

Reference	mutation (protein)	mutation (cDNA level)	age	sex	Hb (g/dl)	ferritin (µg/L)	transferrin saturation (%)	iron removed	Fibrosis (ISAHK)	Cirrhosis	HII	HIC (µmol/g)	MRI	viral hepatitis	steatosis	BMI	metabolic syndrome	Ethanol (g/week)	other comorbidity	Biopsied	Histology M = Kupffer cells exclusively or prominent H = both or hepatocellular only
Subramaniam GUT 05	A77D	c.230C>A	45	m	12.2	3340	29	n.a.	0	0	2.1	96		no	no	normal	neg	20		Kupffer cell iron loading, min Hep	M
Lim BCM2D 2008	A77D	c.230C>A	36	m		4000	45	yes	0			156,25		no	no		neg			prominent kupffer cell siderosis, grade 2 hep siderosis	M
Lim BCM2D 2008	A77D	c.230C>A	34	m		4050	54	yes	1			368,4		no	no					prominent kupffer cell siderosis, grade 3 hep siderosis	M
Pietrangolo NEJM 99	A77D	c.230C>A	61	f	14.5	5846	89	12	1		5.1	310		no	no			<70		mixed: hep, reticuloendothelial	H
Pietrangolo NEJM 99	A77D	c.230C>A	59	m	16	5750	75	35	1		10.9	646		no	no			<70		mixed: hep, reticuloendothelial	H
Pietrangolo NEJM 99	A77D	c.230C>A	30	m	14.3	5600	88	16	1		27.2	815		no	no			<70		mixed: hep, reticuloendothelial	H
Pietrangolo NEJM 99	A77D	c.230C>A	34	m	15.7	2410	60	31	0		23.8	810		no	no			<70		mixed: hep, reticuloendothelial	H
Pietrangolo NEJM 99	A77D	c.230C>A	20	m	15	2700	40	12	0		11	220		no	no			<70		mixed: hep, reticuloendothelial	H
Pietrangolo NEJM 99	A77D	c.230C>A	21	f	14.2	890	30	6	0		5.2	110		no	no			<70		mixed: hep, reticuloendothelial	H
Pietrangolo NEJM 99	A77D	c.230C>A	37	m	15	3250	65	20	0		28.4	1050		no	no			<70		mixed: hep, reticuloendothelial	H
Pietrangolo NEJM 99	A77D	c.230C>A	14	m	13.8	815	28	4	0		5.4	75		no	no			<70		mixed: hep, reticuloendothelial	H
Liu Int Med 05		c.117A>G	43	f		9660	92	yes	0				liver/spleen	no			imp Glc Tol	0		mixed: hep&Kupff	H
Sham BCM2D 05	C326S	c.977G>C	35	m				yes	0	1										marked Hep iron	H
Sham BCM2D 05	C326S	c.977G>C	16	f				yes	0		2.3									marked Hep iron	H
Sham BCM2D 05	C326S	c.977G>C	16	m				yes	0											marked Hep iron	H
Cemones BJH 05	D181V	c.846A>T	40	m		1400	40	yes	0											mixed: hep & kupffer	H
Lee ActHaem 2007		splice site (prec.c.1402G>A)	59	m		1364	25	yes	0						mild		DM, Hyperlipid.	0		mixed: hep & kupffer	H
Jouanolle JHep 03	G490D	c.1468G>A	54	f	14.2	8943	53	yes	0		6.4	347	1							mixed: hep & kupffer	H
De Domenico Haemat 06	G90S	c.238G>A	34	m	15.8	4420	60	n.a.	0			954								yes	H
Girelli JHep 08	I152F	c.758A>T	59	f		1771	22,7	yes	2				liver	HCV	mild	29	DM	no		Kupffer cell iron loading, min Hep	M
Girelli JHep 08	L233P	c.1012T>C	59	m		9000	74,8	yes	1				liver/spleen		grade 1			140		Hepatocellular iron	H
Njaou BCM2D 02	N144H	c.430A>G	76	n/a		219	40,4	no	0	1					yes			abuse		mixed	H
Njaou BCM2D 02	N144H	c.430A>G	64	n/a		223	34,4	na	0	1					yes	obesity				mixed	H
Njaou BCM2D 02	N144H	c.430A>G	63	n/a		113	17	yes	1		27.5									mixed	H
Njaou BCM2D 02	N144H	c.430A>G	80	n/a		133	27,9	yes	1		12,3									mixed	H
Njaou BCM2D 02	N144H	c.430A>G	69	n/a		35	34	yes	1		12,8						DM			mixed	H
Njaou BCM2D 02	N144H	c.430A>G	66	n/a				yes	1						yes		DM			mixed	H
Njaou BCM2D 02	N144H	c.430A>G	53	n/a		31	24,1	yes	0							obesity				mixed	H
Njaou BCM2D 02	N144H	c.430A>G	45	n/a		180	23,1	yes	1							obesity				mixed	H
Wallace JHep 04	N144D	c.430A>G	32	m		10510	99	yes	0		12,3	393	0						Sarcoidosis	Hepatocellular iron	H
Arden GUT 03	N144T	c.431A>C	48	m		2937	80	no	0		7,6	363								grade 4 hep iron, significant kupffer cell iron	M
De Domenico Haemat 06	N174I	c.521A>T	66	f	14,9	5815	48	n.a.	0			450								no hist info	
