# Stephen E Humphries

## List of Publications by Year in Descending Order



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| 162 papers | 19,432 citations | 51 h-index | 139 g-index |
| 171 ext. papers | 23,779 ext. citations | 7.6 avg, IF | 5.58 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 162 | LDL-C Concentrations and the 12-SNP LDL-C Score for Polygenic Hypercholesterolaemia in Self-Reported South Asian, Black and Caribbean Participants of the UK Biobank.. *Frontiers in Genetics*, **2022**, 13, 845498 | 4.5 | 2 |
| 161 | Inborn Errors of Lipoprotein Metabolism Presenting in Childhood **2022**, 677-691 | | |
| 160 | The Clinical Genome Resource (ClinGen) Familial Hypercholesterolemia Variant Curation Expert Panel consensus guidelines for LDLR variant classification.. *Genetics in Medicine*, **2021**, | 8.1 | 6 |
| 159 | Genetic testing for Familial Hypercholesterolaemia - Past, Present and Future. *Journal of Lipid Research*, **2021**, 100139 | 6.3 | 1 |
| 158 | Intake of food rich in saturated fat in relation to subclinical atherosclerosis and potential modulating effects from single genetic variants. *Scientific Reports*, **2021**, 11, 7866 | 4.9 | 0 |
| 157 | Alcohol consumption in relation to carotid subclinical atherosclerosis and its progression: results from a European longitudinal multicentre study. *European Journal of Nutrition*, **2021**, 60, 123-134 | 5.2 | 1 |
| 156 | The overlap of genetic susceptibility to schizophrenia and cardiometabolic disease can be used to identify metabolically different groups of individuals. *Scientific Reports*, **2021**, 11, 632 | 4.9 | 4 |
| 155 | Comparison of the mutation spectrum and association with pre and post treatment lipid measures of children with heterozygous familial hypercholesterolaemia (FH) from eight European countries. *Atherosclerosis*, **2021**, 319, 108-117 | 3.1 | 5 |
| 154 | Case-finding and genetic testing for familial hypercholesterolaemia in primary care. *Heart*, **2021**, 107, 1956-1961 | 5.1 | 1 |
| 153 | Analysis of the genetic variants associated with circulating levels of sgp130. Results from the IMPROVE study. *Genes and Immunity*, **2020**, 21, 100-108 | 4.4 | 3 |
| 152 | Higher Responsiveness to Rosuvastatin in Polygenic versus Monogenic Hypercholesterolaemia: A Propensity Score Analysis. *Life*, **2020**, 10, | 3 | 4 |
| 151 | The familial hypercholesterolaemia phenotype: Monogenic familial hypercholesterolaemia, polygenic hypercholesterolaemia and other causes. *Clinical Genetics*, **2020**, 97, 457-466 | 4 | 15 |
| 150 | Comparison of the characteristics at diagnosis and treatment of children with heterozygous familial hypercholesterolaemia (FH) from eight European countries. *Atherosclerosis*, **2020**, 292, 178-187 | 3.1 | 17 |
| 149 | PCSK6 Is a Key Protease in the Control of Smooth Muscle Cell Function in Vascular Remodeling. *Circulation Research*, **2020**, 126, 571-585 | 15.7 | 12 |
| 148 | Non-HDL or LDL cholesterol in heterozygous familial hypercholesterolaemia: findings of the Simon Broome Register. *Current Opinion in Lipidology*, **2020**, 31, 167-175 | 4.4 | 2 |
| 147 | Sex differences in cardiovascular morbidity associated with familial hypercholesterolaemia: A retrospective cohort study of the UK Simon Broome register linked to national hospital records. *Atherosclerosis*, **2020**, 315, 131-137 | 3.1 | 4 |
| 146 | Management of familial hypercholesterolaemia in childhood. *Current Opinion in Pediatrics*, **2020**, 32, 633-640 | 3.2 | 1 |

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| 145 | Current management of children and young people with heterozygous familial hypercholesterolaemia - HEART UK statement of care. *Atherosclerosis*, **2019**, 290, 1-8 | 3.1 | 32 |
