

**Appendix Table 6. Studies Excluded from Key Question 3**

Study Citation	Reason for Exclusion
Adams PC, Agnew S. Alcoholism in hereditary hemochromatosis revisited: prevalence and clinical consequences among homozygous siblings. <i>Hepatology</i> . 1996;23:724-7. [PMID: 8666324]	Case series
Adams PC, Kertesz AE, Valberg LS. Clinical presentation of hemochromatosis: a changing scene. <i>Am J Med</i> . 1991;90:445-9. [PMID: 2012084]	Case series
Adams PC, Kertesz AE, Valberg LS. Screening for hemochromatosis in children of homozygotes: prevalence and cost-effectiveness. <i>Hepatology</i> . 1995;22:1720-7. [PMID: 7489980]	<18 y included
Adams PC. Haemochromatosis: find them or forget about them? <i>Eur J Gastroenterol Hepatol</i> . 2004;16:857-8. [PMID: 15316408]	Editorial
Assy N, Adams PC. Predictive value of family history in diagnosis of hereditary hemochromatosis. <i>Dig Dis Sci</i> . 1997;42:1312-5. [PMID: 9201100]	No HFE testing
Bacon BR, Olynyk JK, Brunt EM, Britton RS, Wolff RK. HFE genotype in patients with hemochromatosis and other liver diseases. <i>Ann Intern Med</i> . 1999;130:953-62. [PMID: 10383365]	Does not meet our definition of clinical hemochromatosis
Bassett ML, Halliday JW, Ferris RA, Powell LW. Diagnosis of hemochromatosis in young subjects: predictive accuracy of biochemical screening tests. <i>Gastroenterology</i> . 1984;87:628-33. [PMID: 6745616]	Does not include primary results
Bassett ML, Halliday JW, Powell LW. Value of hepatic iron measurements in early hemochromatosis and determination of the critical iron level associated with fibrosis. <i>Hepatology</i> . 1986;6:24-9. [PMID: 3943787]	Case series
Bhavnani M, Lloyd D, Bhattacharyya A, Marples J, Elton P, Worwood M. Screening for genetic haemochromatosis in blood samples with raised alanine aminotransferase. <i>Gut</i> . 2000;46:707-10. [PMID: 10764716]	Quality
Bonkovsky HL, Jawaid Q, Tortorelli K, LeClair P, Cobb J, Lambrecht RW, et al. Non-alcoholic steatohepatitis and iron: increased prevalence of mutations of the HFE gene in non-alcoholic steatohepatitis. <i>J Hepatol</i> . 1999;31:421-9. [PMID: 10488699]	Does not meet our definition of clinical hemochromatosis
Bregman H, Gelfand MC, Winchester JF, Manz HJ, Kneppshield JH, Schreiner GE. iron-overload-associated myopathy in patients on maintenance haemodialysis: a histocompatibility-linked disorder. <i>Lancet</i> . 1980;2:882-5. [PMID: 6107546]	Not the correct population
Brissot P, Moirand R, Jouanolle AM, Guyader D, Le Gall JY, Deugnier Y, et al. A genotypic study of 217 unrelated probands diagnosed as "genetic hemochromatosis" on "classical" phenotypic criteria. <i>J Hepatol</i> . 1999;30:588-93. [PMID: 10207799]	Does not report relevant prevalence or risk measures
Campo S, Restuccia T, Villari D, Raffa G, Cucinotta D, Squadrito G, et al. Analysis of haemochromatosis gene mutations in a population from the Mediterranean Basin. <i>Liver</i> . 2001;21:233-6. [PMID: 11454185]	Not the correct population
Cavanaugh JA, Wilson SR, Bassett ML. Genetic testing for HFE hemochromatosis in Australia: the value of testing relatives of simple heterozygotes. <i>J Gastroenterol Hepatol</i> . 2002;17:800-3. [PMID: 12121511]	Does not include primary results
Conte D, Manachino D, Colli A, Guala A, Aimo G, Andreoletti M, et al. Prevalence of genetic hemochromatosis in a cohort of Italian patients with diabetes mellitus. <i>Ann Intern Med</i> . 1998;128:370-3. [PMID: 9490597]	Not the correct population
