

Supplemental table 1: Marker characteristics.

Marker	Length [bp]	Repeat	FW-Seq	RV-Seq	genomic coordinates: [hg38]	Flanking feature:
D5S1466	218-239	GATA	[TAM]GTATCAGAACTTCATGTTGTACACC	GGCACCTAGGTTTGTCTGA	chr5:109683852-109684147	flanking MAN2A1 on 5'
D5S2065	109-135	CA	[6FAM]CAGCCTCATTGTTTATTGACAG	AATGGCATAGTTTTGGCTC	chr5:114385482-114385592	intronic KCNN2
D5S1484	264-282	ATT	[6FAM]CTACCTATGATGGGGCTGTG	GCTGGGCACTGAAGATTTAG	chr5:115057082-115057360	flanking TRIM36 on 5'
D5S2860	188-220	ATCT	[6FAM]TGTTTCTAGCCTGCTTGCTT	AGTGATGGTTCCTCCAATGA	chr5:115706509-115706729	flanking CDO1 on 5'
D5S404	180-198	GT	[HEX]CTGGAGATGTAATGCTGTGC	GATCACACATTCCACCTAAT	chr5:117511504-117511685	intronic LINC00992
D5S471	236-248	CA	[6FAM]TTTTACACATTTTCCCAGC	AAAACCTCATTACAAAAACAGGAG	chr5:119713364-119713582	flanking FAM170A on 3'
D5S1505	243-248	GATA	[HEX]TAAGTGCCAGAGTCTCCCAC	TAAGGCATGTCTCGGAGCTA	chr5:119765951-119766207	flanking FAM170A on 3'
D5S1712	88-106	ATA	[HEX]GCTTCCAAGTCAGTCTTTGG	GATTAGATACCAGTCCCTGCC	chr5:121367389-121367495	flanking FTMT on 5'
D5S1975	246-260	CA	[TAM]GCTGTGCTGTTTGGCTA	ACCTCATCTTTCGGGCTA	chr5:122789279-122789530	intronic SNX2
D5S818	156-157	GATA	[TAM]GGGTGATTTTCTCTTTGGT	TGATTCCAATCATAGCCACA	chr5:123775491-123775639	flanking CSNK1G3 on 3'
D5S1495	383-384	GATA	[TAM]TCCAAAAATGATTTGAGGC	TAAAAGATGTCTCTTTTTGCG	chr5:126014300-126014686	flanking GRAMD3 on 5'
D5S1500	353-386	GATA	[HEX]CTCTCTGTGCCCTCTCTAA	CAGTGAAAATTAATGAAATATGGC	chr5:144839079-144839440	flanking KCTD16 on 3'

Supplemental table 3: S.D. measurements among all or individual markers derived from duplicate analysis.