| 144 | Risk of cardiovascular disease outcomes in primary care subjects with familial hypercholesterolaemia: A cohort study. *Atherosclerosis*, **2019**, 287, 8-15 | 3.1 | 17 |
| 143 | Genetic variation in CADM2 as a link between psychological traits and obesity. *Scientific Reports*, **2019**, 9, 7339 | 4.9 | 18 |
| 142 | Polygenic Hypercholesterolemia and Cardiovascular Disease Risk. *Current Cardiology Reports*, **2019**, 21, 43 | 4.2 | 21 |
| 141 | Data on the association between a simplified Mediterranean diet score and the incidence of combined, cardio and cerebro vascular events. *Data in Brief*, **2019**, 23, 103789 | 1.2 | |
| 140 | Estimation of the prevalence of cholesteryl ester storage disorder in a cohort of patients with clinical features of familial hypercholesterolaemia. *Annals of Clinical Biochemistry*, **2019**, 56, 112-117 | 2.2 | 3 |
| 139 | Phenome-wide association analysis of LDL-cholesterol lowering genetic variants in PCSK9. *BMC Cardiovascular Disorders*, **2019**, 19, 240 | 2.3 | 8 |
| 138 | Familial hypercholesterolaemia: what's new?. *Paediatrics and Child Health (United Kingdom)*, **2019**, 29, 127-136 | 0.6 | 0 |
| 137 | Statins for children with familial hypercholesterolemia. *The Cochrane Library*, **2019**, 2019, | 5.2 | 19 |
| 136 | Coronary heart disease mortality in severe vs. non-severe familial hypercholesterolaemia in the Simon Broome Register. *Atherosclerosis*, **2019**, 281, 207-212 | 3.1 | 23 |
| 135 | A priori-defined Mediterranean-like dietary pattern predicts cardiovascular events better in north Europe than in Mediterranean countries. *International Journal of Cardiology*, **2019**, 282, 88-92 | 3.2 | 7 |
| 134 | Effect of Coronary Artery Disease risk SNPs on serum cytokine levels and cytokine imbalance in Premature Coronary Artery Disease. *Cytokine*, **2019**, 122, 154060 | 4 | 6 |
| 133 | Association of lifelong occupation and educational level with subclinical atherosclerosis in different European regions. Results from the IMPROVE study. *Atherosclerosis*, **2018**, 269, 129-137 | 3.1 | 5 |
| 132 | Universal screening at age 1-2 years as an adjunct to cascade testing for familial hypercholesterolaemia in the UK: A cost-utility analysis. *Atherosclerosis*, **2018**, 275, 434-443 | 3.1 | 22 |
| 131 | Association of circulating metabolites with healthy diet and risk of cardiovascular disease: analysis of two cohort studies. *Scientific Reports*, **2018**, 8, 8620 | 4.9 | 32 |
| 130 | Genetic Association of Lipids and Lipid Drug Targets With Abdominal Aortic Aneurysm: A Meta-analysis. *JAMA Cardiology*, **2018**, 3, 26-33 | 16.2 | 44 |
| 129 | The UK Paediatric Familial Hypercholesterolaemia Register: Statin-related safety and 1-year growth data. *Journal of Clinical Lipidology*, **2018**, 12, 25-32 | 4.9 | 30 |
| 128 | Increased Levels of Circulating Fatty Acids Are Associated with Protective Effects against Future Cardiovascular Events in Nondiabetics. *Journal of Proteome Research*, **2018**, 17, 870-878 | 5.6 | 9 |

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| 127 | GWAS and colocalization analyses implicate carotid intima-media thickness and carotid plaque loci in cardiovascular outcomes. *Nature Communications*, **2018**, 9, 5141 | 17.4 | 64 |
| 126 | Clinical utility of the polygenic LDL-C SNP score in familial hypercholesterolemia. *Atherosclerosis*, **2018**, 277, 457-463 | 3.1 | 28 |
| 125 | ClinVar database of global familial hypercholesterolemia-associated DNA variants. *Human Mutation* , **2018**, 39, 1631-1640 | 4.7 | 55 |
| 124 | Cost-utility analysis of searching electronic health records and cascade testing to identify and diagnose familial hypercholesterolaemia in England and Wales. *Atherosclerosis*, **2018**, 275, 80-87 | 3.1 | 7 |
| 123 | Influence of cytokine gene polymorphisms on proinflammatory/anti-inflammatory cytokine imbalance in premature coronary artery disease. *Postgraduate Medical Journal*, **2017**, 93, 209-214 | 2 | 9 |