Dalury DF, Ewald FC, Christie MJ, Scott RD. Total knee arthroplasty in a group of patients less than 45 years of age. <i>J Arthroplasty</i> . 1995;10:598-602. [PMID: 9273369]	Does not report relevant prevalence or risk measures
Ellervik C, Mandrup-Poulsen T, Nordestgaard BG, Larsen LE, Appleyard M, Frandsen M, et al. Prevalence of hereditary haemochromatosis in late-onset type 1 diabetes mellitus: a retrospective study. <i>Lancet</i> . 2001;358:1405-9. [PMID: 11705485]	Not the correct population
Feller ER, Pont A, Wands JR, Carter EA, Foster G, Kourides IA, et al. Familial hemochromatosis. Physiologic studies in the precirrhotic stage of the disease. <i>N Engl J Med</i> . 1977;296:1422-6. [PMID: 194151]	Case series
Fiel MI, Schiano TD, Bodenheimer HC, Thung SN, King TW, Varma CR, et al. Hereditary hemochromatosis in liver transplantation. <i>Liver Transpl Surg</i> . 1999;5:50-6. [PMID: 9873093]	Does not report relevant prevalence or risk measures
Gleeson F, Ryan E, Barrett S, Crowe J. Clinical expression of haemochromatosis in Irish C282Y homozygotes identified through family screening. <i>Eur J Gastroenterol Hepatol</i> . 2004;16:859-63. [PMID: 15316409]	Does not report relevant prevalence or risk measures
Guyader D, Jacquelinet C, Moirand R, Turlin B, Mendler MH, Chaperon J, et al. Noninvasive prediction of fibrosis in C282Y homozygous hemochromatosis. <i>Gastroenterology</i> . 1998;115:929-36. [PMID: 9753496]	Not the correct population
Hultcrantz R, Gabrielsson N. Patients with persistent elevation of aminotransferases: investigation with ultrasonography, radionuclide imaging and liver biopsy. <i>J Intern Med</i> . 1993;233:7-12. [PMID: 8429291]	Not relevant outcomes
Jeffrey GP, Adams PC. Pitfalls in the genetic diagnosis of hereditary hemochromatosis. <i>Genet Test</i> . 2000;4:143-6. [PMID: 10953953]	Editorial
Jordan JM. Arthritis in hemochromatosis or iron storage disease. <i>Curr Opin Rheumatol</i> . 2004;16:62-6. [PMID: 14673391]	Review article
Jorquera F, Dominguez A, Diaz-Golpe V, Espinel J, Munoz F, Herrera A, et al. C282Y and H63D mutations of the haemochromatosis gene in patients with iron overload. <i>Rev Esp Enferm Dig</i> . 2001;93:293-302. [PMID: 11488107]	Does not report relevant prevalence or risk measures
Koefoed P, Dalhoff K, Dissing J, Kramer I, Milman N, Pedersen P, et al. HFE mutations and hemochromatosis in Danish patients admitted for HFE genotyping. <i>Scand J Clin Lab Invest</i> . 2002;62:527-35. [PMID: 12512743]	Quality
Krawczak M, Cooper DN, Schmidtke J. Estimating the efficacy and efficiency of cascade genetic screening. <i>Am J Hum Genet</i> . 2001;69:361-70. [PMID: 11431707]	Does not include primary results
Li J, Zhu Y, Singal DP. HFE gene mutations in patients with rheumatoid arthritis. <i>J Rheumatol</i> . 2000;27:2074-7. [PMID: 10990216]	Quality
Mathews JL, Williams HJ. Arthritis in hereditary hemochromatosis. <i>Arthritis Rheum</i> . 1987;30:1137-41. [PMID: 3675659]	Not HFE
McCune CA, Ravine D, Worwood M, Jackson HA, Evans HM, Hutton D. Screening for hereditary haemochromatosis within families and beyond. <i>Lancet</i> . 2003;362:1897-8. [PMID: 14667749]	Does not report relevant prevalence or risk measures
Nassar BA, Zayed EM, Title LM, O'Neill BJ, Bata IR, Kirkland SA, et al. Relation of HFE gene mutations, high iron stores and early onset coronary artery disease. <i>Can J Cardiol</i> . 1998;14:215-20. [PMID: 9520858]	Quality
Nelson RL, Persky V, Davis F, Becker E. Risk of disease in siblings of patients with hereditary hemochromatosis. <i>Digestion</i> . 2001;64:120-4. [PMID: 11684826]	Quality
