

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

SAMPLES 5A06-10/2018

DISPATCHED ON 06TH NOVEMBER 2018

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				IVS3+1G/T		
59	PCR-RE	In-house	Gel			
62	PCR-SSP	Commercial	Gel	IVS3+1G/T		
64						
65	Allelic Discrimination	Commercial	Fluorescence			
70	RT-PCR	Commercial	Fluorescence			
74	PCR and Sequencing					
79	Taqman	In-house	RT-PCR	Genes 4U C282Y + H63D/S65C Toolsets		
80	PCR LightCycler, Melt curve analysis	Commercial	Fluorescence			
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 Zhou et al. (2004) Clinical Chemistry 50:1328-1335		
94	Melt curve analysis	In-house	LightScanner			
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		V53M, V59M, H63H, Q127H, P160delC, E168Q, E168X, E169X, Q283P, E60X, M172K, Y250X, AVAQ594-597del, N144H, V162del	
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).		
412						

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RESULTS

Consensus: HH CC SS HH CY SS DD CC SS HH YY SS HH CC SS

Lab. No.	Assessment	Codon 63			Codon 282			Codon 65			Codon 63			Codon 282			Codon 65			Date Received	Date Tested	Comments
		5A06/2018	5A06/2018	5A06/2018	5A07/2018	5A07/2018	5A07/2018	5A08/2018	5A08/2018	5A08/2018	5A09/2018	5A09/2018	5A09/2018	5A10/2018	5A10/2018	5A10/2018						
1	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
4	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				08-Nov	15-Nov		
5	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS						
14	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	08-Nov		
15	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
16	YES YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				04-Nov	15-Nov		
17	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	08-Nov		
19	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	13-Nov		
22	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			08-Nov	12-Nov		
30	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			12-Nov	13-Nov		
33	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS						
34	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
35	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
36	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			07-Nov	15-Nov		
37	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				14-Nov	20-Nov		
39	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				08-Nov	12-Nov		
42	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
43	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS						
48	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
49	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
50	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
55	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				08-Nov	13-Nov		
56	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	18-Nov		
59	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	12-Nov		
62	YES YES	HH	CC	SS	HH	CY	SS	DD	CC	NT	HH	YY	SS	HH	CC	SS						
64	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	15-Nov		
65	YES YES	HH	CC		HH	CY		NT	CC		HH	YY		HH	CC						H63D mutation was not tested for sample 5A 08/2018 due to a test fail.	
70	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			07-Nov	16-Nov		
74	YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS						
79	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				08-Nov	09-Nov		
80	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
81	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			07-Nov	09-Nov		
82	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				08-Nov	14-Nov		
84	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS						
85	YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			09-Nov	15-Nov		
86	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
88	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	14-Nov		
89	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				13-Nov	14-Nov		
92	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			08-Nov	15-Nov		
94	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			08-Nov	09-Nov		
95	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			07-Nov	15-Nov		
96	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				08-Nov	15-Nov		
97	YES YES	NT	NT		NT	NT		NT	NT		NT	NT		NT	NT						All current clinical cases referred	
99	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	08-Nov		
108	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
123	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	08-Nov		
138	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS						
150	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			07-Nov	12-Nov		
154	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			07-Nov	09-Nov		
269	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS						
270	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			07-Nov	15-Nov		
282	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS						
288	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS						
314	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC							
331	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			09-Nov	13-Nov		
344	YES YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			07-Nov	09-Nov		
369	YES YES	HH	CC		HH	CY		DD	CC		HH	YY		HH	CC				07-Nov	12-Nov		
412	YES YES	HH	CC	SS	HH	CY	SS	DD	CC	SS	HH	YY	SS	HH	CC	SS			09-Nov	14-Nov		