| 122 | Genetics of CHD in 2016: Common and rare genetic variants and risk of CHD. *Nature Reviews Cardiology*, **2017**, 14, 73-74 | 14.8 | 7 |
| 121 | Molecular genetics of familial hypercholesterolemia in Israel-revisited. *Atherosclerosis*, **2017**, 257, 55-63 | 3.1 | 16 |
| 120 | Common variants in the genes of triglyceride and HDL-C metabolism lack association with coronary artery disease in the Pakistani subjects. *Lipids in Health and Disease*, **2017**, 16, 24 | 4.4 | 12 |
| 119 | Genetic risk analysis of coronary artery disease in Pakistani subjects using a genetic risk score of 21 variants. *Atherosclerosis*, **2017**, 258, 1-7 | 3.1 | 17 |
| 118 | Genetic Architecture of Familial Hypercholesterolaemia. *Current Cardiology Reports*, **2017**, 19, 44 | 4.2 | 57 |
| 117 | Causal Associations of Adiposity and Body Fat Distribution With Coronary Heart Disease, Stroke Subtypes, and Type 2 Diabetes Mellitus: A Mendelian Randomization Analysis. *Circulation*, **2017**, 135, 2373-2388 | 16.7 | 182 |
| 116 | Identifying low density lipoprotein cholesterol associated variants in the Annexin A2 (ANXA2) gene. *Atherosclerosis*, **2017**, 261, 60-68 | 3.1 | 8 |
| 115 | PCSK9 genetic variants and risk of type 2 diabetes: a mendelian randomisation study. *Lancet Diabetes and Endocrinology,the*, **2017**, 5, 97-105 | 18.1 | 225 |
| 114 | Carotid plaque-thickness and common carotid IMT show additive value in cardiovascular risk prediction and reclassification. *Atherosclerosis*, **2017**, 263, 412-419 | 3.1 | 39 |
| 113 | Greater preclinical atherosclerosis in treated monogenic familial hypercholesterolemia vs. polygenic hypercholesterolemia. *Atherosclerosis*, **2017**, 263, 405-411 | 3.1 | 47 |
| 112 | An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. *Diabetes*, **2017**, 66, 2888-2902 | 8.2 | 414 |
| 111 | Screening for familial hypercholesterolaemia in childhood: Avon Longitudinal Study of Parents and Children (ALSPAC). *Atherosclerosis*, **2017**, 260, 47-55 | 3.1 | 11 |
| 110 | Cost effectiveness of cascade testing for familial hypercholesterolaemia, based on data from familial hypercholesterolaemia services in the UK. *European Heart Journal*, **2017**, 38, 1832-1839 | 9.5 | 58 |

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| 109 | Applicability of the low-density lipoprotein cholesterol gene score in a South European population. *Atherosclerosis*, **2017**, 263, e99-e100 | 3.1 | 2 |
| 108 | How close are we to implementing a genetic risk score for coronary heart disease?. *Expert Review of Molecular Diagnostics*, **2017**, 17, 905-915 | 3.8 | 6 |
| 107 | The UK Paediatric Familial Hypercholesterolaemia Register: preliminary data. *Archives of Disease in Childhood*, **2017**, 102, 255-260 | 2.2 | 29 |
| 106 | Plasma Concentrations of Afamin Are Associated With Prevalent and Incident Type 2 Diabetes: A Pooled Analysis in More Than 20,000 Individuals. *Diabetes Care*, **2017**, 40, 1386-1393 | 14.6 | 39 |
| 105 | Statins for children with familial hypercholesterolemia. *The Cochrane Library*, **2017**, 7, CD006401 | 5.2 | 27 |
| 104 | The UCL low-density lipoprotein receptor gene variant database: pathogenicity update. *Journal of Medical Genetics*, **2017**, 54, 217-223 | 5.8 | 60 |
| 103 | Functional Analysis of the Coronary Heart Disease Risk Locus on Chromosome 21q22. *Disease Markers*, **2017**, 2017, 1096916 | 3.2 | 4 |
| 102 | Mapping of 79 loci for 83 plasma protein biomarkers in cardiovascular disease. *PLoS Genetics*, **2017**, 13, e1006706 | 6 | 102 |
| 101 | Identification of a novel proinsulin-associated SNP and demonstration that proinsulin is unlikely to be a causal factor in subclinical vascular remodelling using Mendelian randomisation. *Atherosclerosis*, **2017**, 266, 196-204 | 3.1 | 2 |
