



Listing and evaluation of the results

1: Dr. med.Max Mustermann
Laboratory Mustermann

Survey of 04 November 2020

Adviser:

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Set 01 - Lactase-gene (LCT)

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
Lactase-Gene (LCT) (T -13910 C)	61	269	ZY		- 13910 CC	- 13910 CC	M	+
	62				T - 13910C	T - 13910C	M	+

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Set 04 - CYP21A2 21-Hydroxylase

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
Cyp21A2 gene (Del 8bp)	61	266	AP		No Del = wt	No Del = wt	M	+
	62				No Del = wt	No Del = wt	M	+
Cyp21A2 gene (E6 Cluster)	61	266	AP		No Mut = wt	No Mut = wt	M	+
	62				No Mut = wt	No Mut = wt	M	+
Cyp21A2 gene (F306 +T)	61	266	AP		No +T = wt	No +T = wt	M	+
	62				No +T = wt	No +T = wt	M	+
Cyp21A2 gene (I 172 N)	61	266	AP		I(I) 172 = wt	I(I) 172 = wt	M	+
	62				I(I) 172 = wt	I(I) 172 = wt	M	+
Cyp21A2 gene (I2 G)	61	266	AP		G(G)	G(G)	M	+
	62				A/C(A/C) =wt	A/C(A/C) =wt	M	+
Cyp21A2 gene (P 30 L)	61	266	AP		P(P)30 = wt	P(P)30 = wt	M	+
	62				P(P)30 = wt	P(P)30 = wt	M	+
Cyp21A2 gene (P 453 S)	61	266	AP		P(P) 453=wt	P(P) 453=wt	M	+
	62				P(P) 453=wt	P(P) 453=wt	M	+
Cyp21A2 gene (Q 318 X)	61	266	AP		Q(Q) 318=wt	Q(Q) 318=wt	M	+
	62				Q(Q) 318=wt	Q(Q) 318=wt	M	+
Cyp21A2 gene (R 356 W)	61	266	AP		R(R) 356=wt	R(R) 356=wt	M	+
	62				R(R) 356=wt	R(R) 356=wt	M	+
Cyp21A2 gene (V 281 L)	61	266	AP		V(V) 281 =wt	V(V) 281 =wt	M	+
	62				V(V) 281 =wt	V(V) 281 =wt	M	+

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Set 06 - CYP2C9 CYP2C19 VKORC1 TPMT

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Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
CYP2C19*2 Allele: G 681 A	61	266	AP		681AA	681AA	M	+
	62				GG681	GG681	M	+
CYP2C19*3 Allele: G 636 A	61	266	AP		GG636	GG636	M	+
	62				GG636	GG636	M	+
CYP2C9*2 Allele: C 430 T	61	266	AP		CC 430	CC 430	M	+
	62				CC 430	CC 430	M	+
CYP2C9*3 Allele: A 1075 C	61	266	AP		AA 1075	AA 1075	M	+
	62				AA 1075	AA 1075	M	+
TPMT (nt. A 719 G)	61	269	ZY		WT	WT	M	+
	62				HET	HET	M	+
TPMT (nt. G 460 A)	61	269	ZY		WT	WT	M	+
	62				HET	HET	M	+
TPMT*2 Allele (nt. G 238 C)	61	269	ZY		WT	WT	M	+
	62				WT	WT	M	+

** BRAVO **

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Set 08 - CYP2D6 DPD IL28B

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
CYP2D6 Gene Duplication	61	271	AP		Duplication	Duplication	M	+
	62				No Duplication	No Duplication	M	+
CYP2D6*3: A2637 del	61	266	AP		No deletion	No deletion	M	+
	62				No deletion	No deletion	M	+
CYP2D6*4: G 1934 A	61	266	AP		GG1934 wt	GG1934 wt	M	+
	62				GG1934 wt	GG1934 wt	M	+
CYP2D6*5: complete Deletion	61	271	AP		No deletion	No deletion	M	+
	62				Del HET	No deletion	M	-
CYP2D6*6: T1795 del	61	266	AP		No deletion	No deletion	M	+
	62				No deletion	No deletion	M	+
DPD (exon 14 skipping)	61	266	ZY		No e14s (WT)	No e14s (WT)	M	+
	62				e14s Het	e14s Het	M	+

