

Appendix Table 4. Studies Excluded from Key Question 1

Study Citation	Reason for Exclusion
Iron overload disorders among Hispanics—San Diego, California, 1995. <i>MMWR Morb Mortal Wkly Rep.</i> 1996;45:991-3. [PMID: 9005307]	Study disease definition does not meet our definition of asymptomatic primary iron overload or clinical disease
A simple genetic test identifies 90% of UK patients with haemochromatosis. The UK Haemochromatosis Consortium. <i>Gut.</i> 1997;41:841-4. [PMID: 9462220]	Not a screening population
Adams PC, Reboussin DM, Barton JC, McLaren CE, Eckfeldt JH, McLaren GD, et al. Hemochromatosis and iron-overload screening in a racially diverse population. <i>N Engl J Med.</i> 2005;352:1769-78. [PMID: 15858186]	Does not report relevant outcomes
Adams PC. Is there a threshold of hepatic iron concentration that leads to cirrhosis in C282Y hemochromatosis? <i>Am J Gastroenterol.</i> 2001;96:567-9. [PMID: 11232708]	Not a screening population
Adams PC, Deugnier Y, Moirand R, Brissot P. The relationship between iron overload, clinical symptoms, and age in 410 patients with genetic hemochromatosis. <i>Hepatology.</i> 1997;25:162-6. [PMID: 8985284]	Not a screening population
Adams PC, Gregor JC, Kertesz AE, Valberg LS. Screening blood donors for hereditary hemochromatosis: decision analysis model based on a 30-year database. <i>Gastroenterology.</i> 1995;109:177-88. [PMID: 7797016]	Does not contain primary data
Adams PC, Kertesz AE, Valberg LS. Clinical presentation of hemochromatosis: a changing scene. <i>Am J Med.</i> 1991;90:445-9. [PMID: 2012084]	Not a screening population
Adams PC, Speechley M, Kertesz AE. Long-term survival analysis in hereditary hemochromatosis. <i>Gastroenterology.</i> 1991;101:368-72. [PMID: 2065912]	Not a screening population
Adams PC. Hepatic iron in hemochromatosis. <i>Dig Dis Sci.</i> 1990;35:690-2. [PMID: 2344801]	Includes data from patients < 18 y
Ammann RW, Muller E, Bansky J, Schuler G, Hacki WH. High incidence of extrahepatic carcinomas in idiopathic hemochromatosis. <i>Scand J Gastroenterol.</i> 1980;15:733-6. [PMID: 6259710]	Not a screening population
Asberg A, Hveem K, Kruger O, Bjerve KS. Persons with screening-detected haemochromatosis: as healthy as the general population? <i>Scand J Gastroenterol.</i> 2002;37:719-24. [PMID: 12126253]	Study disease definition does not meet our definition of asymptomatic primary iron overload or clinical disease
Asberg A, Hveem K, Thorstensen K, Ellekjer E, Kannelonning K, Fjosne U, et al. Screening for hemochromatosis: high prevalence and low morbidity in an unselected population of 65,238 persons. <i>Scand J Gastroenterol.</i> 2001;36:1108-15. [PMID: 11589387]	Does not include C282Y genotyping in screening sequence
Askari AD, Muir WA, Rosner IA, Moskowitz RW, McLaren GD, Braun WE. Arthritis of hemochromatosis. Clinical spectrum, relation to histocompatibility antigens, and effectiveness of early phlebotomy. <i>Am J Med.</i> 1983;75:957-65. [PMID: 6650551]	Not a screening population
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]	Study design
Bacon BR, Sadiq SA. Hereditary hemochromatosis: presentation and diagnosis in the 1990s. <i>Am J Gastroenterol.</i> 1997;92:784-9. [PMID: 9149185]	Not a screening population
Baer DM, Simons JL, Staples RL, Rumore GJ, Morton CJ. Hemochromatosis screening in asymptomatic ambulatory men 30 years of age and older. <i>Am J Med.</i> 1995;98:464-8. [PMID: 7733125]	Does not include C282Y genotyping in screening sequence