| 100 | Variant rs10911021 that associates with coronary heart disease in type 2 diabetes, is associated with lower concentrations of circulating HDL cholesterol and large HDL particles but not with amino acids. *Cardiovascular Diabetology*, **2016**, 15, 115 | 8.7 | 9 |
| 99 | Mitochondrial uncoupling proteins regulate angiotensin-converting enzyme expression: crosstalk between cellular and endocrine metabolic regulators suggested by RNA interference and genetic studies. *BioEssays*, **2016**, 38 Suppl 1, S107-18 | 4.1 | 7 |
| 98 | A 19-SNP coronary heart disease gene score profile in subjects with type 2 diabetes: the coronary heart disease risk in type 2 diabetes (CoRDia study) study baseline characteristics. *Cardiovascular Diabetology*, **2016**, 15, 141 | 8.7 | 6 |
| 97 | Defining severe familial hypercholesterolaemia and the implications for clinical management: a consensus statement from the International Atherosclerosis Society Severe Familial Hypercholesterolemia Panel. *Lancet Diabetes and Endocrinology,the*, **2016**, 4, 850-61 | 18.1 | 215 |
| 96 | Post-GWAS methodologies for localisation of functional non-coding variants: ANGPTL3. *Atherosclerosis*, **2016**, 246, 193-201 | 3.1 | 10 |
| 95 | Plasma urate concentration and risk of coronary heart disease: a Mendelian randomisation analysis. *Lancet Diabetes and Endocrinology,the*, **2016**, 4, 327-36 | 18.1 | 100 |
| 94 | Influence of Genetic Risk Factors on Coronary Heart Disease Occurrence in Afro-Caribbeans. *Canadian Journal of Cardiology*, **2016**, 32, 978-85 | 3.8 | 18 |
| 93 | Effect of six type II diabetes susceptibility loci and an FTO variant on obesity in Pakistani subjects. *European Journal of Human Genetics*, **2016**, 24, 903-10 | 5.3 | 22 |
| 92 | Circulating Apolipoprotein E Concentration and Cardiovascular Disease Risk: Meta-analysis of Results from Three Studies. *PLoS Medicine*, **2016**, 13, e1002146 | 11.6 | 27 |

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| 91 | Identification of the Functional Variant(s) that Explain the Low-Density Lipoprotein Receptor (LDLR) GWAS SNP rs6511720 Association with Lower LDL-C and Risk of CHD. *PLoS ONE*, **2016**, 11, e0167676 | 3.7 | 17 |
| 90 | Genome-Wide DNA Methylation in Mixed Ancestry Individuals with Diabetes and Prediabetes from South Africa. *International Journal of Endocrinology*, **2016**, 2016, 3172093 | 2.7 | 9 |
| 89 | Variants Within TSC2 Exons 25 and 31 Are Very Unlikely to Cause Clinically Diagnosable Tuberous Sclerosis. *Human Mutation*, **2016**, 37, 364-70 | 4.7 | 14 |
| 88 | Cardiovascular risk stratification in familial hypercholesterolaemia. *Heart*, **2016**, 102, 1003-8 | 5.1 | 38 |
| 87 | Marginal role for 53 common genetic variants in cardiovascular disease prediction. *Heart*, **2016**, 102, 1640-7 | 5.1 | 23 |
| 86 | The genetic spectrum of familial hypercholesterolemia in south-eastern Poland. *Metabolism: Clinical and Experimental*, **2016**, 65, 48-53 | 12.7 | 20 |
| 85 | Telomere length, antioxidant status and incidence of ischaemic heart disease in type 2 diabetes. *International Journal of Cardiology*, **2016**, 216, 159-64 | 3.2 | 19 |
| 84 | Genetic loci on chromosome 5 are associated with circulating levels of interleukin-5 and eosinophil count in a European population with high risk for cardiovascular disease. *Cytokine*, **2016**, 81, 1-9 | 4 | 11 |
| 83 | Soluble CD93 Is Involved in Metabolic Dysregulation but Does Not Influence Carotid Intima-Media Thickness. *Diabetes*, **2016**, 65, 2888-99 | 0.9 | 6 |
| 82 | Association of Lipid Fractions With Risks for Coronary Artery Disease and Diabetes. *JAMA Cardiology*, **2016**, 1, 692-9 | 16.2 | 168 |