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Set 09 - CFTR Common Mutations

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
CFTR Gen (1677 delTA)	61	271	ZY		No del / No del = wt	No del / No del = wt	M	+
	62				No del / No del = wt	No del / No del = wt	M	+
CFTR Gen (2143 del T)	61	271	ZY		No del / No del = wt	No del / No del = wt	M	+
	62				No del / No del = wt	No del / No del = wt	M	+
CFTR Gen (2184 delA)	61	271	ZY		No del / No del = wt	No del / No del = wt	M	+
	62				No del / No del = wt	No del / No del = wt	M	+
CFTR Gen (3905insT)	61	271	ZY		No ins / No ins = wt	No ins / No ins = wt	M	+
	62				No ins / No ins = wt	No ins / No ins = wt	M	+
CFTR Gen (A455E)	61	271	ZY		AA 455 = wt	AA 455 = wt	M	+
	62				AA 455 = wt	AA 455 = wt	M	+
CFTR Gen (E 60 X)	61	271	ZY		EE 60 = wt	EE 60 = wt	M	+
	62				EE 60 = wt	EE 60 = wt	M	+
CFTR Gen (E 92 X)	61	271	ZY		EE 92 = wt	EE 92 = wt	M	+
	62				EE 92 = wt	EE 92 = wt	M	+
CFTR Gen (Intron 8)	61	271	ZY		7T / 7T	7T / 7T	M	+
	62				9T / 7T	9T / 7T	M	+
CFTR Gen (delta F 508)	61	271	ZY		Wild Type	Wild Type	M	+
	62				Wild Type	Wild Type	M	+

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Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
CFTR gene (1078 del T)	61	271	ZY		No del / No del = wt	No del / No del = wt	M	+
	62				No del / No del = wt	No del / No del = wt	M	+
CFTR gene (1717-1 G>A)	61	271	ZY		GG = wt	GG = wt	M	+
	62				GG = wt	GG = wt	M	+
CFTR gene (2183A A>G)	61	271	ZY		AA / AA = wt	AA / AA = wt	M	+
	62				AA / AA = wt	AA / AA = wt	M	+
CFTR gene (2184 ins A)	61	271	ZY		No ins / No ins =wt	No ins / No ins =wt	M	+
	62				No ins / No ins =wt	No ins / No ins =wt	M	+
CFTR gene (2789+5 G>A)	61	271	ZY		GG = wt	GG = wt	M	+
	62				GG = wt	GG = wt	M	+
CFTR gene (3272-26 A>G)	61	271	ZY		AA = wt	AA = wt	M	+
	62				AA = wt	AA = wt	M	+
CFTR gene (3659 del C)	61	271	ZY		No del / No del = wt	No del / No del = wt	M	+
	62				No del / No del = wt	No del / No del = wt	M	+
CFTR gene (3849+10kb C>T)	61	271	ZY		CC = wt	CC = wt	M	+
	62				CC = wt	CC = wt	M	+
CFTR gene (621+1 G>T)	61	271	ZY		GG = wt	GG = wt	M	+
	62				GG = wt	GG = wt	M	+
CFTR gene (G 542 X)	61	271	ZY		GG 542 = wt	GG 542 = wt	M	+
	62				G 542 X	G 542 X	M	+
CFTR gene (G 551 D)	61	271	ZY		GG 551 = wt	GG 551 = wt	M	+
	62				GG 551 = wt	GG 551 = wt	M	+
CFTR gene (G 85 E)	61	271	ZY		GG 85 = wt	GG 85 = wt	M	+
	62				GG 85 = wt	GG 85 = wt	M	+
CFTR gene (I 336 K)	61	271	ZY		II 336 = wt	II 336 = wt	M	+
	62				II 336 = wt	II 336 = wt	M	+
CFTR gene (I 507 del)	61	271	ZY		No del / No del = wt	No del / No del = wt	M	+
	62				No del / No del = wt	No del / No del = wt	M	+
CFTR gene (M 1101 K)	61	271	ZY		MM 1101 = wt	MM 1101 = wt	M	+
	62				MM 1101 = wt	MM 1101 = wt	M	+
CFTR gene (N 1303 K)	61	271	ZY		NN 1303 =wt	NN 1303 =wt	M	+
	62				NN 1303 =wt	NN 1303 =wt	M	+
CFTR gene (R 1162 X)	61	271	ZY		RR 1162 = wt	RR 1162 = wt	M	+
	62				RR 1162 = wt	RR 1162 = wt	M	+
CFTR gene (R 117 H)	61	271	ZY		RR 117 = wt	RR 117 = wt	M	+
	62				RR 117 = wt	RR 117 = wt	M	+
CFTR gene (R 334 W)	61	271	ZY		RR 334 = wt	RR 334 = wt	M	+
	62				RR 334 = wt	RR 334 = wt	M	+
CFTR gene (R 347 P/H)	61	271	ZY		RR 347 = wt	RR 347 = wt	M	+
	62				RR 347 = wt	RR 347 = wt	M	+
CFTR gene (R 553 X)	61	271	ZY		RR 553 = wt	RR 553 = wt	M	+
	62				RR 553 = wt	RR 553 = wt	M	+
CFTR gene (W 1282 X)	61	271	ZY		WW 1282 = wt	WW 1282 = wt	M	+
	62				WW 1282 = wt	WW 1282 = wt	M	+
CFTR gene (Y 1092 X (C>A))	61	271	ZY		YY 1092 = wt	YY 1092 = wt	M	+
	62				YY 1092 = wt	YY 1092 = wt	M	+
CFTR gene (dele2,3 (21kb))	61	271	ZY		No Del = wt	No Del = wt	M	+
	62				No Del = wt	No Del = wt	M	+


