

SAMPLES 5A01-05/2018

DISPATCHED ON 22ND MAY 2018

UK NEQAS for H&I Scheme 5A - HFE Typing

METHODOLOGY

Lab. No.	Typing methods used	Primer / oligo source	Detection method used	Reference to primer / oligo sequences	Comments on HFE typing method	Other HFE mutations or associated polymorphisms
1						
4						
5	LightCycler 480 melt curve genotyping	Roche-TibMolBiol Lightmix kit	Fluorescence	Feder. J.N, et al., 1996 A novel MHC class I-like gene is mutated in patients with hereditary haemochromatosis. Nature Genetics. 13, pp.399-408.		
14	PCR-RFLP		Gel			
15	PCR-SSP	In-house	Gel			
16						
17						
19						
22	RT-PCR. FRET probe melting curve analysis	In-house	Fluorescence		Meadows et al., Rapid-Cycler Real-Time-PCR, Springer (2001) 127-134	
30	HotStart RT-PCR	Commercial	Fluorescence			
33						
34	PCR-SSP	In-house	Gel		Gutteridge et al, 1997 Vox Sanguinis	
35						
36	PCR	Commercial	Fluorescence			
37						
39	PCR-SSP	In-house	Gel			
42	PCR-SSP	Commercial	Gel			
43	RT-PCR (melt curve analysis)	Commercial	Fluorescence			
48	PCR-SSP	In-house	Gel			
49	Allelic Discrimination	Commercial	Fluorescence			
50						
55	PCR-SSP	Commercial	Gel			
56						
59	PCR-RE	In-house	Gel			
62	PCR-SSP	Commercial	Gel			
64						
65	Allelic Discrimination	Commercial	Fluorescence			
70	RT-PCR	Commercial	Fluorescence			
74	PCR and Sequencing					
79	Taqman	In-house	RT-PCR			
80	PCR LightCycler, Melt curve analysis	Commercial	Fluorescence	Genes 4U C282Y + H63D/S65C Toolsets		
81	RT-PCR. FRET analysis	In-house	Fluorescence			
82						
84						
85						
86						
88						
89						
92	PCR LightCycler-Roche Melting Curve Analysis	Commercial	Fluorescence	HFE Duplex Tib Mol Biol		
94	Melt curve analysis	In-house	LightScanner	Zhou et al. (2004) Clinical Chemistry 50:1328-1335		
95	RT-PCR	Commercial	Melting curve analysis			
96						
97						
99						
108	RT-PCR	Commercial	Fluorescence			
123						
138	RT-PCR		Fluorescence			
150	Sequencing	Commercial				
154	PCR-SSO	Commercial	Hybridization on membrane			
269						
270						
282						
288						
314						
331	RT-PCR	Commercial	Fluorescence	Jacques B. et al, Human mutation 19:554-559, 2002		
344						
369	Loop-mediated isothermal amplification	Commercial	Fluorescence	Primers included in the commercial kit (LaCAR Mdx).		

IVS3+1G/T
IVS3+1G/T

V53M, V59M, H63H, Q127H, P160delC, E168Q, E168X, E169X, Q283P, E60X, M172K, Y250X, AVAQ594-597del, N144H, V162del

NEQ-125

Issue 2

UK NEQAS for H&I is operated by Velindre NHS Trust, a UKAS proficiency testing provider No 8351.

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RESULTS

Consensus:			HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	ss	Date Received	Date Tested	Comments
Lab No.	Assessment		Codon 63	Codon 282	Codon 65	Codon 63	Codon 282	Codon 65	Codon 63	Codon 282	Codon 65	Codon 63	Codon 282	Codon 65	Codon 63	Codon 282	Codon 65			
	63 282 65		5A01/2018	5A01/2018	5A01/2018	5A02/2018	5A02/2018	5A02/2018	5A03/2018	5A03/2018	5A03/2018	5A04/2018	5A04/2018	5A04/2018	5A05/2018	5A05/2018	5A05/2018			
1	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	30-May	
4	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	05-Jun	
5	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS		30-May	
14	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	30-May	
15	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY			05-Jun	
16	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	24-May	30-May	
17	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		24-May	30-May	
19	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	25-May	
22	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	29-May	
30	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	08-Jun	08-Jun	
33	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS			
34	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY			04-Jun	
35	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	24-May	
36	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	25-May	29-May	
37	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	04-Jun	
39	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY			05-Jun	
42	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY			05-Jun	
43	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	01-Jun	
48	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY			04-Jun	
49	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY			31-May	
50	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY				
55	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	23-May	
56	YES YES		HH	YY		HH	CC		HH	CC		HH	CC	Failed	HH	YY		23-May	31-May	
59	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		24-May	24-May	
62	YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS		05-Jun	
64	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	31-May	
65	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY			01-Jun	
70	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	22-May	05-Jun	
74	YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	30-May	
79	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		24-May	29-May	
80	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		25-May	31-May	
81	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	30-May	
82	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		25-May	31-May	
84	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	04-Jun	
85	YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	30-May	
86	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	29-May	
88	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	29-May	
89	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		28-May	07-Jun	
92	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	25-May	04-Jun	
94	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	24-May	30-May	
95	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS		07-Jun	
96	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		24-May	24-May	
97	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY				
99	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	24-May	
108	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY			04-Jun	
123	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	30-May	
138	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	25-May	
150	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	24-May	28-May	
154	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS		04-Jun	
269	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS			
270	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	29-May	
282	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS			
288	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	23-May	29-May	
314	YES YES		NT	NT		NT	NT		NT	NT		NT	NT		NT	NT				Sample delivery delay
331	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS			
344	YES YES YES		HH	YY	SS	HH	CC	SS	HH	CC	SS	HH	CC	SS	HH	YY	SS	22-May	24-May	
369	YES YES		HH	YY		HH	CC		HH	CC		HH	CC		HH	YY		23-May	28-May	

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