Olynyk J, Hall P, Ahern M, Kwiatek R, Mackinnon M. Screening for genetic haemochromatosis in a rheumatology clinic. <i>Aust N Z J Med</i> . 1994;24:22-5. [PMID: 8002853]	Quality

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**Appendix Table 6—Continued**

Study Citation	Reason for Exclusion
Panajotopoulos N, Piperno A, Conte D, Mandelli C, Cesana M, Mercuriali F, et al. HLA typing in 67 Italian patients with idiopathic hemochromatosis and their relatives. <i>Tissue Antigens</i> . 1989;33:431-6. [PMID: 2734773]	Not the correct population
Peterlin B, Globocnik Petrovic M, Makuc J, Hawlina M, Petrovic D. A hemochromatosis-causing mutation C282Y is a risk factor for proliferative diabetic retinopathy in Caucasians with type 2 diabetes. <i>J Hum Genet</i> . 2003;48:646-9. [PMID: 14618419]	Not the correct population
Piperno A, D'Alba R, Fargion S, Roffi L, Sampietro M, Parma S, et al. Liver iron concentration in chronic viral hepatitis: a study of 98 patients. <i>Eur J Gastroenterol Hepatol</i> . 1995;7:1203-8. [PMID: 8789313]	Not the correct population
Rasmussen ML, Folsom AR, Catellier DJ, Tsai MY, Garg U, Eckfeldt JH. A prospective study of coronary heart disease and the hemochromatosis gene (HFE) C282Y mutation: the Atherosclerosis Risk in Communities (ARIC) study. <i>Atherosclerosis</i> . 2001;154:739-46. [PMID: 11257277]	Does not report relevant prevalence or risk measures
Roberts AG, Whatley SD, Morgan RR, Worwood M, Elder GH. Increased frequency of the haemochromatosis Cys282Tyr mutation in sporadic porphyria cutanea tarda. <i>Lancet</i> . 1997;349:321-3. [PMID: 9024376]	Does not meet our definition of clinical hemochromatosis
Rosenqvist M, Hultcrantz R. Prevalence of a haemochromatosis among men with clinically significant bradyarrhythmias. <i>Eur Heart J</i> . 1989;10:473-8. [PMID: 2788086]	No HFE testing
Sampietro M, Piperno A, Lupica L, Arosio C, Vergani A, Corbetta N, et al. High prevalence of the His63Asp HFE mutation in Italian patients with porphyria cutanea tarda. <i>Hepatology</i> . 1998;27:181-4. [PMID: 9425935]	Does not meet our definition of clinical hemochromatosis
Schmid H, Struppeler C, Braun GS, Kellner W, Kellner H. Ankle and hindfoot arthropathy in hereditary hemochromatosis. <i>J Rheumatol</i> . 2003;30:196-9. [PMID: 12508413]	Not the correct population
Sham RL, Raubertas RF, Braggins C, Cappuccio J, Gallagher M, Phatak PD. Asymptomatic hemochromatosis subjects: genotypic and phenotypic profiles. <i>Blood</i> . 2000;96:3707-11. [PMID: 11090050]	Not the correct population
Shoaf EH Jr. Hemochromatosis discovered through blood donor screening for alanine aminotransferase. <i>N C Med J</i> . 1990;51:443-5. [PMID: 2234109]	Case report
Siezenga MA, Rasp E, Wijermans PW. Testing families with HFE-related hereditary haemochromatosis. <i>Neth J Med</i> . 2004;62:156-9. [PMID: 15366698]	Case report
Simon M, Alexandre JL, Bourel M, Le Marec B, Scordia C. Heredity of idiopathic hemochromatosis: a study of 106 families. <i>Clin Genet</i> . 1977;11:327-41. [PMID: 862210]	Quality
Tannapfel A, Stolzel U, Kostler E, Melz S, Richter M, Keim V, et al. C282Y and H63D mutation of the hemochromatosis gene in German porphyria cutanea tarda patients. <i>Virchows Arch</i> . 2001;439:1-5. [PMID: 11499833]	Does not meet our definition of clinical hemochromatosis
Timms AE, Sathananthan R, Bradbury L, Athanasou NA, Wordsworth BP, Brown MA. Genetic testing for hemochromatosis in patients with chondrocalcinosis. <i>Ann Rheum Dis</i> . 2002;61:745-7. [PMID: 12117686]	Quality