** BRAVO **

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781 Set 12 - FMR1 Fragile X

Analyte	Sample	Unit	Your value	Target value	TV-Type	Lower limit	Upper limit	Deviation	Z-Score	Meets criteria	
Allele 1: FMR1 CGG repeats in 5' UTR	61		30.0	30.0	ET	27.0	33.0	0.0%	0	+	
	62		>200		ET	>=200			0	+	

Analyte	Sample	Your unit	Stated value	Conversion factor	Method	Manufacturer	Device
Allele 1: FMR1 CGG repeats in 5' UTR	61		30.0	1.00	9999	ZY	
	62		>200	1.00			

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
FMR1 gene - evaluation / Allele 1	61	9999	ZY		Normal	Normal	M	+
	62				Full Mutation	Full Mutation	M	+
Global evaluation	61	9999	ZY		Normal	Normal	M	+
	62				Abnormal	Abnormal	M	+





782 Set 13 - GJB2 Connexin 26

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
GJB2 35del G (Connexin 26)	61	266	AP		No del / No del	No del / No del	M	+
	62				35delG / No del	35delG / No del	M	+

783 Set 14 - MSH2 MLH1 (HNPCC)

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
MLH1 mutation type	61	266	ZY		Wild Type	Wild Type	M	+
	62				c.551C>A	c.551C>A	M	+
MSH2 mutation type	61	266	ZY		Wild Type	Wild Type	M	+
	62				Wild Type	Wild Type	M	+

784 Set 15 - HTT Huntingtin

Analyte	Sample	Unit	Your value	Target value	TV-Type	Lower limit	Upper limit	Deviation	Z-Score	Meets criteria	
HTT Gen - CAG repeats allele 1	61		17.0	17.0	ET	14.0	20.0	0.0%	0	+	
	62		20.0	20.0	ET	17.0	23.0	0.0%	0	+	
HTT Gen - CAG repeats allele 2	61		33.0	33.0	ET	30.0	36.0	0.0%	0	+	
	62		32.0	32.0	ET	29.0	35.0	0.0%	0	+	

Analyte	Sample	Your unit	Stated value	Conversion factor	Method	Manufacturer	Device
HTT Gen - CAG repeats allele 1	61		17.0	1.00	9999	ZY	
	62		20.0	1.00			
HTT Gen - CAG repeats allele 2	61		33.0	1.00	9999	ZY	
	62		32.0	1.00			



