16E-21	28.0	1.43E-26	20.6
INS	rs3842753	Type 1 diabetes	Type 1 diabetes	0.79	0.72	5.71E-46	0.28	6.31E-23	0.35
IGHG3	rs74093865	Immunodeficiency w. predominantly antibody defects	D80 Immunodeficiency w. predominantly antibody defects	0.4	0.3	4.44E-08	0.50	0.027	0.53
UGT1A6	rs1105879	Cholelithiasis, broad definition with cholecystitis	Cholelithiasis and cholecystitis	0.45	0.35	1.53E-11	0.08	1.58E-10	0.16
SLCO1A2	rs10841795	Cardiac murmurs and other cardiac sounds	R01 Cardiac murmurs and other cardiac sounds	0.11	0.14	1.38E-08	1.03	0.47	0.14
TMEM214	rs1124649	Pain (limb, back, neck, head abdominally)	General pain for 3+ months	0.28	0.31	3.79E-11	-0.06	0.41	-0.10
ZNF415	rs10410030	Mild mental retardation	F81 Specific developmental disorders of scholastic skills	0.097	0.15	1.56E-08	2.10	0.42	-0.37
FUT2	rs601338	Intestinal infectious diseases	Intestinal infection	0.37	0.48	4.01E-09	-0.07	0.21	-0.03
CASP7	rs2227309	Other cataract	H26 Other cataract	0.28	0.26	NA	NA	0.016	0.07
APPL2	rs2272495	noninflammatory disorders of uterus, except cervix	N85 noninflammatory disorders of uterus, except cervix	0.23	0.20	NA	NA	0.0085	0.18
PCK1	rs1804160	Glucose regulation and pancreatic secretion	E16 Other disorders of pancreatic internal secretion	0.043	0.023	NA	NA	0.038	1.67

Supplementary Table S7. Variants with recessive associations in FinnGen that could be tested for additive associations in the UK biobank (UKBB)[4]. The columns show gene, rsid, phenotype name in UKBB, additive p-value and beta in UKBB and the phenotype name in FinnGen.

Gene	rsid	Phenotype UKBB	p-value	beta	Phenotype FinnGen
ACOXL	rs80265967	Gestational diabetes	>0.05	NA	Diabetes mellitus in pregnancy
APPL2	rs138491961	noninflammatory disorders of uterus, except cervix	>0.05	NA	N85 Other noninflammatory disorders of uterus, except cervix
CASP7	rs141266925	Cataract	4.50E-12	0.7	Cataract
C10orf90	rs139123090	Conductive and sensorineural hearing loss	>0.05	NA	Conductive and sensorineural hearing loss
C15orf40	rs115868366	Demyelenating diseases of the CNS	1.40E-02	0.34	multiple sclerosis
CEP250	rs56259282	Varicose veins	>0.05	NA	Varicose veins
CLCN1	rs55960271	Diseases of the myoneural junction and muscle	>0.05	NA	Myasthenia gravis and other myoneural disorders
CNGB1	rs201162411	Hereditary retinal dystrophy	>0.05	NA	Hereditary retinal dystrophy
DMBT1	rs189221852	Nephropathy	2.20E-04	5.3	Non-proliferative glomerulonephritis
EBAG9	rs1422	Female infertility	>0.05	NA	Female infertility
IGHG3	rs74093865	Immunodeficiency w. predominantly antibody defects	>0.05	NA	Immunodeficiency with predominantly antibody defects
INS	rs3842753	Type 1 diabetes	9.10E-20	0.28	Type 1 diabetes
PCK1	rs1804160	Other disorders of pancreatic internal secretion	>0.05	NA	Glucose regulation and pancreatic secretion
PLTP	rs56126980	Unspecified lump in breast	>0.05	NA	Lump or mass in breast
SERPINA1	rs28929474	Emphysema	2.30E-19	1.3	Emphysema
SLCO1A2	rs10841795	Cardiac murmurs and other cardiac sounds	>0.05	NA	Cardiac murmurs and other cardiac sounds
TMEM214	rs1124649	Pain (limb, back, neck, head abdominally)	4.00E-03	-0.13	polymyalgia rheumatica
UGT1A6	rs200711686	Cholelithiasis	1.40E-06	0.064	Cholelithiasis
USP5	rs200723728	Endometriosis of ovary	>0.05	NA	Endometriosis
USP8	rs147742292	Postpartum haemorrhage	>0.05	NA	Postpartum haemorrhage
ZNF415	rs10410030	Mild mental retardation	2.90E-02	0.31	Mild cognitive impairment