De Domenico Haemat 06	N174I	c.521A>T	38	f	13,4	5430	45	n.a.	0			850								no hist info	
Koyama Int Med 05	R495S	c.1467A>C	43	m		922	24,8	no	0					no			no	no		selective iron deposits in kupffer cells	M
Bach BCM2D 06	R88T	c.263G>C	61	m		9075	91	16,5	1	0				no			no	mild		severe iron deposits in both	H
Bach BCM2D 06	R88T	c.263G>C	56	m		2830	43	9,5	1	0	200									severe iron deposits in both	H
Bach BCM2D 06	R88T	c.263G>C	53	m		5291	70	17,7	1	0				HBV/HCV						severe iron deposits in both	H
Bach BCM2D 06	R88T	c.263G>C	23	m		1870	64	11	0											severe iron deposits in both	H
Wallace JHep 07	S338R	c.1014T>G	72	m		1990	90	yes	0		210	0			mild		imp. glc.tol	175		grade 4 iron in hep, Kupff a periportal macros	H
Cazzola BJH 02	V162del	c.484_486del3	31	f	12	1396	15	no	0											selective iron deposits in kupffer cells	M
Devalia Blood 02	V162del	c.484_486del3	38	f		2855	40	yes	0											heavy iron deposits in both, kupffer and hepat	H
Devalia Blood 02	V162del	c.484_486del3	34	f		1150	31	yes	0				1, liver and spleen							marked iron accumulation in kupffer cells	M
Rocetto Blood 02	V162del	c.484_486del3	26	f	11,5	1022	18	yes	0											positive especially in kupffer cells, with few hepatocyte granules	M
Wallace Blood 02	V162del	c.484_486del3	56	m		12000	81	no	1		8,3	475								prominent kupffer cell loading in addition to hepatocell iron	M
Wallace Blood 02	V162del	c.484_486del3	73	m		10000	81	no	1											prominent kupffer cell loading in addition to hepatocell iron	M
Wallace Blood 02	V162del	c.484_486del3	20	m		1768	35	yes	1		5,2									prominent kupffer cell loading in addition to hepatocell iron	M
Wallace Blood 02	V162del	c.484_486del3	19	f		1182	19	yes	0		3,5									prominent kupffer cell loading in addition to hepatocell iron	M
Wallace GUT 05	V162del	c.484_486del3	36	f		3145	29	yes	0	0	9,1	316,1	1, liver				no			iron is prominent in kupffer cells and hepatocytes	H
Zoller Hep 05	V162del	c.484_486del3	28	m	15,5	4935	23	no	0											predominantly in kupffer cells	M
Speletes BCM2D 08	V162del	c.484_486del3	31	f		2419	22,1	yes	1				1, liver							prominent iron in kupffer cells	M
Lim BCM2D 2008	V162del	c.484_486del3	65	m	15,2	4559	yes	0			0,80	52,1		neg						grade 3 siderosis in hepatocytes and prominent in kupffer cells	H
Pelucchi ClinGent 08	V72F	c.214G>T	58	m	15,7	1091	81	yes	2	0	2,7	160						regular drinks		largely hepatocytes but also kupffer cells	H
Pelucchi ClinGent 08	V72F	c.214G>T	48	m	15,4	956	73	yes	3		2,5	120								largely hepatocytes but also kupffer cells	H
Rivard BJH 03	Y64N	c.190T>C	56	m		1759	96	yes	1											iron deposition hepatocytes and kupffer cells	H
Rivard BJH 03	Y64N	c.190T>C	24	m		647	98	yes	0				1, liver							iron deposition hepatocytes and kupffer cells	H
Rivard BJH 03	Y64N	c.190T>C	30	f		176	47	no	0											iron deposition hepatocytes and kupffer cells	H
Cunat ClinChem 2007	G490S	c.1468G>A	71	f		2710	83	unknown	0			350								yes	M
Cunat ClinChem 2007	G490S	c.1468G>A	24	m		1262	32	unknown	0											yes	M
Cunat ClinChem 2007	R89G	c.262A>G	41	m		2200	yes	1				65								mixed1	M
Cunat ClinChem 2007	S UTR	c.[59_45del]	73	m		1400	80	yes	0			240								yes	M
Griffith	R489K	c.1466G>A	54	m		3400	yes	0	0											grade 1 hepatocytes strong kupffer cell	H
Lutocart BJH 2009	Y501C	c.1502A>G	35	m	15,3	642	94	no	0				1, liver no spleen	neg						significant hepatocellular iron overload, although kupffer cell was also evident	H