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Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
HTT Gen - evaluation allele 1	61	9999	ZY		Normal	Normal	M	+
	62				Normal	Normal	M	+
HTT Gen - evaluation allele 2	61	9999	ZY		Intermediate	Intermediate	M	+
	62				Intermediate	Intermediate	M	+

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Set 17 - SMN1 (Spinal Muscular Atrophy)

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
SMN1 Gen - exon 7 deletion	61	9999	ZY		No deletion	No deletion	M	+
	62				Del HOM	Del HOM	M	+
SMN1 Gen - exon 8 deletion	61	9999	ZY		No deletion	No deletion	M	+
	62				Del HOM	Del HOM	M	+

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Set 18 - AZF (Y-Chromosome Microdeletions)

Analyte	Sample	Method	Manufacturer	Device	Your specification(s)	Correct specification(s)	TV-Type	Meets criteria
Y-Chromosome AZFa deletion	61	271	ZY		No deletion	No deletion	M	+
	62				No deletion	No deletion	M	+
Y-Chromosome AZFb deletion	61	271	ZY		Deletion	Deletion	M	+
	62				No deletion	No deletion	M	+
Y-Chromosome AZFc deletion	61	271	ZY		Deletion	Deletion	M	+
	62				No deletion	No deletion	M	+

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Individual summary of results

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770 Set 01 - Lactase-gene (LCT)

Lactase-Gene (LCT) (T -13910 C) (N = 69, Rate of success: 97,1%)

Sample 61

Collective	- 13910 CC	total
all methods	69 ●	69

Sample 62

Collective	TT -13910	T - 13910C	total
all methods	1	68 ●	69

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

773 Set 04 - CYP21A2 21-Hydroxylase

Cyp21A2 gene (Del 8bp) (N = 16, Rate of success: 100%)

Sample 61

Collective	No Del = wt	total
all methods	16 ●	16

Sample 62

Collective	No Del = wt	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Cyp21A2 gene (E6 Cluster) (N = 16, Rate of success: 100%)

Sample 61

Collective	No Mut = wt	total
all methods	16 ●	16

Sample 62

Collective	No Mut = wt	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Cyp21A2 gene (F306 +T) (N = 16, Rate of success: 100%)

Sample 61

Collective	No +T = wt	total
all methods	16 ●	16

Sample 62

Collective	No +T = wt	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Cyp21A2 gene (I 172 N) (N = 16, Rate of success: 100%)

Sample 61

Collective	I(I) 172 = wt	total
all methods	16 ●	16

Sample 62

Collective	I(I) 172 = wt	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective



Cyp21A2 gene (I2 G) (N = 16, Rate of success: 93,8%)

Sample 61

Collective	A/C & G	G(G)	total
all methods	1	15 ●	16

Sample 62

Collective	A/C(A/C) =wt	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Cyp21A2 gene (P 30 L) (N = 16, Rate of success: 100%)

Sample 61

Collective	P(P)30 = wt	total
all methods	16 ●	16

Sample 62

Collective	P(P)30 = wt	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Cyp21A2 gene (P 453 S) (N = 16, Rate of success: 100%)

Sample 61

Collective	P(P) 453=wt	total
all methods	16 ●	16

Sample 62

Collective	P(P) 453=wt	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Cyp21A2 gene (Q 318 X) (N = 16, Rate of success: 100%)

Sample 61

Collective	Q(Q) 318=wt	total
all methods	16 ●	16

Sample 62

Collective	Q(Q) 318=wt	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Cyp21A2 gene (R 356 W) (N = 16, Rate of success: 100%)

Sample 61

Collective	R(R) 356=wt	total
all methods	16 ●	16

Sample 62

Collective	R(R) 356=wt	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Cyp21A2 gene (V 281 L) (N = 16, Rate of success: 93,8%)

Sample 61

Collective	V(V) 281=wt	total
all methods	16 ●	16

Sample 62

Collective	V(V) 281 =wt	V 281 L	total
all methods	15 ●	1	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective



Set 06 - CYP2C9 CYP2C19 VKORC1 TPMT

CYP2C19*2 Allele: G 681 A (N = 23, Rate of success: 91,3%)

Sample 61

Collective	GG681	G681A	681AA	total
all methods	1	1	21 ●	23

Sample 62

Collective	GG681	681AA	total
all methods	22 ●	1	23

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CYP2C19*3 Allele: G 636 A (N = 23, Rate of success: 100%)

Sample 61

Collective	GG636	total
all methods	23 ●	23

Sample 62

Collective	GG636	total
all methods	23 ●	23

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CYP2C9*2 Allele: C 430 T (N = 23, Rate of success: 100%)

Sample 61

Collective	CC 430	total
all methods	23 ●	23

Sample 62

Collective	CC 430	total
all methods	23 ●	23

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CYP2C9*3 Allele: A 1075 C (N = 23, Rate of success: 100%)

Sample 61

Collective	AA 1075	total
all methods	23 ●	23

Sample 62

Collective	AA 1075	total
all methods	23 ●	23

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

TPMT (nt. A 719 G) (N = 22, Rate of success: 95,5%)

Sample 61

Collective	WT	total
all methods	22 ●	22

Sample 62

Collective	WT	HET	total
all methods	1	21 ●	22

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

TPMT (nt. G 460 A) (N = 21, Rate of success: 95,2%)

Sample 61

Collective	WT	total
all methods	21 ●	21

Sample 62

Collective	WT	HET	total
all methods	1	20 ●	21

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

TPMT*2 Allele (nt. G 238 C) (N = 21, Rate of success: 100%)
Sample 61

Collective	WT	total
all methods	21 ●	21

Sample 62

Collective	WT	total
all methods	21 ●	21

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

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Set 08 - CYP2D6 DPD IL28B
CYP2D6 Gene Duplication (N = 10, Rate of success: 90%)
Sample 61

Collective	No Duplication	Duplication	total
all methods	1	9 ●	10

Sample 62

Collective	No Duplication	total
all methods	10 ●	10

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CYP2D6*3: A2637 del (N = 16, Rate of success: 100%)
Sample 61

Collective	No deletion	total
all methods	16 ●	16

Sample 62

Collective	No deletion	total
all methods	16 ●	16

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CYP2D6*4: G 1934 A (N = 17, Rate of success: 100%)
Sample 61

Collective	GG1934 wt	total
all methods	17 ●	17

Sample 62

Collective	GG1934 wt	total
all methods	17 ●	17

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CYP2D6*5: complete Deletion (N = 11, Rate of success: 90,9%)
Sample 61

Collective	No deletion	total
all methods	11 ●	11

Sample 62

Collective	No deletion	Del HET	total
all methods	10 ●	1	11

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CYP2D6*6: T1795 del (N = 14, Rate of success: 100%)
Sample 61

Collective	No deletion	total
all methods	14 ●	14

Sample 62

Collective	No deletion	total
all methods	14 ●	14

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective



DPD (exon 14 skipping) (N = 26, Rate of success: 100%)

Sample 61

Collective	No e14s (WT)	total
all methods	26 ●	26

Sample 62

Collective	e14s Het	total
all methods	26 ●	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

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Set 09 - CFTR Common Mutations

CFTR Gen (1677 delTA) (N = 26, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt	total
all methods	26 ●	26

Sample 62

Collective	No del / No del = wt	total
all methods	26 ●	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR Gen (2143 del T) (N = 28, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt	total
all methods	28 ●	28

Sample 62

Collective	No del / No del = wt	total
all methods	28 ●	28

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR Gen (2184 delA) (N = 27, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt	total
all methods	27 ●	27

Sample 62

Collective	No del / No del = wt	total
all methods	27 ●	27

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR Gen (3905insT) (N = 26, Rate of success: 100%)

Sample 61

Collective	No ins / No ins = wt	total
all methods	26 ●	26

Sample 62

Collective	No ins / No ins = wt	total
all methods	26 ●	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR Gen (A455E) (N = 26, Rate of success: 100%)