Supplementary Table S8. Most significant disease associations of all 15140 coding variants annotated in ClinVar.

Supplementary Table S9. Likely benign ClinVar variants that are the most likely causative (top PIP) variant in one GWAS locus. Genomic locations are given as rsids and GRCh38 coordinates (chromosome, position in bp, reference and alternate allele, separated by ":"). PIP, posterior inclusion probability (statistical finemapping[5]); AF, allele frequency in FinnGen; AD, autosomal dominant; AR autosomal recessive; KELA, Finnish Social Insurance Institution. All listed variants are missense variants.

Gene	GRCh38	rsid	AF	OMIM phenotype of gene	GWAS phenotype in FinnGen	beta	p-value	PIP
ARMS2	10:122454932:G:T	rs10490924	0.24	Age-related macular degeneration (association)	Age-related mac. degen.	1.06	1.90E-104	0.08
					Other retinal disorders	0.27	1.78E-46	0.08
					Diabetic retinopathy	0.16	3.92E-22	0.08
ATM	11:108272729:C:G	rs1800057	0.01	Ataxia-telangiectasia AR;	Malignant neoplasm	0.28	2.58E-08	0.4
				Breast cancer susceptibility	Leiomyoma of uterus	0.45	1.13E-09	0.22
INS	11:2159830:T:G	rs3842753	0.79	Diabetes m. (neonatal, AR, AD, MODY)	Diabetic maculopathy	0.32	1.04E-10	0.44
					Diabetic retinopathy	0.32	5.55E-19	0.51
HNF1A	12:120978847:A:C	rs1169288	0.37	Diabetes m. (AR, AD MODY)	Statin medication	0.08	1.55E-14	0.86
KRT5	12:52519884:C:T	rs11170164	0.1	Epidermolysis bullosa simplex, AR, AD	Malignant neoplasm of skin	0.21	1.34E-12	0.94
RPL3L	16:1947063:G:A	rs147972626	0.01	Cardiomyopathy, dilated, AR	Atrial fibrillation and flutter	0.45	6.23E-11	0.49
TYK2	19:10352442:G:C	rs34536443	0.03	Immunodeficiency, AR	KELA Reimbursement (Gastrointestinal)	-0.3	6.09E-09	0.7
CFH	1:196690107:C:T	rs1061170	0.56	Complement factor H deficiency, AR, AD; Macular deg. (association)	Disorders of choroid and retina	-0.11	5.18E-16	0.02
					Diabetic retinopathy	-0.09	2.05E-10	0.02
PCSK9	1:55039974:G:T	rs11591147	0.04	Hypercholesterolemia, familial, AD	Hypercholesterolaemia	-0.43	1.33E-16	1
					Coronary atherosclerosis	-0.22	4.59E-09	0.41
					Myocardial infarction	-0.32	1.48E-11	0.94
					Coronary revascular.	-0.36	1.10E-12	0.93
					Statin medication	-0.61	5.26E-119	1
HNF4A	20:44413724:C:T	rs1800961	0.05	MODY, type I, AD	Cholelithiasis	0.38	1.27E-30	1
					Intrahepatic Cholestasis of Pregnancy	0.82	2.43E-09	1
AIRE	21:44294411:C:T	rs74203920	0.04	Autoimm. polyendocrinopathy syndrome AR, AD	Diabetic maculopathy	0.64	2.39E-09	0.99
					Type 1 diabetes	0.38	2.19E-10	0.96
					Diabetic retinopathy	0.48	9.48E-11	1
IFIH1	2:162267541:C:T	rs1990760	0.58	Aicardi-Goutieres AD;	Hypothyroidism	0.08	8.88E-11	0.62
				Singleton-Merten, AD	Disorders of the thyroid gland	0.07	5.43E-09	0.51
APOB	2:21041028:G:A	rs1367117	0.28	Hypercholesterolemia AD	Disorders of lipoprotein metabolism	0.11	6.64E-12	0.51
				Hypobetalipoproteinemia, AR				
ZAP70	2:97725153:C:T	rs145955907	0.019	Autoimmune disease, infantile, AR	Hypothyroidism	0.24	1.83E-08	0.43
LRBA	4:150277928:G:A	rs2290846	0.22	Immunodeficiency, with autoimmunity, AR	Cholelithiasis	0.12	1.31E-13	0.52
					Disorders of biliary tract and pancreas	0.1	2.19E-12	0.7
DBH	9:133636606:C:T	rs77273740	0.05	Orthostatic hypotension due to DBH deficiency, AR	Hypertensive diseases	-0.16	6.16E-13	0.99