Balan V, Baldus W, Fairbanks V, Michels V, Burritt M, Klee G. Screening for hemochromatosis: a cost-effectiveness study based on 12,258 patients. <i>Gastroenterology.</i> 1994;107:453-9. [PMID: 8039622]	Does not include C282Y genotyping in screening sequence
Barosi G, Salvaneschi L, Grasso M, Martinetti M, Marchetti M, Bodini U, et al. High prevalence of a screening-detected, HFE-unrelated, mild idiopathic iron overload in Northern Italy. <i>Haematologica.</i> 2002;87:472-8. [PMID: 12010659]	Does not report relevant outcomes
Barton JC, Cheatwood SM, Key TJ, Acton RT. Hemochromatosis detection in a health screening program at an Alabama forest products mill. <i>J Occup Environ Med.</i> 2002;44:745-51. [PMID: 12185795]	Does not report relevant outcomes
Barton JC, Barton NH, Alford TJ. Diagnosis of hemochromatosis probands in a community hospital. <i>Am J Med.</i> 1997;103:498-503. [PMID: 9428833]	Not a screening population
Barton JC, Shih WW, Sawada-Hirai R, Acton RT, Harmon L, Rivers C, et al. Genetic and clinical description of hemochromatosis probands and heterozygotes: evidence that multiple genes linked to the major histocompatibility complex are responsible for hemochromatosis. <i>Blood Cells Mol Dis.</i> 1997;23:135-45; discussion 145a-b. [PMID: 9215758]	Not a screening population
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]	Participants < 18 y included
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]	Does not report relevant outcomes
Bell H, Thordal C, Raknerud N, Hansen T, Bosnes V, Halvorsen R, et al. Prevalence of hemochromatosis among first-time and repeat blood donors in Norway. <i>J Hepatol.</i> 1997;26:272-9. [PMID: 9059946]	Does not include C282Y genotyping in screening sequence
Bell H, Berg JP, Undlien DE, Distant S, Raknerud N, Heier HE, et al. The clinical expression of hemochromatosis in Oslo, Norway. Excessive oral iron intake may lead to secondary hemochromatosis even in HFE C282Y mutation negative subjects. <i>Scand J Gastroenterol.</i> 2000;35:1301-7. [PMID: 11199371]	Not a screening population
Borwein ST, Ghent CN, Flanagan PR, Chamberlain MJ, Valberg LS. Genetic and phenotypic expression of hemochromatosis in Canadians. <i>Clin Invest Med.</i> 1983;6:171-9. [PMID: 6652983]	Does not report relevant outcomes
Bradbear RA, Bain C, Siskind V, Schofield FD, Webb S, Axelsen EM, et al. Cohort study of internal malignancy in genetic hemochromatosis and other chronic nonalcoholic liver diseases. <i>J Natl Cancer Inst.</i> 1985;75:81-4. [PMID: 2989605]	Not a screening population
Bradley LA, Haddow JE, Palomaki GE. Population screening for haemochromatosis: a unifying analysis of published intervention trials. <i>J Med Screen.</i> 1996;3:178-84. [PMID: 9041481]	Review article
Bulaj ZJ, Ajioka RS, Phillips JD, LaSalle BA, Jorde LB, Griffen LM, et al. Disease-related conditions in relatives of patients with hemochromatosis. <i>N Engl J Med.</i> 2000;343:1529-35. [PMID: 11087882]	Quality
Buysschaert M, Paris I, Selvais P, Hermans MP. Clinical aspects of diabetes secondary to idiopathic haemochromatosis in French-speaking Belgium. <i>Diabetes Metab.</i> 1997;23:308-13. [PMID: 9342544]	Case series

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Appendix Table 4—Continued

Study Citation	Reason for Exclusion
Cadet E, Capron D, Gallet M, Omanga-Leke ML, Boutignon H, Julier C, et al. Reverse cascade screening of newborns for hereditary haemochromatosis: a model for other late onset diseases? <i>J Med Genet.</i> 2005;42:390-5. [PMID: 15863667]	Includes data from patients < 18 y Cannot separate C282Y homozygotes from C282Y heterozygotes
