

## Suggested Reading List

- 1 Berglund L, Brunzell JD, Goldberg AC et al. Evaluation and treatment of hypertriglyceridemia: an Endocrine Society clinical practice guideline. *J Clin Endocrinol Metab* 2012; **97**:2969–89.
- 2 Boullart AC, de Graaf J, Stalenhoef AF. Serum triglycerides and risk of cardiovascular disease. *Biochim Biophys Acta* 2012; **1821**:867–75.
- 3 Chapman MJ, Ginsberg HN, Amarenco P et al. Triglyceride-rich lipoproteins and high-density lipoprotein cholesterol in patients at high risk of cardiovascular disease: evidence and guidance for management. *Eur Heart J* 2011; **32**:1345–61.
- 4 Yuan G, Al-Shali KZ, Hegele RA. Hypertriglyceridemia: its etiology, effects and treatment. *CMAJ* 2007; **176**:1113–20.
- 5 Executive Summary of The Third Report of The National Cholesterol Education Program (NCEP) Expert Panel on Detection, Evaluation, And Treatment of High Blood Cholesterol In Adults (Adult Treatment Panel III). *JAMA* 2001; **285**:2486–97.
- 6 Catapano AL, Reiner Z, De Backer G et al. ESC/EAS Guidelines for the management of dyslipidaemias: the Task Force for the management of dyslipidaemias of the European Society of Cardiology (ESC) and the European Atherosclerosis Society (EAS). *Atherosclerosis* 2011; **217** Suppl 1:S1–44.
- 7 Reiner Z, Catapano AL, De Backer G et al. ESC/EAS Guidelines for the management of dyslipidaemias: the Task Force for the management of dyslipidaemias of the European Society of Cardiology (ESC) and the European Atherosclerosis Society (EAS). *Eur Heart J* 2011; **32**:1769–818.
- 8 Goussault Y, Turpin E, Neel D et al. 'Pseudohypertriglyceridemia' caused by hyperglycerolemia due to congenital enzyme deficiency. *Clin Chim Acta* 1982; **123**:269–74.
- 9 Langsted A, Nordestgaard BG. Nonfasting lipids, lipoproteins, and apolipoproteins in individuals with and without diabetes: 58 434 individuals from the Copenhagen General Population Study. *Clin Chem* 2011; **57**:482–9.
- 10 Guerin M, Egger P, Soudant C et al. Cholestryler ester flux from HDL to VLDL-1 is preferentially enhanced in type IIB hyperlipidemia in the postprandial state. *J Lipid Res* 2002; **43**:1652–60.
- 11 Nordestgaard BG, Benn M, Schnohr P, Tybjaerg-Hansen A. Nonfasting triglycerides and risk of myocardial infarction, ischemic heart disease, and death in men and women. *JAMA* 2007; **298**:299–308.
- 12 Freiberg JJ, Tybjaerg-Hansen A, Jensen JS, Nordestgaard BG. Nonfasting triglycerides and risk of ischemic stroke in the general population. *JAMA* 2008; **300**:2142–52.
- 13 Varbo A, Benn M, Tybjærg-Hansen A, Jørgensen AB, Frikke-Schmidt R, Nordestgaard BG. Remnant cholesterol as a causal risk factor for ischemic heart disease. *J Am Coll Cardiol* 2013; **61**:427–36.