Supplementary Table S10. Summary statistics of all coding variants in FinnGen release 4 (any additive association with p-value $<1x10^{-4}$).

Supplementary Notes.

1) Variants causing disease with recessive inheritance are at higher MAF in Finnish than Non-Finnish Europeans. In light of a global enrichment of deleterious variants in Finland[6,7] and the well-described "Finnish Disease Heritage"[8,9], we hypothesized that a set of known disease-causing variants should exist at higher frequencies in Finland. We thus compared minor allele frequencies (MAF) of 2419 unique variants that were listed in the disease variant database ClinVar[10] (version 2018) as pathogenic or likely pathogenic (likely pathogenic) in genes with only recessive inheritance annotated in the Online Mendelian Inheritance in Man, OMIM® (omim.org) between 10,824 Finnish (FIN) and 10,824 Non-Finnish Europeans (NFE) taken from the gnomAD population database[11] (Supplementary Figure S1). Concordant with a known lower rare variant diversity[9], only 554 likely pathogenic variants were present in FIN while 2169 were present in NFE at same sample sizes. As expected, however, observed likely pathogenic variants were at higher MAF in FIN (median MAF 9.2×10^{-5}) than NFE (median MAF = 4.7×10^{-5}, p-value = 6×10^{-7}, Wilcoxon rank test), with the difference particularly pronounced in 133 variants in Finnish disease heritage genes[8] (see Supplementary Figure S1). In FIN, 12% (67/554) of likely pathogenic variants were above MAF 0.001 (39% [217/554] above MAF 10^{-4}) compared to 2.5% (54/2169) in NFE (26% [559/2169] above MAF 10^{-4}).

2) Simulations of recessive and additive associations. We performed simulations of genotype and phenotype data to assess the output of our recessive and additive model in the presence of a strictly recessive or additive association. We simulated genotype and phenotype data aiming to resemble our own data. Specifically, we generated genotype counts of wildtype, heterozygotes and mutant homozygotes in 200,000 individuals of a variant at four different MAF thresholds following Hardy Weinberg Equilibrium (using R library HardyWeinberg[12]). We then generated random phenotype data of cases versus controls of a disease with a prevalence of 0.05 using the rbinom function in base R. We tested a recessive and an additive scenario. For the recessive scenario, we set the probability of homozygotes to develop the disease to 5x compared to wildtype and set heterozygote effect sizes to 1 corresponding to no heterozygous effect. For the additive scenario, we set the heterozygote effect to 1.5 and homozygous effect to 2.25 corresponding to a strictly additive effect on a logarithmic scale. We then performed logistic regression using the glm function in R with an additive and a recessive model, respectively.