Cartwright GE, Edwards CQ, Kravitz K, Skolnick M, Amos DB, Johnson A, et al. Hereditary hemochromatosis. Phenotypic expression of the disease. <i>N Engl J Med.</i> 1979;301:175-9. [PMID: 449974]	Does not report relevant outcomes
Cecchetti G, Binda A, Piperno A, Nador F, Fargion S, Fiorelli G. Cardiac alterations in 36 consecutive patients with idiopathic haemochromatosis: polygraphic and echocardiographic evaluation. <i>Eur Heart J.</i> 1991;12:224-30. [PMID: 2044557]	Not a screening population
Cogswell ME, Gallagher ML, Steinberg KK, Caudill PhD SP, Looker AC, Bowman BA, et al. HFE genotype and transferrin saturation in the United States. <i>Genet Med.</i> 2003;5:304-10. [PMID: 12865759]	Study disease definition does not meet our definition of asymptomatic primary iron overload or clinical disease
Crawford DH, Jazwinska EC, Cullen LM, Powell LW. Expression of HLA-linked hemochromatosis in subjects homozygous or heterozygous for the C282Y mutation. <i>Gastroenterology.</i> 1998;114:1003-8. [PMID: 9558290]	Not a screening population
Cundy T, Bomford A, Butler J, Wheeler M, Williams R. Hypogonadism and sexual dysfunction in hemochromatosis: the effects of cirrhosis and diabetes. <i>J Clin Endocrinol Metab.</i> 1989;69:110-6. [PMID: 2732293]	Not a screening population
Deugnier YM, Charalambous P, Le Quilleuc D, Turlin B, Searle J, Brissot P, et al. Preneoplastic significance of hepatic iron-free foci in genetic hemochromatosis: a study of 185 patients. <i>Hepatology.</i> 1993;18:1363-9. [PMID: 7902316]	Not a screening population
Distante S, Berg JP, Lande K, Haug E, Bell H. HFE gene mutation (C282Y) and phenotypic expression among a hospitalised population in a high prevalence area of haemochromatosis. <i>Gut.</i> 2000;47:575-9. [PMID: 10986220]	Inconsistent application of exclusion criteria
Edwards CQ, Griffen LM, Kushner JP. The morbidity of hemochromatosis among clinically unselected homozygotes: preliminary report. <i>Adv Exp Med Biol.</i> 1994;356:303-8. [PMID: 7887235]	Does not report relevant outcomes
Edwards CQ, Griffen LM, Kushner JP. Comparison of stainable liver iron between symptomatic and asymptomatic hemochromatosis homozygotes and their homozygous relatives. <i>Am J Med Sci.</i> 1991;301:44-6. [PMID: 1994729]	Not a screening population
Edwards CQ, Griffen LM, Goldgar D, Drummond C, Skolnick MH, Kushner JP. Prevalence of hemochromatosis among 11,065 presumably healthy blood donors. <i>N Engl J Med.</i> 1988;318:1355-62. [PMID: 3367936]	Does not include C282Y genotyping in screening sequence
Edwards CQ, Cartwright GE, Skolnick MH, Amos DB. Homozygosity for hemochromatosis: clinical manifestations. <i>Ann Intern Med.</i> 1980;93:519-25. [PMID: 7436183]	Does not report relevant outcomes
Elliott R, Lin BP, Dent OF, Tait A, Smith CI. Prevalence of hemochromatosis in a random sample of asymptomatic men. <i>Aust N Z J Med.</i> 1986;16:491-5. [PMID: 3467692]	Does not include C282Y genotyping in screening sequence
Elmberg M, Hultcrantz R, Ekblom A, Brandt L, Olsson S, Olsson R, et al. Cancer risk in patients with hereditary hemochromatosis and in their first-degree relatives. <i>Gastroenterology.</i> 2003;125:1733-41. [PMID: 14724826]	Not a screened population
Fargion S, Fracanzani AL, Piperno A, Braga M, D'Alba R, Ronchi G, et al. Prognostic factors for hepatocellular carcinoma in genetic hemochromatosis. <i>Hepatology.</i> 1994;20:1426-31. [PMID: 7982640]	Not a screening population