Sample 61

Collective	AA 455 = wt	total
all methods	26 ●	26

Sample 62

Collective	AA 455 = wt	total
all methods	26 ●	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR Gen (E 60 X) (N = 26, Rate of success: 100%)
Sample 61

Collective	EE 60 = wt	total
all methods	26 ●	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	EE 60 = wt	total
all methods	26 ●	26

CFTR Gen (E 92 X) (N = 21, Rate of success: 100%)
Sample 61

Collective	EE 92 = wt	total
all methods	21 ●	21

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	EE 92 = wt	total
all methods	21 ●	21

CFTR Gen (Intron 8) (N = 23, Rate of success: 91,3%)
Sample 61

Collective	9T / 7T	7T / 7T	total
all methods	1	22 ●	23

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	9T / 7T	7T / 7T	total
all methods	22 ●	1	23

CFTR Gen (delta F 508) (N = 32, Rate of success: 100%)
Sample 61

Collective	Wild Type	total
all methods	32 ●	32

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	Wild Type	total
all methods	32 ●	32

CFTR gene (1078 del T) (N = 27, Rate of success: 100%)
Sample 61

Collective	No del / No del = wt	total
all methods	27 ●	27

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	No del / No del = wt	total
all methods	27 ●	27

CFTR gene (1717-1 G>A) (N = 29, Rate of success: 100%)
Sample 61

Collective	GG = wt	total
all methods	29 ●	29

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	GG = wt	total
all methods	29 ●	29

CFTR gene (2183A A>G) (N = 26, Rate of success: 96,2%)
Sample 61

Collective	AA / AA = wt	AA / G	total
all methods	25 ●	1	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	AA / AA = wt	AA / G	total
all methods	25 ●	1	26

CFTR gene (2184 ins A) (N = 23, Rate of success: 100%)
Sample 61

Collective	No ins / No ins =wt	total
all methods	23 ●	23

Sample 62

Collective	No ins / No ins =wt	total
all methods	23 ●	23

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (2789+5 G>A) (N = 29, Rate of success: 100%)
Sample 61

Collective	GG = wt	total
all methods	29 ●	29

Sample 62

Collective	GG = wt	total
all methods	29 ●	29

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (3272-26 A>G) (N = 28, Rate of success: 100%)
Sample 61

Collective	AA = wt	total
all methods	28 ●	28

Sample 62

Collective	AA = wt	total
all methods	28 ●	28

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (3659 del C) (N = 27, Rate of success: 100%)
Sample 61

Collective	No del / No del = wt	total
all methods	27 ●	27

Sample 62

Collective	No del / No del = wt	total
all methods	27 ●	27

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (3849+10kb C>T) (N = 29, Rate of success: 100%)
Sample 61

Collective	CC = wt	total
all methods	29 ●	29

Sample 62

Collective	CC = wt	total
all methods	29 ●	29

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (621+1 G>T) (N = 30, Rate of success: 100%)
Sample 61

Collective	GG = wt	total
all methods	30 ●	30

Sample 62

Collective	GG = wt	total
all methods	30 ●	30

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (G 542 X) (N = 31, Rate of success: 87,1%)
Sample 61

Collective	GG 542 = wt	G 542 X	total
all methods	30 ●	1	31

Sample 62

Collective	GG 542 = wt	G 542 X	total
all methods	3	28 ●	31

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective



CFTR gene (G 551 D) (N = 30, Rate of success: 100%)

Sample 61

Collective	GG 551 = wt	total
all methods	30 ●	30

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	GG 551 = wt	total
all methods	30 ●	30

CFTR gene (G 85 E) (N = 29, Rate of success: 100%)

Sample 61

Collective	GG 85 = wt	total
all methods	29 ●	29

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	GG 85 = wt	total
all methods	29 ●	29

CFTR gene (I 336 K) (N = 22, Rate of success: 100%)

Sample 61

Collective	II 336 = wt	total
all methods	22 ●	22

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	II 336 = wt	total
all methods	22 ●	22

CFTR gene (I 507 del) (N = 30, Rate of success: 100%)

Sample 61

Collective	No del / No del = wt	total
all methods	30 ●	30

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	No del / No del = wt	total
all methods	30 ●	30