For visualization and a table of results please see Extended Data Figure 2 and Supplementary Table S4. In summary, we find that if a simulated recessive effect reaches genomewide significance in the recessive model (recessive p-value <5x10e-8), the recessive p-value is 2 orders of magnitude below the additive p-value in 298,414/298,418 instances across 4x100,000 simulations. Thus, if truly recessive associations reach genomewide significance, we will likely identify them as recessive with our approach. Conversely, we find that for a simulated strictly additive effect in logarithmic space (heterozygous effect 1.5, homozygous effect 2.25; 100,000 simulations each at minor allele frequency thresholds 0.01, 0.02, 0.05, 0.1) recessive p-value is 2 orders of magnitude below the additive p-value in 0/400,000 simulations. We can thus with high confidence reject strict additivity when recessive p-value is 2 orders of magnitude below the additive p-value.

In an earlier version of this manuscript, we found in additional simulations that for increasing sizes of heterozygous effects the additive model's significance increased accordingly [13]. We found similar results for varying effect sizes and MAF.

3) Novel recessive disease associations. Here, we highlight two variants with large homozygous effects in genes with no previous disease associations, in *CASP7* associated with cataract, *C10orf90* associated with hearing loss and *EBAG9* associated with female infertility.

We would first like to present a missense variant in *CASP7* with a MAF of 0.015 in Finland (4x enriched in Finns) that we found associated with adult-onset cataract. The variant had large homozygous effects on the distinct phenotypes “other cataract” (p-value = $2x10^{-16}$, beta 4.8, most frequent ICD10 codes in 43 homozygotes: 13x secondary cataract and 7x presenile cataract) and “senile cataract” (p-value = $2x10^{-12}$, beta 2.1). While there were too few homozygotes in the UKBB we could replicate a genomewide significant effect on phenotype “all cataracts” in the additive model in UKBB (p-value = $5x10^{-12}$, beta 0.7) suggesting a potential heterozygous effect or homozygous effect of other variants in LD. There was also some evidence for an effect on ”other cataracts” in heterozygous carriers in FinnGen (SAiGE GWAS p-value 0.003, beta 0.25). Age at first diagnosis was nominally significantly earlier in homozygotes (see Supplementary Figure 5). In the age group below 63 (mean retirement age in Finland) 50% of homozygotes (13/26) but only 3.4% (94/2750) of heterozygous and 3.1% (2710/87799) of wildtype carriers had already reported cataracts in our data. *CASP7* encodes Caspase-7, an executing enzyme in apoptosis (programmed cell death) [14]. In lens development, cell organelles are degraded to create transparent cells by a process very similar to

apoptosis. Further, deactivating apoptosis genes in mice (e.g. *Casp3*[15] or *Apaf-1* and *p53*[16]) has been shown to lead to cataract and there has been evidence for apoptosis involved in human non-congenital cataract[17,18]. *CASP7* is therefore a biologically plausible and to the best of our knowledge first Mendelian gene for adult-onset cataract, the leading cause of vision loss worldwide with no other effective treatment than surgery and therefore particularly affecting low/middle income countries (Source: WHO, 2012).

The missense variant in *C10orf90* was associated with sensorineural hearing loss also with Mendelian effects (recessive p-value = $5x10^{-12}$, beta 6.0; additive p-value > 10^{-4}), which we could replicate in the UKBB (recessive p-value $1x10^{-4}$, beta 56). The variant was 2.8x enriched in Finns with a MAF of 0.0083 in Finland. Homozygous carriers had significantly earlier onset of diseases: among 14k individuals diagnosed with hearing loss carriers of homozygous variants in *C10orf90* were first diagnosed at a mean age of 40±17 (mean±sd) compared to wildtype carriers (63±16, p-value $8*10^{-5}$, Wilcoxon-rank test). There is less known about the gene *C10orf90* which is also called fragile-site associated tumor suppressor (*FATS*)[19]. However, mutations in its most closely related paralog gene *ALMS1* cause the ciliopathy "Alstrom syndrome" with sensorineural hearing loss as one of its main symptoms[20]. As there is also some evidence for a role of *C10orf90* in cilia formation[21], it is plausible that *C10orf90* would also cause hearing via an effect on cilia. However, since there are no other symptoms in variant carriers beyond expectation apart from hearing loss, this is likely restricted to hearing loss and therefore distinct from *ALMS1*.