Fargion S, Mandelli C, Piperno A, Cesana B, Fracanzani AL, Fraquelli M, et al. Survival and prognostic factors in 212 Italian patients with genetic hemochromatosis. <i>Hepatology.</i> 1992;15:655-9. [PMID: 1312985]	Not a screening population
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]	Not a screening population
Fleming DJ, Jacques PF, Tucker KL, Massaro JM, D'Agostino RB Sr, Wilson PW, et al. Iron status of the free-living, elderly Framingham Heart Study cohort: an iron-replete population with a high prevalence of elevated iron stores. <i>Am J Clin Nutr.</i> 2001;73:638-46. [PMID: 11237943]	Does not report relevant outcomes
Fletcher LM, Dixon JL, Purdie DM, Powell LW, Crawford DH. Excess alcohol greatly increases the prevalence of cirrhosis in hereditary hemochromatosis. <i>Gastroenterology.</i> 2002;122:281-9. [PMID: 11832443]	Not a screening population
Fox CJ, Cullen DJ, Knuiaman MW, Cumpston GN, Divitini ML, Rossi E, et al. Effects of body iron stores and haemochromatosis genotypes on coronary heart disease outcomes in the Busseton health study. <i>J Cardiovasc Risk.</i> 2002;9:287-93. [PMID: 12394323]	Study disease definition does not meet our definition of asymptomatic primary iron overload or clinical disease
Fracanzani AL, Conte D, Fraquelli M, Taioli E, Mattioli M, Losco A, et al. Increased cancer risk in a cohort of 230 patients with hereditary hemochromatosis in comparison to matched control patients with non-iron-related chronic liver disease. <i>Hepatology.</i> 2001;33:647-51. [PMID: 11230745]	Not a screening population
Fracanzani AL, Fargion S, Romano R, Conte D, Piperno A, D'Alba R, et al. Portal hypertension and iron depletion in patients with genetic hemochromatosis. <i>Hepatology.</i> 1995;22:1127-31. [PMID: 7557861]	Not a screening population
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]	Includes data from patients < 18 y
Hallberg L, Bjorn-Rasmussen E, Jungner I. Prevalence of hereditary haemochromatosis in two Swedish urban areas. <i>J Intern Med.</i> 1989;225:249-55. [PMID: 2723582].	Does not include C282Y genotyping in screening sequence
Halliday JW, Russo AM, Cowlishaw JL, Powell LW. Serum-ferritin in diagnosis of haemochromatosis. A study of 43 families. <i>Lancet.</i> 1977;2:621-4. [PMID: 71445]	Does not report relevant outcomes
Hamilton EB, Bomford AB, Laws JW, Williams R. The natural history of arthritis in idiopathic haemochromatosis: progression of the clinical and radiological features over ten years. <i>Q J Med.</i> 1981;50:321-9. [PMID: 7330169]	Not a screening population
Jackson HA, Carter K, Darke C, Guttridge MG, Ravine D, Hutton RD, et al. HFE mutations, iron deficiency and overload in 10,500 blood donors. <i>Br J Haematol.</i> 2001;114:474-84. [PMID: 11529872]	Study disease definition does not meet our definition of asymptomatic primary iron overload or clinical disease
Jiang R, Manson JE, Meigs JB, Ma J, Rifai N, Hu FB. Body iron stores in relation to risk of type 2 diabetes in apparently healthy women. <i>JAMA.</i> 2004;291:711-7. [PMID: 14871914]	Study design

Appendix Table 4—Continued

Study Citation	Reason for Exclusion
Jonsson JJ, Johannesson GM, Sigfusson N, Magnusson B, Thjodleifsson B, Magnusson S. Prevalence of iron deficiency and iron overload in the adult Icelandic population. <i>J Clin Epidemiol.</i> 1991;44:1289-97. [PMID: 1753260]	Does not include C282Y genotyping in screening sequence
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]	Not a screening population