| 81 | Mitochondrial uncoupling proteins regulate angiotensin-converting enzyme expression: crosstalk between cellular and endocrine metabolic regulators suggested by RNA interference and genetic studies. *Inside the Cell*, **2016**, 1, 70-81 | | 2 |
| 80 | Improving detection of familial hypercholesterolaemia in primary care using electronic audit and nurse-led clinics. *Journal of Evaluation in Clinical Practice*, **2016**, 22, 341-8 | 2.5 | 20 |
| 79 | Improving identification of familial hypercholesterolaemia in primary care: derivation and validation of the familial hypercholesterolaemia case ascertainment tool (FAMCAT). *Atherosclerosis*, **2015**, 238, 336-43 | 3.1 | 65 |
| 78 | A genome-wide association study identifies multiple loci for variation in human ear morphology. *Nature Communications*, **2015**, 6, 7500 | 17.4 | 42 |
| 77 | Mendelian randomization of blood lipids for coronary heart disease. *European Heart Journal*, **2015**, 36, 539-50 | 9.5 | 417 |
| 76 | Comparison of coronary heart disease genetic assessment with conventional cardiovascular risk assessment in primary care: reflections on a feasibility study. *Primary Health Care Research and Development*, **2015**, 16, 607-17 | 1.6 | 1 |
| 75 | Familial hypercholesterolaemia in children and adolescents: gaining decades of life by optimizing detection and treatment. *European Heart Journal*, **2015**, 36, 2425-37 | 9.5 | 430 |
| 74 | Sex-Specific Effects of Adiponectin on Carotid Intima-Media Thickness and Incident Cardiovascular Disease. *Journal of the American Heart Association*, **2015**, 4, e001853 | 6 | 25 |

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| 73 | Functional Analysis of a Carotid Intima-Media Thickness Locus Implicates BCAR1 and Suggests a Causal Variant. *Circulation: Cardiovascular Genetics*, **2015**, 8, 696-706 | | 12 |
| 72 | The UK10K project identifies rare variants in health and disease. *Nature*, **2015**, 526, 82-90 | 50.4 | 776 |
| 71 | Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. *Nature Genetics*, **2015**, 47, 1415-25 | 36.3 | 292 |
| 70 | Functional analysis of four LDLR 5'UTR and promoter variants in patients with familial hypercholesterolaemia. *European Journal of Human Genetics*, **2015**, 23, 790-5 | 5.3 | 17 |
| 69 | Sixty-five common genetic variants and prediction of type 2 diabetes. *Diabetes*, **2015**, 64, 1830-40 | 0.9 | 76 |
| 68 | Refinement of variant selection for the LDL cholesterol genetic risk score in the diagnosis of the polygenic form of clinical familial hypercholesterolemia and replication in samples from 6 countries. *Clinical Chemistry*, **2015**, 61, 231-8 | 5.5 | 130 |
| 67 | Identifying genetic risk variants for coronary heart disease in familial hypercholesterolemia: an extreme genetics approach. *European Journal of Human Genetics*, **2015**, 23, 381-7 | 5.3 | 10 |
| 66 | HMG-coenzyme A reductase inhibition, type 2 diabetes, and bodyweight: evidence from genetic analysis and randomised trials. *Lancet, The*, **2015**, 385, 351-61 | 40 | 409 |
| 65 | Effectiveness of a self-management intervention with personalised genetic and lifestyle-related risk information on coronary heart disease and diabetes-related risk in type 2 diabetes (CoRDia): study protocol for a randomised controlled trial. *Trials*, **2015**, 16, 547 | 2.8 | 8 |
| 64 | Demonstration of the presence of the "deleted" MIR122 gene in HepG2 cells. *PLoS ONE*, **2015**, 10, e0122471 | 3.7 | 5 |
| 63 | Networks in Coronary Heart Disease Genetics As a Step towards Systems Epidemiology. *PLoS ONE*, **2015**, 10, e0125876 | 3.7 | 14 |
| 62 | Clinical Utility of a Coronary Heart Disease Risk Prediction Gene Score in UK Healthy Middle Aged Men and in the Pakistani Population. *PLoS ONE*, **2015**, 10, e0130754 | 3.7 | 18 |
| 61 | Effect of the PPARG2 Pro12Ala Polymorphism on Associations of Physical Activity and Sedentary Time with Markers of Insulin Sensitivity in Those with an Elevated Risk of Type 2 Diabetes. *PLoS ONE*, **2015**, 10, e0124062 | 3.7 | 9 |