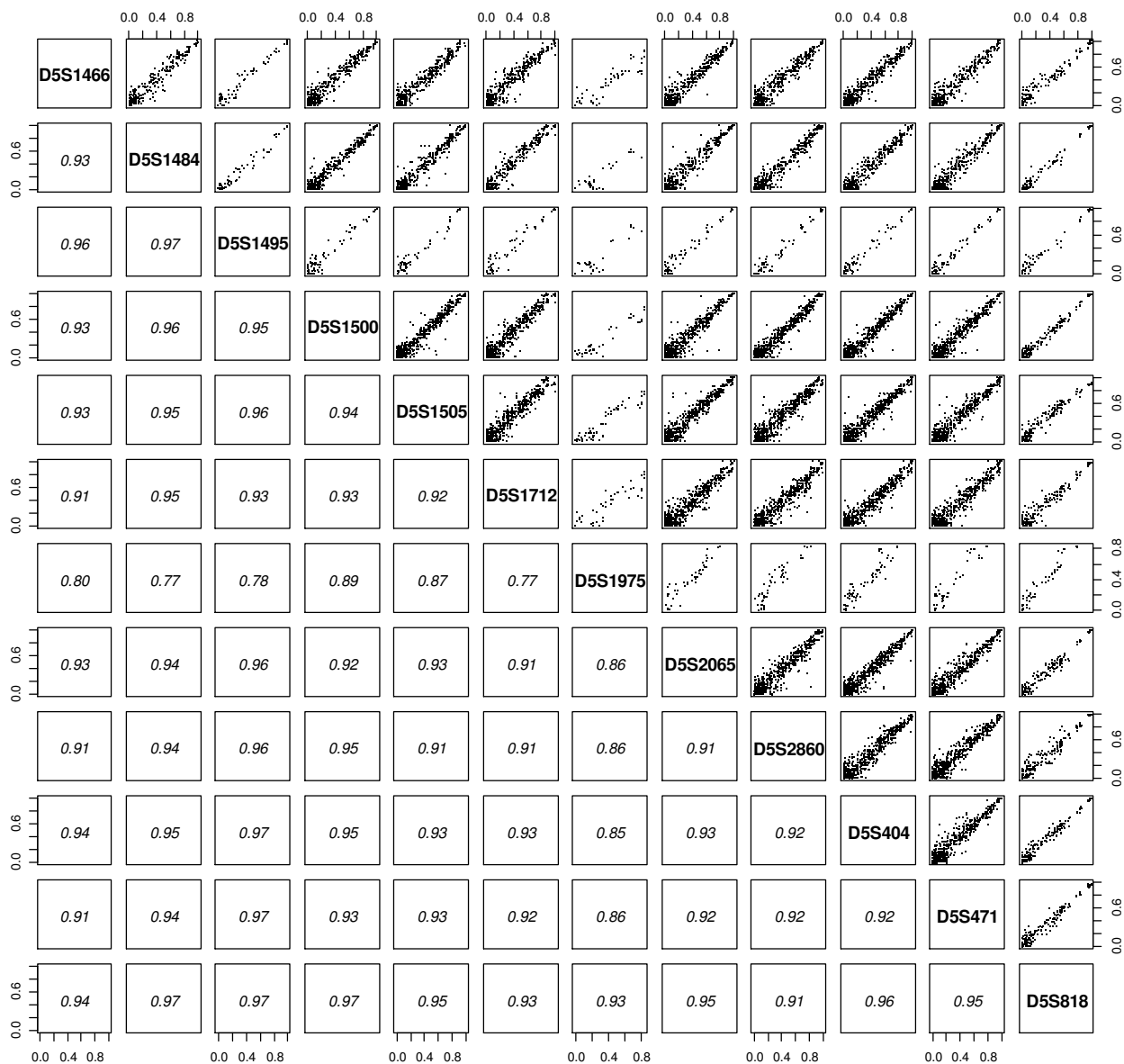
<b>name</b>	<b>mean S.D. [%]</b>	<b>95% CI</b>
<b>Overall*</b>	1.7	1.5-1.9
<b>D5S2065</b>	2.9	2.2-3.7
<b>D5S1484</b>	2.0	1.5-2.4
<b>D5S2860</b>	3.1	2.6-3.6
<b>D5S471</b>	2.7	2.3-3
<b>D5S1712</b>	3.6	3-4.2
<b>D5S1500</b>	2.7	2.2-3.2
<b>D5S404</b>	2.9	2.4-3.5
<b>D5S1505</b>	3.0	2.5-3.6
<b>D5S1466</b>	4.6	3.4-5.8
<b>D5S1975</b>	2.5	0.2-4.8
<b>D5S1495</b>	2.1	0.6-3.5
<b>D5S818</b>	2.3	1.5-3

\* Overall S.D. for del(5q) quantification was derived from duplicate multiplex reactions after averaging individual marker values

Supplemental table 4: Parameters of diagnostic accuracy for PCR based identification of del(5q).