Karlsson M, Ikkala E, Reunanen A, Takkunen H, Vuori E, Makinen J. Prevalence of hemochromatosis in Finland. <i>Acta Med Scand.</i> 1988;224:385-90. [PMID: 3188989]	Does not include C282Y genotyping in screening sequence
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]	Not a screening population
Lalouel JM, Le Mignon L, Simon M, Fauchet R, Bourel M, Rao DC, et al. Genetic analysis of idiopathic hemochromatosis using both qualitative (disease status) and quantitative (serum iron) information. <i>Am J Hum Genet.</i> 1985;37:700-18. [PMID: 9556659]	Does not report relevant outcomes
Leggett BA, Halliday JW, Brown NN, Bryant S, Powell LW. Prevalence of haemochromatosis amongst asymptomatic Australians. <i>Br J Haematol.</i> 1990;74:525-30. [PMID: 2346731]	Does not include C282Y genotyping in screening sequence
Lin E, Adams PC. Biochemical liver profile in hemochromatosis. A survey of 100 patients. <i>J Clin Gastroenterol.</i> 1991;13:316-20. [PMID: 2066547]	Not a screening population
Lindmark B, Eriksson S. Regional differences in the idiopathic hemochromatosis gene frequency in Sweden. <i>Acta Med Scand.</i> 1985;218:299-304. [PMID: 4072776]	Does not include C282Y genotyping in screening sequence
Livesey KJ, Wilmhurst VL, Carter K, Worwood M, Cadet E, Rochette J, et al. The 16189 variant of mitochondrial DNA occurs more frequently in C282Y homozygotes with haemochromatosis than those without iron loading. <i>J Med Genet.</i> 2004;41:6-10. [PMID: 14729817]	Not a screening population
Mainous AG 3rd, Gill JM, Pearson WS. Should we screen for hemochromatosis? An examination of evidence of downstream effects on morbidity and mortality. <i>Arch Intern Med.</i> 2002;162:1769-74. [PMID: 12153381]	Does not report relevant outcomes
Mainous AG 3rd, King DE, Pearson WS, Garr DR. Is an elevated serum transferrin saturation associated with the development of diabetes? <i>J Fam Pract.</i> 2002;51:933-6. [PMID: 12485546]	Does not include C282Y genotyping in screening sequence
Mainous AG 3rd, Wells B, Carek PJ, Gill JM, Geesey ME. The mortality risk of elevated serum transferrin saturation and consumption of dietary iron. <i>Ann Fam Med.</i> 2004;2:139-44. [PMID: 15083854]	Does not include C282Y genotyping in screening sequence
Mainous AG 3rd, Gill JM, Carek PJ. Elevated serum transferrin saturation and mortality. <i>Ann Fam Med.</i> 2004;2:133-8. [PMID: 15083853]	Does not include C282Y genotyping in screening sequence
Mainous AG 3rd, Gill JM, Everett CJ. Transferrin saturation, dietary iron intake, and risk of cancer. <i>Ann Fam Med.</i> 2005;3:131-7. [PMID: 15798039]	Does not report relevant outcomes
Mathews JL, Williams HJ. Arthritis in hereditary hemochromatosis. <i>Arthritis Rheum.</i> 1987;30:1137-41. [PMID: 3675659]	Ineligible study design
McCune CA, Al-Jader LN, May A, Hayes SL, Jackson HA, Worwood M. Hereditary haemochromatosis: only 1% of adult HFE C282Y homozygotes in South Wales have a clinical diagnosis of iron overload. <i>Hum Genet.</i> 2002;111:538-43. [PMID: 12436244]	Not a screening population
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]	Not a screening population Quality
Merryweather-Clarke AT, Worwood M, Parkinson L, Mattock C, Pointon JJ, Shearman JD, et al. The effect of HFE mutations on serum ferritin and transferrin saturation in the Jersey population. <i>Br J Haematol.</i> 1998;101:369-73. [PMID: 9609537]	Does not report relevant outcomes