Next, we find a noncoding variant in *EBAG9* with a homozygous effect on female infertility. This is the only noncoding variant we investigated in our study but we believe it is the more likely causal candidate than the linked PTV variant in *PKHD1L1* we initially identified as it has a larger effect (beta 2.0 compared to 0.21), it is more likely causal after finemapping (PIP 0.2 while the PTV in *PKHD1L1* is not in the credible set) and *EBAG9* is a known estrogen responsive gene. 48 of 4,505 women (1.1%) with infertility in FinnGen are homozygous for the *EBAG9* variant. In FinnGen R6 data, we investigated the effect of the *EBAG9* variant on fertility in 106,732 women born between 1925 and 1975 for whom we had thus approximately the whole reproductive life span available. We find that 357 homozygotes of the *EBAG9* variant had fewer children (1.7±1.3 [mean±sd], p-value 0.0002, Wilcoxon rank test) than wildtypes (2.0±1.3) and a later age at first child (27.2±5.2 versus 28.3±5.5, p-value 0.01, Wilcoxon rank test). This effect is mainly driven by 71 *EBAG9* homozygotes

diagnosed with infertility as it is in the expected direction but not significant in 302 *EBAG9* homozygotes without the infertility diagnosis (mean 1.85 children, p-value 0.08, Wilcoxon rank test). Among all 7,980 women diagnosed with infertility 71 *EBAG9* homozygotes were more severely affected as they had fewer children (1.0±0.9 [mean±sd]) than other women with infertility diagnoses (1.5±1.2 [mean±sd]) p-value 0.005, Wilcoxon rank test) and had their first child significantly later (32.3±4.6 versus 30.0±5.6, p-value 0.005, Wilcoxon rank test).

4) Compound heterozygous effects. The genes *GJB2* and *EYS* harbored more than one pathogenic variant with significant recessive associations in our data so we sought to explore compound heterozygous (=comp-het) effects. We computed linkage disequilibrium (LD) between those variants with a Finnish population reference panel consisting of 2,244 high-coverage (30x) whole genome sequenced individuals[22]. LD between the two *GJB2* variants (r^2=0.002) was negligible but the two *EYS* variants were in strong LD (r^2=0.47) so we could not investigate comp-het effects for *EYS*. In *GJB2,* we found early onset sensorineural hearing loss in 100 *GJB2* comp-hets, i.e. individuals with one pathogenic missense and one pLoF *GJB2* variant (p-value $6x10^{-28}$, HR 6.1, coxph) that each also had effects in homozygous state. In agreement with the variants' different homozygous effect sizes, the effect of comp-het was in between the homozygous pLoF (HR 15, p-value $7x10^{-37}$) and homozygous missense variants' effect sizes (HR 3.9, p-value $8x10^{-15}$) (see Figure 3C). We also tested the effects of another likely pathogenic missense variant in *GJB2.* LD with the other *GJB2* variants was negligible (r^2~0.00 with both). That variant was at a much lower MAF of 0.004. Therefore, our dataset included only 3 individuals in homozygous state, 4 in compound heterozygous state with the pLoF *GJB2* variant and 5 in compound heterozygous state with the other missense *GJB2* variant. Combined, all 12 variants were associated with early onset sensorineural hearing loss with similar effect as the other pathogenic variants in *GJB2* (p-value $3x10^{-12}$, HR 11.8, coxph). We also investigated comp-het effects in *CERKL* as there was an additional known pathogenic pLoF variant in our data chr2:181656814:C:A (hg38) at low frequency (MAF 0.0019) which was thus not well-powered to reach significance in homozygous state in the pheWAS. When excluding individuals with homozygous and comp-het *CERKL* variants we did not find a heterozygous effect of any of the pathogenic variants on the phenotype of retinal dystrophy anymore suggesting comp-het effects drove the initial nominally significant heterozygous effect.