| 60 | New genetic loci link adipose and insulin biology to body fat distribution. *Nature*, **2015**, 518, 187-196 | 50.4 | 920 |
| 59 | Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206 | 50.4 | 2687 |
| 58 | PLA2G10 Gene Variants, sPLA2 Activity, and Coronary Heart Disease Risk. *Circulation: Cardiovascular Genetics*, **2015**, 8, 356-62 | | 15 |
| 57 | GWAS-identified loci for coronary heart disease are associated with intima-media thickness and plaque presence at the carotid artery bulb. *Atherosclerosis*, **2015**, 239, 304-10 | 3.1 | 26 |
| 56 | Plasma IL-5 concentration and subclinical carotid atherosclerosis. *Atherosclerosis*, **2015**, 239, 125-30 | 3.1 | 29 |

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| 55 | Analysis of the role of interleukin 6 receptor haplotypes in the regulation of circulating levels of inflammatory biomarkers and risk of coronary heart disease. *PLoS ONE*, **2015**, 10, e0119980 | 3.7 | 17 |
| 54 | Plasma autoantibodies against apolipoprotein B-100 peptide 210 in subclinical atherosclerosis. *Atherosclerosis*, **2014**, 232, 242-8 | 3.1 | 22 |
| 53 | Low levels of IgM antibodies against phosphorylcholine are associated with fast carotid intima media thickness progression and cardiovascular risk in men. *Atherosclerosis*, **2014**, 236, 394-9 | 3.1 | 19 |
| 52 | A systematic review and meta-analysis of 130,000 individuals shows smoking does not modify the association of APOE genotype on risk of coronary heart disease. *Atherosclerosis*, **2014**, 237, 5-12 | 3.1 | 24 |
| 51 | A rare variant in APOC3 is associated with plasma triglyceride and VLDL levels in Europeans. *Nature Communications*, **2014**, 5, 4871 | 17.4 | 46 |
| 50 | Defining the role of common variation in the genomic and biological architecture of adult human height. *Nature Genetics*, **2014**, 46, 1173-86 | 36.3 | 1339 |
| 49 | A serum 25-hydroxyvitamin D concentration-associated genetic variant in DHCR7 interacts with type 2 diabetes status to influence subclinical atherosclerosis (measured by carotid intima-media thickness). *Diabetologia*, **2014**, 57, 1159-72 | 10.3 | 24 |
| 48 | Association between alcohol and cardiovascular disease: Mendelian randomisation analysis based on individual participant data. *BMJ, The*, **2014**, 349, g4164 | 5.9 | 406 |
| 47 | Whole exome sequencing of familial hypercholesterolaemia patients negative for LDLR/APOB/PCSK9 mutations. *Journal of Medical Genetics*, **2014**, 51, 537-44 | 5.8 | 77 |
| 46 | Association of vitamin D status with arterial blood pressure and hypertension risk: a mendelian randomisation study. *Lancet Diabetes and Endocrinology,the*, **2014**, 2, 719-29 | 18.1 | 250 |
| 45 | The polygenic nature of hypertriglyceridaemia: implications for definition, diagnosis, and management. *Lancet Diabetes and Endocrinology,the*, **2014**, 2, 655-66 | 18.1 | 357 |
| 44 | Human genetic evidence for involvement of CD137 in atherosclerosis. *Molecular Medicine*, **2014**, 20, 456-65 | 6.2 | 8 |
| 43 | Clinical utility gene card for: hyperlipoproteinemia, TYPE II. *European Journal of Human Genetics*, **2014**, 22, | 5.3 | 7 |
| 42 | The genetic architecture of the familial hyperlipidaemia syndromes: rare mutations and common variants in multiple genes. *Current Opinion in Lipidology*, **2014**, 25, 274-81 | 4.4 | 24 |
| 41 | Common genetic determinants of lung function, subclinical atherosclerosis and risk of coronary artery disease. *PLoS ONE*, **2014**, 9, e104082 | 3.7 | 30 |
| 40 | Association of TLL1 gene polymorphism (rs1503298, T > C) with coronary heart disease in PREDICT, UDACS and ED cohorts. *Journal of the College of Physicians and Surgeons--Pakistan: JCPSP*, **2014**, 24, 615-9 | 0.7 | 2 |