- 14 Jørgensen AB, Frikke-Schmidt R, West AS, Grande P, Nordestgaard BG, Tybjærg-Hansen A. Genetically elevated nonfasting triglycerides and calculated remnant cholesterol as causal risk factors for myocardial infarction. *Eur Heart J* 2013; **34**:1826–33.
- 15 Zilversmit DB. Atherogenesis: a postprandial phenomenon. *Circulation* 1979; **60**:473–85.
- 16 Nordestgaard BG, Wootton R, Lewis B. Selective retention of VLDL, IDL, and LDL in the arterial intima of genetically hyperlipidemic rabbits in vivo. Molecular size as a determinant of fractional loss from the intima-inner media. *Arterioscler Thromb Vasc Biol* 1995; **15**:534–42.
- 17 Norata GD, Grigore L, Raselli S et al. Post-prandial endothelial dysfunction in hypertriglyceridemic subjects: molecular mechanisms and gene expression studies. *Atherosclerosis* 2007; **193**:321–7.
- 18 Malloy MJ, Kane JP. A risk factor for atherosclerosis: triglyceride-rich lipoproteins. *Adv Intern Med* 2001; **47**:111–36.
- 19 Mamo JC, Proctor SD, Smith D. Retention of chylomicron remnants by arterial tissue; importance of an efficient clearance mechanism from plasma. *Atherosclerosis* 1998; **141** Suppl 1:S63–9.
- 20 Di Angelantonio E, Sarwar N, Perry P et al. Major lipids, apolipoproteins, and risk of vascular disease. *JAMA* 2009; **302**:1993–2000.
- 21 Ng DS, Wong NC, Hegele RA. HDL-is it too big to fail? *Nat Rev Endocrinol* 2013; **9**:308–12.
- 22 Sarwar N, Sandhu MS, Ricketts SL et al. Triglyceride-mediated pathways and coronary disease: collaborative analysis of 101 studies. *Lancet* 2010; **375**:1634–9.
- 23 Jun M, Foote C, Lv J et al. Effects of fibrates on cardiovascular outcomes: a systematic review and meta-analysis. *Lancet* 2010; **375**:1875–84.
- 24 Sacks FM, Carey VJ, Fruchart JC. Combination lipid therapy in type 2 diabetes. *N Engl J Med* 2010; **363**:692–4.
- 25 AIM-HIGH Investigators; Boden WE, Probstfield JL, Anderson T et al. Niacin in patients with low HDL cholesterol levels receiving intensive statin therapy. *N Engl J Med* 2011; **365**:2255–67.
- 26 Rizos EC, Ntzani EE, Bika E, Kostapanos MS, Elisaf MS. Association between omega-3 fatty acid supplementation and risk of major cardiovascular disease events: a systematic review and meta-analysis. *JAMA* 2012; **308**:1024–33.
- 27 Hegele RA. Plasma lipoproteins: genetic influences and clinical implications. *Nat Rev Genet* 2009; **10**:109–21.
- 28 Soutar AK, Naoumova RP. Mechanisms of disease: genetic causes of familial hypercholesterolemia. *Nat Clin Pract Cardiovasc Med* 2007; **4**:214–25.
- 29 Hegele RA, Ban MR, Hsueh N et al. A polygenic basis for four classical Fredrickson hyperlipoproteinemia phenotypes that are characterized by hypertriglyceridemia. *Hum Mol Genet* 2009; **18**:4189–94.

- 30 Johansen CT, Wang J, Lanktree MB et al. An increased burden of common and rare lipid-associated risk alleles contributes to the phenotypic spectrum of hypertriglyceridemia. *Arterioscler Thromb Vasc Biol* 2011; **31**:1916–26.
- 31 Johansen CT, Kathiresan S, Hegele RA. Genetic determinants of plasma triglycerides. *J Lipid Res* 2011; **52**:189–206.
- 32 Johansen CT, Hegele RA. Genetic bases of hypertriglyceridemic phenotypes. *Curr Opin Lipidol* 2011; **22**:247–53.
- 33 Johansen CT, Hegele RA. Allelic and phenotypic spectrum of plasma triglycerides. *Biochim Biophys Acta* 2012; **1821**:833–42.
- 34 Talmud PJ, Shah S, Whittall R et al. Use of low-density lipoprotein cholesterol gene score to distinguish patients with polygenic and monogenic familial hypercholesterolaemia: a case-control study. *Lancet* 2013; **381**:1293–301.
- 35 Beil U, Grundy SM, Crouse JR, Zech L. Triglyceride and cholesterol metabolism in primary hypertriglyceridemia. *Arteriosclerosis* 1982; **2**:44–57.
- 36 Rahalkar AR, Giffen F, Har B et al. Novel LPL mutations associated with lipoprotein lipase deficiency: two case reports and a literature review. *Can J Physiol Pharmacol* 2009; **87**:151–60.
- 37 Babirak SP, Iverius PH, Fujimoto WY, Brunzell JD. Detection and characterization of the heterozygote state for lipoprotein lipase deficiency. *Arteriosclerosis* 1989; **9**:326–34.
- 38 Hegele RA, Breckenridge WC, Cox DW, Maguire GF, Little JA, Connelly PW. Interaction between variant apolipoproteins C-II and E that affects plasma lipoprotein concentrations. *Arterioscler Thromb* 1991; **11**:1303–9.
- 39 Nordestgaard BG, Abildgaard S, Wittrup HH, Steffensen R, Jensen G, Tybjaerg-Hansen A. Heterozygous lipoprotein lipase deficiency: frequency in the general population, effect on plasma lipid levels, and risk of ischemic heart disease. *Circulation* 1997; **96**:1737–44.
- 40 Priore Oliva C, Pisciotta L, Li Volti G et al. Inherited apolipoprotein A-V deficiency in severe hypertriglyceridemia. *Arterioscler Thromb Vasc Biol* 2005; **25**:411–7.
- 41 Johansen CT, Wang J, Lanktree MB et al. Excess of rare variants in genes identified by genome-wide association study of hypertriglyceridemia. *Nat Genet* 2010; **42**:684–7.
- 42 Johansen CT, Wang J, McIntyre AD et al. Excess of rare variants in non-genome-wide-association study candidate genes in patients with hypertriglyceridemia. *Circ Cardiovasc Genet* 2012; **5**:66–72.
- 43 Lee JH, Giannikopoulos P, Duncan SA et al. The transcription factor cyclic AMP-responsive element-binding protein H regulates triglyceride metabolism. *Nat Med* 2011; **17**:812–5.
- 44 Teslovich TM, Musunuru K, Smith AV et al. Biological, clinical and population relevance of 95 loci for blood lipids. *Nature* 2010; **466**:707–13.