CFTR gene (M 1101 K) (N = 23, Rate of success: 100%)

Sample 61

Collective	MM 1101 = wt	total
all methods	23 ●	23

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	MM 1101 = wt	total
all methods	23 ●	23

CFTR gene (N 1303 K) (N = 30, Rate of success: 100%)

Sample 61

Collective	NN 1303 = wt	total
all methods	30 ●	30

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	NN 1303 = wt	total
all methods	30 ●	30

CFTR gene (R 1162 X) (N = 28, Rate of success: 100%)

Sample 61

Collective	RR 1162 = wt	total
all methods	28 ●	28

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	RR 1162 = wt	total
all methods	28 ●	28

CFTR gene (R 117 H) (N = 24, Rate of success: 100%)
Sample 61

Collective	RR 117 = wt	total
all methods	24 ●	24

Sample 62

Collective	RR 117 = wt	total
all methods	24 ●	24

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (R 334 W) (N = 28, Rate of success: 100%)
Sample 61

Collective	RR 334 = wt	total
all methods	28 ●	28

Sample 62

Collective	RR 334 = wt	total
all methods	28 ●	28

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (R 347 P/H) (N = 29, Rate of success: 100%)
Sample 61

Collective	RR 347 = wt	total
all methods	29 ●	29

Sample 62

Collective	RR 347 = wt	total
all methods	29 ●	29

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (R 553 X) (N = 30, Rate of success: 96,7%)
Sample 61

Collective	RR 553 = wt	total
all methods	30 ●	30

Sample 62

Collective	RR 553 = wt	R 553 X	total
all methods	29 ●	1	30

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (W 1282 X) (N = 28, Rate of success: 100%)
Sample 61

Collective	WW 1282 = wt	total
all methods	28 ●	28

Sample 62

Collective	WW 1282 = wt	total
all methods	28 ●	28

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (Y 1092 X (C>A)) (N = 25, Rate of success: 100%)
Sample 61

Collective	YY 1092 = wt	total
all methods	25 ●	25

Sample 62

Collective	YY 1092 = wt	total
all methods	25 ●	25

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

CFTR gene (dele2,3 (21kb)) (N = 29, Rate of success: 100%)
Sample 61

Collective	No Del = wt	total
all methods	29 ●	29

Sample 62

Collective	No Del = wt	total
all methods	29 ●	29

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective



781

Set 12 - FMR1 Fragile X

Allele 1: FMR1 CGG repeats in 5' UTR (N = 12)										
Collective	Sample	Target value	Target range	Participants collective			Rate (%)			
				AVG	CV	Num.	Sam.	total		
all methods	61	30.0	27.0 - 33.0	30.0	0.000	12	100	66.7		
	62		>=200 -	200	0.000	12	66.7			

Rate of success: 66,7%

Graphical representation not useful

781

Set 12 - FMR1 Fragile X

FMR1 gene - evaluation / Allele 1 (N = 12, Rate of success: 75%)

Sample 61

Collective	Normal	total
all methods	11 ●	11

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	Premutation	Full Mutation	total
all methods	1	10 ●	11

Global evaluation (N = 12, Rate of success: 91,7%)

Sample 61

Collective	Normal	total
all methods	12 ●	12

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	Abnormal	total
all methods	11 ●	11

782

Set 13 - GJB2 Connexin 26

GJB2 35del G (Connexin 26) (N = 14, Rate of success: 100%)

Sample 61

Collective	No del / No del	total
all methods	14 ●	14

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	35delG / No del	total
all methods	14 ●	14

783

Set 14 - MSH2 MLH1 (HNPCC)

MLH1 mutation type (N = 11, Rate of success: 100%)

Sample 61

Collective	Wild Type	total
all methods	11 ●	11

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Sample 62

Collective	c.551C>A	total
all methods	11 ●	11



MSH2 mutation type (N = 11, Rate of success: 100%)