| 39 | Statin treatment of children with familial hypercholesterolemia--trying to balance incomplete evidence of long-term safety and clinical accountability: are we approaching a consensus?. *Atherosclerosis*, **2013**, 226, 315-20 | 3.1 | 48 |
| 38 | Analysis of the frequency and spectrum of mutations recognised to cause familial hypercholesterolaemia in routine clinical practice in a UK specialist hospital lipid clinic. *Atherosclerosis*, **2013**, 229, 161-8 | 3.1 | 65 |

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| 37 | Discovery and refinement of loci associated with lipid levels. *Nature Genetics*, **2013**, 45, 1274-1283 | 36.3 | 1904 |
| 36 | Secretory phospholipase A(2)-IIA and cardiovascular disease: a mendelian randomization study. *Journal of the American College of Cardiology*, **2013**, 62, 1966-1976 | 15.1 | 91 |
| 35 | Use of low-density lipoprotein cholesterol gene score to distinguish patients with polygenic and monogenic familial hypercholesterolaemia: a case-control study. *Lancet, The*, **2013**, 381, 1293-301 | 40 | 376 |
| 34 | Familial hypercholesterolaemia is underdiagnosed and undertreated in the general population: guidance for clinicians to prevent coronary heart disease: consensus statement of the European Atherosclerosis Society. *European Heart Journal*, **2013**, 34, 3478-90a | 9.5 | 1551 |
| 33 | Population genomics of cardiometabolic traits: design of the University College London-London School of Hygiene and Tropical Medicine-Edinburgh-Bristol (UCLEB) Consortium. *PLoS ONE*, **2013**, 8, e71345 | 3.7 | 33 |
| 32 | Association of TERC and OBFC1 haplotypes with mean leukocyte telomere length and risk for coronary heart disease. *PLoS ONE*, **2013**, 8, e83122 | 3.7 | 34 |
| 31 | The interleukin-6 receptor as a target for prevention of coronary heart disease: a mendelian randomisation analysis. *Lancet, The*, **2012**, 379, 1214-24 | 40 | 658 |
| 30 | Paraoxonase-1 Is Not Associated with Coronary Artery Calcification in Type 2 Diabetes: Results from the PREDICT Study. *Disease Markers*, **2012**, 33, 101-112 | 3.2 | 10 |
| 29 | Cochrane Review: Statins for children with familial hypercholesterolemia. *Evidence-Based Child Health: A Cochrane Review Journal*, **2011**, 6, 1086-1129 | | 1 |
| 28 | Coronary heart disease risk prediction in the era of genome-wide association studies: current status and what the future holds. *Circulation*, **2010**, 121, 2235-48 | 16.7 | 49 |
| 27 | Cross-sectional analysis of baseline data to identify the major determinants of carotid intima-media thickness in a European population: the IMPROVE study. *European Heart Journal*, **2010**, 31, 614-22 | 9.5 | 99 |
| 26 | Identification and management of familial hypercholesterolaemia: what does it mean to primary care?. *British Journal of General Practice*, **2009**, 59, 773-6 | 1.6 | 48 |
| 25 | Reductions in all-cause, cancer, and coronary mortality in statin-treated patients with heterozygous familial hypercholesterolaemia: a prospective registry study. *European Heart Journal*, **2008**, 29, 2625-33 | 9.5 | 321 |
| 24 | Development of sensitive and specific age- and gender-specific low-density lipoprotein cholesterol cutoffs for diagnosis of first-degree relatives with familial hypercholesterolaemia in cascade testing. *Clinical Chemistry and Laboratory Medicine*, **2008**, 46, 791-803 | 5.9 | 109 |
| 23 | Candidate gene genotypes, along with conventional risk factor assessment, improve estimation of coronary heart disease risk in healthy UK men. *Clinical Chemistry*, **2007**, 53, 8-16 | 5.5 | 74 |
| 22 | Mutational analysis in UK patients with a clinical diagnosis of familial hypercholesterolaemia: relationship with plasma lipid traits, heart disease risk and utility in relative tracing. *Journal of Molecular Medicine*, **2006**, 84, 203-14 | 5.5 | 55 |