5) Known disease causing variants in FinnGen. We studied 15,140 variants that were annotated in ClinVar[10] in 176,899 Finns in the FinnGen research project. These included 311 coding variants that were annotated as likely pathogenic, 147 of them by multiple submitters or an expert panel (corresponds to ClinVar review status of 2-4 out of 4 possible stars) which we thus describe as being annotated as likely pathogenic with "high confidence". 298 further variants had "conflicting interpretations of pathogenicity" but were classified as likely pathogenic by at least one submitter. 3.4% of study participants carried at least one of ten high confidence likely pathogenic variant in one of nine genes causing disease with dominant inheritance according to OMIM. For 6 out of the 10 likely pathogenic variants we found expected genomewide significant disease associations (see also Supplementary Table S1) in the genes *FLG* (atopic dermatitis, 2690 and 1050 heterozygotes, respectively), *MYOC* (glaucoma, 1023 heterozygotes), *PALB2* (breast cancer, 512 heterozygotes), *JAK2* (Polycythaemia vera, 194 heterozygotes) and *MYBPC3* (Hypertrophic cardiomyopathy, 116 heterozygotes). Accordingly, we did not find genomewide significant associations for 4 of 10 high confidence likely pathogenic variants of which *CHEK2* was genomewide significant for breast cancer in the later FinnGen datafreeze R6. Variants in *COL5A2, G6PD* and *PKD1* were not significant in later releases. For *G6PD* this may be expected as the variant is rare (58 hemizygous males) and functionally predicted to lead to Glucose-6-phosphate dehydrogenase activity of 15 to 25 percent of wildtype (Cappellini 1994, Cappellini 1995, De Vita 1989, Frigerio 1994, Lenzerini 1969, Rattazzi 1969), thus characteristic of a moderate (Class III) deficiency where hemolysis is only triggered by stressors. This rare and moderate effect may thus be underpowered to be detected by our EHR data. For variants in *COL5A2* and *PKD1* we would expect at least nominally significant disease associations in later FinnGen releases, given they contain > 60,000 individuals with joint disorders and >1,000 individuals with cystic kidney disease. The fact that there are only two submitters for the variant in *COL5A2* and one submitter classifying the variant in *PKD1* as VUS in a later ClinVar release adds to the suspicion that they may in fact not be truly disease associated.

All of 0.41% of participants (n=726) carried one of 98 high confidence likely pathogenic variants in homozygous state in one of 86 disease genes with known recessive inheritance, 41.8% were heterozygous carriers. Most frequent homozygous mutants were in the genes *BTD* (partial biotinidase deficiency, 523 homozygotes), *GJB2* (nonsyndromic sensorineural hearing loss, 113 homozygotes) and *SERPINA1* (Alpha-1-antitrypsin deficiency, 77 homozygotes). We found

expected homozygous phenotype associations of *GJB2* and *SERPINA1* variants with genomewide significance. We were however surprised by the lack of genomewide signifiant disease associations for the known pathogenic variant [*BTD*:p.(Asp444His)] linked to biotinidase deficiency. *BTD*:p.(Asp444His) has been reported as pathogenic for biotinidase deficiency by 17 submitters in ClinVar and experimentally led to a ~25% reduction in biotinidase activity[23]. Homozygotes for *BTD*:p.(Asp444His) are thus expected to have ca. 50% of normal serum biotinidase activitiy. As partial biotinidase deficiency is however characterized by 10%-30% of normal serum biotinidase activity[24] the lack of disease associations above chance expectation for *BTD*:p.(Asp444His) homozygotes makes sense. We can thus empirically confirm the experimental hypothesis that the variant should only cause disease when compound heterozygous with a variant leading to near-zero biotinidase activity.

