Replication validity of genetic association studies
REFERENCE APPENDIX

A. Eligible meta-analyses (ID number corresponds to table 1 of the main paper)

1. Agerholm-Larsen B, Nordestgaard BG, Tybjaerg-Hansen A. ACE gene polymorphism in cardiovascular disease. Meta-analyses of small and large studies in whites. Arterioscler Thromb Vasc Biol 2000;20:484-92 **(ID: 1,2,3)**
2. Arranz MJ, Munro J, Sham P, Kirov G, Murray RM, Collier DA, Kerwin RW. Meta-analysis of studies on genetic variation in 5-HT2A receptors and clozapine response. Schizophrenia Res 1998;32:93-9 **(ID: 4,5)**
3. Brattstrom L, Wilcken DEL, Ohrvik J, Brudin L. Common methylenetetrahydrofolate reductase gene mutation leads to hyperhomocysteinemia but not to vascular disease. Circulation 1998;98:2520-6 **(ID: 6)**
4. Christensen PM, Gotzsche PC, Brosen K. The sparteine/debrisoquine (CYP2D6) oxidation polymorphism and the risk of lung cancer: a meta-analysis. Eur J Clin Pharmacol 1997;51:389-93 **(ID: 7)**
5. Deb S, Braganza J, Norton N, Williams H, Kehoe PG, Williams J, Owen MJ. APOE å4 influences the manifestation of Alzheimer's disease in adults with Down's syndrome. Br J Psychiatry 2000;176:468-72 **(ID: 8)**
6. Dubertret C, Gorwood P, Ades J, Feingold J, Schwartz JC, Sokoloff P. Meta-analysis of DRD3 gene and schizophrenia: Ethnic heterogeneity and significant association in Caucasians. Am J Med Genet 1998;81:318-22 **(ID: 9)**
7. Furlong RA, Ho L, Rubinnsztein JS, Walsh C, Paykel ES, Rubinsztein DC. Analysis of the monoamine oxidase A (MAOA) gene in bipolar affective disorder by association studies, meta-analyses, and sequencing of the promoter. Am J Med Genet 1999;88:398-406 **(ID: 10,11)**
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12. Iacoviello L, Burzotta F, Di Castelnuovo A, Zito F, Marchioli R, Donati MB. The 4G/5G polymorphism of PAI-1 promoter gene and the risk of myocardial infraction: a meta-analysis. Thromb Heamost 1998;80:1029-30 **(ID: 18)**
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14. Kato N, Sugiyama T, Morita H, Kurihara H, Yamori Y, Yazaki Y. Angiotensinogen gene and essential hypertension in the Japanese: extensive

association study and meta-analysis on six reported studies. J Hypertens 1999;17:757-63 **(ID: 20)**
15. Krontiris TG, Devlin B, Karp DD, Robert NJ, Risch N. An association between the risk of cancer and mutations in the HRAS1 minisatellite locus. N Engl J Med 1993;329;517-23 **(ID: 21)**
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17. Marcus PM, Vineis P, Rothman N. NAT2 slow acetylation and bladder cancer risk: a meta-analysis of 22 case-control studies conducted in the general population. Pharmacogenetics 2000;10:115-22 **(ID: 23)**
18. McCarron MO, Delong D, Alberts MJ. APOE genotype as a risk factor for ischemic cerebrovascular disease: a meta-analysis. Neurology 1999;53:1308-11 **(ID: 24)**
19. Mitchell LE. Transforming growth factor á locus and nonsyndromic cleft lip with or without cleft palate: a reappraisal. Genet Epidemiol 1996;14:231-40 **(ID: 25)**
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23. van der Put NMJ, Eskes TKAB, Blom HJ. Is the common 677C-T mutation in the methylenetetrahydrofolate reductase gene a risk factor for neural tube defects? A meta-analysis. QJM;90:111-5 **(ID: 29,30,31)**
24. Wilson PWF, Schaefer EJ, Larson MG, Ordovas JM. Apolipoprotein E alleles and risk of coronary heart disease: a meta-analysis. Arterioscler Thromb Vasc Biol 1996;16:1250-5 **(ID: 32)**
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B. First studies for 27 genetic associations where there was a single first study (for 9 genetic associations multiple "first" studies were published in the same year at different journals) **(ID number corresponds to table 1 of the main paper)**

1. Cambien F, Poirier O, Lecerf L, Evans A, Cambou JP, Arveiler D, Luc G, Bard JM, Bara L, Ricard S, et al. Deletion polymorphism in the gene for angiotensin-converting enzyme is a potent risk for myocardial infraction. Nature 1992;359:641-4 **(ID 1)**
2. Miettinen HE, Korpela K, Hamalainen L, Kontula K. Polymorphisms of the apolipoprotein and angiotensin-converting enzyme genes in young North

Karelian patients with coronary heart disease. Hum Genet 1994;94:189-92 **(ID 2)**
3. Arranz MJ, Collier D, Sodhi M, Ball D, Roberts G, Price J, Sham P, Kerwin R Association between clozapine response and allelic variation in 5-HT2A receptor gene. Lancet 1995;346:281-2 **((ID 4,5)**
4. Ayesh R, Idle JR, Ritchie JC, Crothers MJ, Hetzel MR. Metabolic oxidation phenotypes as markers of susceptibility to lung cancer. Nature 1984;312:169-70 **(ID 7)**
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6. Crocq MA, Mant R, Asherson P, Williams J, Hode Y, Mayerova A, Collier D, Lannfelt L, Sokoloff P, Schwartz JC, et al. Association between schizophrenia and homozygosity at the dopamine D3 receptor gene. J Med Genet 1992;29:858-60 **(ID 9)**
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14. Armstrong M, Daly AK, Cholerton S, Bateman DN, Idle JR. Mutant debrisoquine hydroxylation genes in Parkinson's disease. Lancet 1992;339:1017-8 **(ID 19)**
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C. Complete list of 370 genetic association studies included in the 36 meta-analyses (ID numbers correspond to table 1) (Some articles include data for more than one meta-analysis).

ID1

1. Cambien F, Poirier O, Lecerf L, Evans A, Cambou JP, Arveiler D, Luc G, Bard JM, Bara L, Ricard S, et al. Deletion polymorphism in the gene for angiotensin-converting enzyme is a potent risk for myocardial infraction. Nature 1992;359:641-4
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presenting with myocardial infraction or unstable angina. J Hum Hypertens 1994;8:635-8.
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ID2

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populations and relevance to myocardial infraction and coronary artery disease. Arterioscler Thromb Vasc Biol 1996;16:115-119.
13. Corbo RM, Vilardo T, Mantuano E, Ruggeri M, Gemma AT, Scacchi R. Apolipoproteins B, and E, and angiotensin I-converting enzyme (ACE) genetic polymorphisms in Italian women with coronary artery disease (CAD) and their relationships with plasma lipid and apolipoprotein levels. Clin Genet 1997;52:77-82.
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