| 21 | Common variants in the TCF7L2 gene and predisposition to type 2 diabetes in UK European Whites, Indian Asians and Afro-Caribbean men and women. *Journal of Molecular Medicine*, **2006**, 84, 1005-14 | 5.5 | 112 |
| 20 | Insight into the nature of the CRP-coronary event association using Mendelian randomization. *International Journal of Epidemiology*, **2006**, 35, 922-31 | 7.8 | 137 |

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| 19 | Genetic risk factors for stroke and carotid atherosclerosis: insights into pathophysiology from candidate gene approaches. *Lancet Neurology, The*, **2004**, 3, 227-35 | 24.1 | 179 |
| 18 | Psychological impact of genetic testing for familial hypercholesterolemia within a previously aware population: a randomized controlled trial. *American Journal of Medical Genetics Part A*, **2004**, 128A, 285-93 | | 148 |
| 17 | Genetic causes of monogenic heterozygous familial hypercholesterolemia: a HuGE prevalence review. *American Journal of Epidemiology*, **2004**, 160, 407-20 | 3.8 | 406 |
| 16 | A review on the diagnosis, natural history, and treatment of familial hypercholesterolaemia. *Atherosclerosis*, **2003**, 168, 1-14 | 3.1 | 426 |
| 15 | Cost effectiveness analysis of different approaches of screening for familial hypercholesterolaemia. *BMJ, The*, **2002**, 324, 1303 | 5.9 | 146 |
| 14 | Angiotensin-I converting enzyme genotype-dependent benefit from hormone replacement therapy in isometric muscle strength and bone mineral density. *Journal of Clinical Endocrinology and Metabolism*, **2001**, 86, 2200-4 | 5.6 | 46 |
| 13 | Epidemiological and genetic associations of activated factor XII concentration with factor VII activity, fibrinopeptide A concentration, and risk of coronary heart disease in men. *Circulation*, **2000** , 102, 2058-62 | 16.7 | 71 |
| 12 | Linkage of the cholesteryl ester transfer protein (CETP) gene to LDL particle size: use of a novel tetranucleotide repeat within the CETP promoter. *Circulation*, **2000**, 101, 2461-6 | 16.7 | 62 |
| 11 | ApoCIII gene variants modulate postprandial response to both glucose and fat tolerance tests. *Circulation*, **1999**, 99, 1872-7 | 16.7 | 77 |
| 10 | Lipoprotein lipase gene variation is associated with a paternal history of premature coronary artery disease and fasting and postprandial plasma triglycerides: the European Atherosclerosis Research Study (EARS). *Arteriosclerosis, Thrombosis, and Vascular Biology*, **1998**, 18, 526-34 | 9.4 | 128 |
| 9 | Simplified detection of a mutation causing familial hypercholesterolaemia throughout Britain: evidence for an origin in a common distant ancestor. *Annals of Clinical Biochemistry*, **1998**, 35 ( Pt 2), 226-35 | 2.2 | 1 |
| 8 | Common founder mutation in the LDL receptor gene causing familial hypercholesterolaemia in the Icelandic population. *Human Mutation*, **1997**, 10, 36-44 | 4.7 | 30 |
| 7 | Familial lipoprotein lipase (LPL) deficiency: a catalogue of LPL gene mutations identified in 20 patients from the UK, Sweden, and Italy. *Human Mutation*, **1997**, 10, 465-73 | 4.7 | 36 |
| 6 | The use of a highly informative CA repeat polymorphism within the abetalipoproteinaemia locus (4q22-24). *Prenatal Diagnosis*, **1997**, 17, 1181-6 | 3.2 | 10 |
| 5 | Lipoprotein lipase variants D9N and N291S are associated with increased plasma triglyceride and lower high-density lipoprotein cholesterol concentrations: studies in the fasting and postprandial states: the European Atherosclerosis Research Studies. *Circulation*, **1997**, 96, 733-40 | 16.7 | 74 |
| 4 | The 10-Base-Pair Insertion in the Promoter of the Factor VII Gene Is not Associated with Lower Levels of Factor VI Ic in Afrocarribeans. *Thrombosis and Haemostasis*, **1997**, 77, 213-214 | 7 | 2 |
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