We also demonstrate how our results help to verify disease associations of variants with low sample sizes. One example is a missense variant in *SPINK1* with conflicting interpretations of likely benign (1x), likely pathogenic (8x) and VUS (3x) on ClinVar. In our data, it is the only coding variant, most significant and most likely causal variant (finemapping probability 0.19 in FinnGen datafreeze 8) at the respective locus associated with alcohol-induced acute and chronic pancreatitis as well as acute and chronic pancreatitis more generally with odd's ratios between 2.8-8.5. Its inheritance is best described as semidominant with an additive genotype dosage effect. Another example is a variant in *SOD1* with conflicting interpretations of likely benign (1x), likely pathogenic (5x) and VUS (4x) on ClinVar. In our data, we find a large homozygous effect on motor neuron disease with odd's ratio > 100 while not detecting a heterozygous effect which we can confirm with R6 data in a total of 8 cases. We thus classify inheritance as recessive. We are currently preparing the full list of associations of unclear disease variants for submission to ClinVar. We can thus show that we are well-powered to identify known associations across a wide range of disease phenotypes in our dataset. In addition, it can serve as a resource to verify or falsify previously described disease associations. For verification purposes the statistical finemapping can be additionally helpful to identify associations' causal variants.

6) Benign and likely benign (likely benign) variants in FinnGen. We studied effects of variants listed in ClinVar [10] as likely benign. Those are regarded as "not the cause for a patient's disorder" ACMG [25]. However, we found more phenotype associations than expected in likely benign ClinVar variants as a group.

To test whether these might potentially just correlate with actual causal variants in linkage disequilibrium we assessed how often they were included in the 95%-credible set (cs) for a given GWAS locus [5]. We found on the contrary, that 142 likely benign missense variants were slightly more often in the 95%-cs than 307 missense variants not annotated in ClinVar (Fisher's Exact test, p-value 0.05, OR 1.5, 95%-CI 1.0-2.4). 16 represented even the most likely causal variant for a GWAS locus. The ClinVar annotation labeling these variants as likely benign was above average quality (1.6; 7 of the 16 likely benign top causal variants had a one star and 9 of 16 a two-star review status in ClinVar) compared to an average 1.2 stars for all likely benign variants in ClinVar (range: zero to three stars). Thus, poor quality of ClinVar classification as likely benign cannot explain why so many benign variants in our dataset are disease associated.

Expectedly, the disease phenotypes in FinnGen – though often related - are mostly different from the known OMIM phenotypes of the respective genes. Nevertheless, disease associations of likely benign variants may still be counterintuitive as they are often interpreted as generally disease-irrelevant [26,27]. How could we then explain global disease associations for them? Firstly, it is plausible that the frequent diagnostic sequencing and analysis of disease-associated genes with high clinical interest also results in submission of many likely benign variants to ClinVar for those genes. Hypothesizing that genes with many disease-causing variants would therefore also get many ClinVar submissions of likely benign variants, we found indeed a positive correlation of the number of likely pathogenic and likely benign variants per gene in ClinVar (r^2 0.41, rho 0.07, respective p $<10^{-16}$). Genes with many disease-causing variants - particularly relevant to human disease - may be more likely involved in additional diseases than disease-irrelevant genes [28]. In line with that we found indeed that likely benign ClinVar missense variants had more significant phenotype associations than missense variants in different genes but that signal disappeared mostly when missense variants were matched to the same genes. In addition, likely benign variants are of significantly higher MAF (0.07±0.12, mean, sd) than other ClinVar variants (0.01±0.05) in FinnGen which is expected as MAF is a criterion for labeling a variant as likely benign according to official ACMG guidelines [25]. So we are well-powered to find significant disease associations for likely benign variants, particularly in a bottleneck population like Finland that enriches disease variants to higher frequencies [9]. Of note, when investigating unique ClinVar variants in the 95%-cs, 19 out of 67 likely benign ClinVar variants had protective effects on disease, while only 1 out of 27 ClinVar risk variants (i.e. likely pathogenic, conflicting or risk factor variants) was protective (Fisher's

Exact test, p-value 0.01 OR 10, 95%-CI 1.4-443). While their identified effects are more often protective than for known risk variants the majority of associations increase disease risk also in benign variants! In conclusion, we think it is important to keep in mind that variants labeled as likely benign are not generally neutral and can still contribute to disease.

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