Milman N, Pedersen P, Steig T, Byg KE, Graudal N, Fenger K. Clinically overt hereditary hemochromatosis in Denmark 1948-1985: epidemiology, factors of significance for long-term survival, and causes of death in 179 patients. <i>Ann Hematol.</i> 2001;80:737-44. [PMID: 11797115]	Quality
Milman N. Iron status markers in hereditary haemochromatosis: distinction between individuals being homozygous and heterozygous for the haemochromatosis allele. <i>Eur J Haematol.</i> 1991;47:292-8. [PMID: 1954989]	Does not report relevant outcomes
Moirand R, Jouanolle AM, Brissot P, Le Gall JY, David V, Deugnier Y. Phenotypic expression of HFE mutations: a French study of 1110 unrelated iron-overloaded patients and relatives. <i>Gastroenterology.</i> 1999;116:372-7. [PMID: 9922318]	Does not report relevant outcomes
Moodie SJ, Ang L, Stenner JM, Finlayson C, Khotari A, Levin GE, et al. Testing for haemochromatosis in a liver clinic population: relationship between ethnic origin, HFE gene mutations, liver histology and serum iron markers. <i>Eur J Gastroenterol Hepatol.</i> 2002;14:223-9. [PMID: 11953685]	Not a screening population
Morrison ED, Brandhagen DJ, Phatak PD, Barton JC, Krawitt EL, El-Serag HB, et al. Serum ferritin level predicts advanced hepatic fibrosis among U.S. patients with phenotypic hemochromatosis. <i>Ann Intern Med.</i> 2003;138:627-33. [PMID: 12693884]	Not a screening population
Mura C, Noubbaum JB, Verger P, Moalic MT, Ragueneas O, Mercier AY, et al. Phenotype-genotype correlation in haemochromatosis subjects. <i>Hum Genet.</i> 1997;101:271-6. [PMID: 9439654]	Not a screening population
Nash S, Marconi S, Sikorska K, Naeem R, Nash G. Role of liver biopsy in the diagnosis of hepatic iron overload in the era of genetic testing. <i>Am J Clin Pathol.</i> 2002;118:73-81. [PMID: 12109859]	Not a screening population
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
Niederer C, Niederer CM, Lange S, Littauer A, Abdel-Jalil N, Maurer M, et al. Screening for hemochromatosis and iron deficiency in employees and primary care patients in Western Germany. <i>Ann Intern Med.</i> 1998;128:337-45. [PMID: 9490593]	Does not include C282Y genotyping in screening sequence
Olsson KS, Eriksson K, Ritter B, Heedman PA. Screening for iron overload using transferrin saturation. <i>Acta Med Scand.</i> 1984;215:105-12. [PMID: 6702489]	Does not include C282Y genotyping in screening sequence

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Appendix Table 4—Continued

Study Citation	Reason for Exclusion
Olsson KS, Ritter B, Lundin PM. Liver affection in iron overload studied with serum ferritin and serum aminotransferases. <i>Acta Med Scand.</i> 1985;217:79-84. [PMID: 3976436]	Not a screening population
Olynyk JK, Luxon BA, Britton RS, Bacon BR. Hepatic iron concentration in hereditary hemochromatosis does not saturate or accurately predict phlebotomy requirements. <i>Am J Gastroenterol.</i> 1998;93:346-50. [PMID: 9517637]	Does not report relevant outcomes
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]	Study design
Phatak PD, Sham RL, Raubertas RF, Dunnigan K, O'Leary MT, Braggins C, et al. Prevalence of hereditary hemochromatosis in 16031 primary care patients. <i>Ann Intern Med.</i> 1998;129:954-61. [PMID: 9867748]	Does not include C282Y genotyping in screening sequence
Piperno A, Vergani A, Salvioni A, Trombini P, Vigana M, Riva A, et al. Effects of venesections and restricted diet in patients with the insulin-resistance hepatic iron overload syndrome. <i>Liver Int.</i> 2004;24:471-6. [PMID: 15482345]	Not a screening population