- 45 Anderson TJ, Grégoire J, Hegele RA et al. 2012 update of the Canadian cardiovascular society guidelines for the diagnosis and treatment of dyslipidemia for the prevention of cardiovascular disease in the adult. *Can J Cardiol* 2013; **29**:151–67.
- 46 Sniderman AD, Williams K, Contois JH et al. A meta-analysis of low-density lipoprotein cholesterol, non-high-density lipoprotein cholesterol, and apolipoprotein B as markers of cardiovascular risk. *Circ Cardiovasc Qual Outcomes* 2011; **4**:337–345.
- 47 Baigent C, Keech A, Kearney PM et al; Cholesterol Treatment Trialists' (CTT) Collaborators. Efficacy and safety of cholesterol-lowering treatment: prospective meta-analysis of data from 90,056 participants in 14 randomised trials of statins. *Lancet* 2005; **366**:1267–78.
- 48 Boekholdt SM, Arsenault BJ, Mora S et al. Association of LDL cholesterol, non-HDL cholesterol, and apolipoprotein B levels with risk of cardiovascular events among patients treated with statins: a meta-analysis. *JAMA* 2012; **307**:1302–9.
- 49 Huijgen R, Abbink EJ, Bruckert E et al; Triple Study Group. Colesevelam added to combination therapy with a statin and ezetimibe in patients with familial hypercholesterolemia: a 12-week, multicenter, randomized, double-blind, controlled trial. *Clin Ther* 2010; **32**:615–25.
- 50 Asselbergs FW, Guo Y, van Iperen EP et al. Large-scale gene-centric meta-analysis across 32 studies identifies multiple lipid loci. *Am J Hum Genet* 2012; **91**:823–38.
- 51 Chen R, Mias GI, Li-Pook-Than J et al. Personal omics profiling reveals dynamic molecular and medical phenotypes. *Cell* 2012; **148**:1293–307.
- 52 Hawrylycz MJ, Lein ES, GUILLOZET-BONGAARTS AL et al. An anatomically comprehensive atlas of the adult human brain transcriptome. *Nature* 2012; **489**:391–9.
- 53 Gaudet D, de Wal J, Tremblay K et al. Review of the clinical development of alipogene tiparvovec gene therapy for lipoprotein lipase deficiency. *Atheroscler Suppl* 2010; **11**:55–60.
- 54 Stroes ES, Nierman MC, Meulenbergh JJ et al. Intramuscular administration of AAV1-lipoprotein lipase S447X lowers triglycerides in lipoprotein lipase-deficient patients. *Arterioscler Thromb Vasc Biol* 2008; **28**:2303–4.
- 55 Cuchel M, Meagher EA, du Toit Theron H et al; Phase 3 HoFH Lomitapide Study investigators. Efficacy and safety of a microsomal triglyceride transfer protein inhibitor in patients with homozygous familial hypercholesterolaemia: a single-arm, open-label, phase 3 study. *Lancet* 2013; **381**:40–6.
- 56 Raal FJ, Santos RD, Blom DJ et al. Mipomersen, an apolipoprotein B synthesis inhibitor, for lowering of LDL cholesterol concentrations in patients with homozygous familial hypercholesterolaemia: a randomised, double-blind, placebo-controlled trial. *Lancet* 2010; **375**:998–1006.
- 57 Pollin TI, Damcott CM, Shen H et al. A null mutation in human APOC3 confers a favorable plasma lipid profile and apparent cardioprotection. *Science* 2008; **322**:1702–5.

58 Musunuru K, Pirruccello JP, Do R et al. Exome sequencing, ANGPTL3 mutations, and familial combined hypolipidemia. *N Engl J Med* 2010; **363**:2220–7.