Sample 61

Collective	Wild Type	total
all methods	11 ●	11

Sample 62

Collective	Wild Type	total
all methods	11 ●	11

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

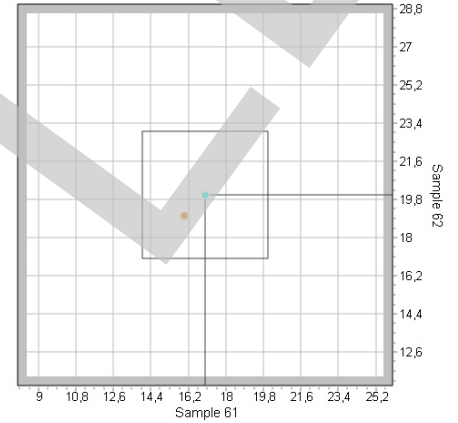
784

Set 15 - HTT Huntingtin

HTT Gen - CAG repeats allele 1 (N = 8)

Collective	Sample	Target value	Target range	Participants collective			Rate (%)	
				AVG	CV	Num.	Sam.	total
all methods	61	17.0	14.0 - 20.0	17.0	0.000	8	100	100
	62	20.0	17.0 - 23.0	20.0	0.000	8	100	100

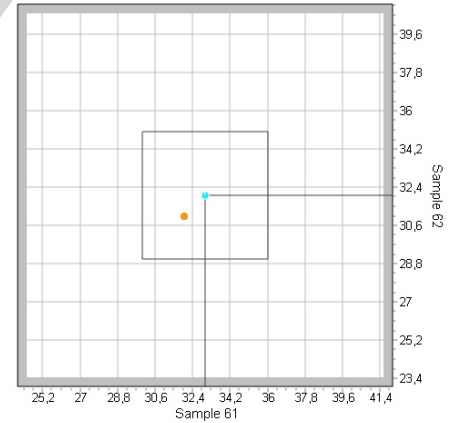
Rate of success: 100%



HTT Gen - CAG repeats allele 2 (N = 8)

Collective	Sample	Target value	Target range	Participants collective			Rate (%)	
				AVG	CV	Num.	Sam.	total
all methods	61	33.0	30.0 - 36.0	33.0	0.000	8	100	100
	62	32.0	29.0 - 35.0	32.0	0.000	8	100	100

Rate of success: 100%



784

Set 15 - HTT Huntingtin

HTT Gen - evaluation allele 1 (N = 8, Rate of success: 100%)

Sample 61

Collective	Normal	total
all methods	8 ●	8

Sample 62

Collective	Normal	total
all methods	8 ●	8

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective



HTT Gen - evaluation allele 2 (N = 8, Rate of success: 75%)

Sample 61

Collective	Normal	Intermediate	total
all methods	2	6 ●	8

Sample 62

Collective	Normal	Intermediate	total
all methods	2	6 ●	8

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

787

Set 17 - SMN1 (Spinal Muscular Atrophy)

SMN1 Gen - exon 7 deletion (N = 11, Rate of success: 100%)

Sample 61

Collective	No deletion	total
all methods	11 ●	11

Sample 62

Collective	Del HOM	total
all methods	11 ●	11

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

SMN1 Gen - exon 8 deletion (N = 10, Rate of success: 100%)

Sample 61

Collective	No deletion	total
all methods	10 ●	10

Sample 62

Collective	Del HOM	total
all methods	10 ●	10

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

788

Set 18 - AZF (Y-Chromosome Microdeletions)

Y-Chromosome AZFa deletion (N = 26, Rate of success: 100%)

Sample 61

Collective	No deletion	total
all methods	26 ●	26

Sample 62

Collective	No deletion	total
all methods	26 ●	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Y-Chromosome AZFb deletion (N = 26, Rate of success: 100%)

Sample 61

Collective	Deletion	total
all methods	26 ●	26

Sample 62

Collective	No deletion	total
all methods	26 ●	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective

Y-Chromosome AZFc deletion (N = 26, Rate of success: 100%)

Sample 61

Collective	Deletion	total
all methods	26 ●	26

Sample 62

Collective	No deletion	total
all methods	26 ●	26

The point corresponds to the correct result, the horizontal bar corresponds to your specification, the vertical bar corresponds to your collective