Porto G, Vicente C, Fraga J, da Silva BM, de Sousa M. Importance of establishing appropriate local reference values for the screening of hemochromatosis: a study of three different control populations and 136 hemochromatosis family members. Hemochromatosis Clinical and Research Group. <i>J Lab Clin Med.</i> 1992;119:295-305. [PMID: 1541878]	Includes data from patients < 18 y
Porto G, Vicente C, Teixeira MA, Martins O, Cabeda JM, Lacerda R, et al. Relative impact of HLA phenotype and CD4-CD8 ratios on the clinical expression of hemochromatosis. <i>Hepatology.</i> 1997;25:397-402. [PMID: 9021953]	Not a screening population
Poullis A, Moodie SJ, Ang L, Finlayson CJ, Levin GE, Maxwell JD. Routine transferrin saturation measurement in liver clinic patients increases detection of hereditary haemochromatosis. <i>Ann Clin Biochem.</i> 2003;40:521-7. [PMID: 14503989]	Not a screening population
Powell LW, Summers KM, Board PG, Axelsen E, Webb S, Halliday JW. Expression of hemochromatosis in homozygous subjects. Implications for early diagnosis and prevention. <i>Gastroenterology.</i> 1990;98:1625-32. [PMID: 2338199]	Includes data from patients < 18 y
Poynard T, Mathurin P, Lai CL, Guyader D, Poupon R, Tainturier MH, et al. A comparison of fibrosis progression in chronic liver diseases. <i>J Hepatol.</i> 2003;38:257-65. [PMID: 12586290]	Not a screening population
Press RD, Flora K, Gross C, Rabkin JM, Corless CL. Hepatic iron overload: direct HFE (HLA-H) mutation analysis vs quantitative iron assays for the diagnosis of hereditary hemochromatosis. <i>Am J Clin Pathol.</i> 1998;109:577-84. [PMID: 9576576]	Not a screening population
Rhodes DA, Raha-Chowdhury R, Cox TM, Trowsdale J. Homozygosity for the predominant Cys282Tyr mutation and absence of disease expression in hereditary haemochromatosis. <i>J Med Genet.</i> 1997;34:761-4. [PMID: 9321765]	Does not report relevant outcomes
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 report relevant outcomes
Rossi E, Henderson S, Chin CY, Olynyk J, Beilby JP, Reed WD, et al. Genotyping as a diagnostic aid in genetic haemochromatosis. <i>J Gastroenterol Hepatol.</i> 1999;14:427-30. [PMID: 10355506]	Not a screening population
Rowe JW, Wands JR, Mezey E, Waterbury LA, Wright JR, Tobin J, et al. Familial hemochromatosis: characteristics of the precirrhotic stage in a large kindred. <i>Medicine (Baltimore).</i> 1977;56:197-211. [PMID: 870791]	Does not report relevant outcomes
Ryan E, Byrnes V, Coughlan B, Flanagan AM, Barrett S, O'Keane JC, et al. Underdiagnosis of hereditary haemochromatosis: lack of presentation or penetration? <i>Gut.</i> 2002;51:108-12. [PMID: 12077102]	Includes data from patients < 18 y
Salonen JT, Tuomainen TP, Kontula K. Role of C282Y mutation in haemochromatosis gene in development of type 2 diabetes in healthy men: prospective cohort study. <i>BMJ.</i> 2000;320:1706-7. [PMID: 10864547]	Does not report relevant outcomes
Scotet V, Merour MC, Mercier AY, Chanu B, Le Faou T, Raguene O, et al. Hereditary hemochromatosis: effect of excessive alcohol consumption on disease expression in patients homozygous for the C282Y mutation. <i>Am J Epidemiol.</i> 2003;158:129-34. [PMID: 12851225]	Does not report relevant outcomes
Sham RL, Ou CY, Cappuccio J, Braggins C, Dunnigan K, Phatak PD. Correlation between genotype and phenotype in hereditary hemochromatosis: analysis of 61 cases. <i>Blood Cells Mol Dis.</i> 1997;23:314-20. [PMID: 9410475]	Not a screening population
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