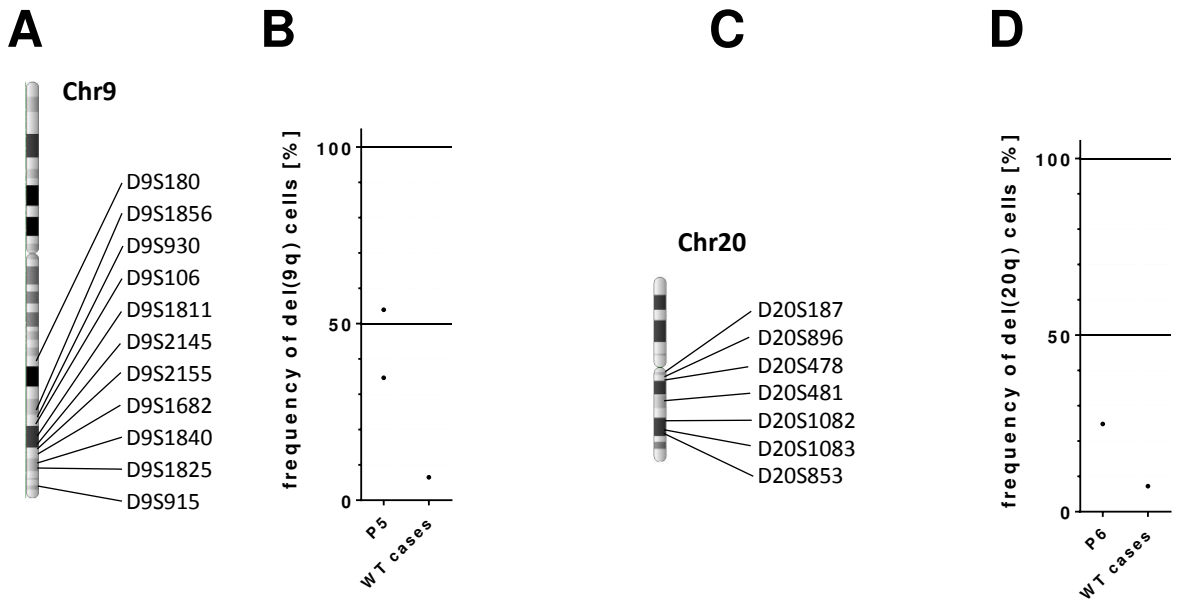
<b>threshold (95% CI)</b>	<b>4.9</b>	<b>7.2</b>	<b>7.5</b>	<b>7.6</b>	<b>9</b>	<b>11.4*</b>	<b>13.5</b>
<b>sensitivity</b>	0.99 (0.969-1)	0.979 (0.948-1)	0.969 (0.927-1)	0.958 (0.917-0.99)	0.948 (0.895-0.99)	0.938 (0.885-0.979)	0.885 (0.823-0.938)
<b>specificity</b>	0.25 (0.125-0.375)	0.688 (0.562-0.813)	0.729 (0.604-0.854)	0.771 (0.646-0.875)	0.875 (0.771-0.958)	0.979 (0.938-1)	1 (1-1)
<b>PPV</b>	0.725 (0.696-0.76)	0.862 (0.812-0.913)	0.877 (0.826-0.929)	0.893 (0.846-0.939)	0.938 (0.894-0.979)	0.989 (0.938-1)	1 (1-1)
<b>NPV</b>	0.923 (0.75-1)	0.943 (0.86-1)	0.921 (0.837-1)	0.902 (0.814-0.976)	0.894 (0.813-0.977)	0.887 (0.804-0.96)	0.814 (0.739-0.889)

PPV: positive predictive value; NPV: negative predictive value; \* optimal diagnostic cut-off.



## Supplemental Figure 1

Correlation of any possible two-marker comparisons. Numbers indicate  $R^2$ .



## Supplemental Figure 2

A) Chromosomal distribution of selected STR-markers specific for the chromosome 9q (del(9q)) B) Quantification of the fraction of cells carrying del(9q) in patient P5 and a wildtype (WT) case. C) Chromosomal distribution of selected STR-markers specific for the chromosome 20q (del(20q)). D) Quantification of the fraction of cells carrying del(20q) in patient P6 and a wildtype (WT) case.