

DB4403

DB4403/T 126 2020

Technical Regulation of DNA ID

..... 2

1 3

2 3

3 3

4 5

5 5

6 6

7 7

A DNA 10

B 12

..... 13

DB4403/T 126 2020

GB/T 1.1 2020

1

DB4403/T 126 2020

ABO

3.4

Rh

Rh gene

1 RHD RHCE 1 SMP1 RHD RHCE 3
30000 bp

3.5

Rh

Rh genotyping

Rh RHD D D DEL

3.6

HLA

HLA high resolution genotyping

HLA

3.7

ancestry trace

SNP

3.8

ancestry informative markers, AIMS

3.9

probe capture

90 120 nt

DNA

3.10

↑

principal component analysis PCA

[GB/T 29859 2013 2.6.5]

3.11

DNA

DNA genotyping

DNA

DNA

DNA

3.12

reads

[GB/T 35890 2018 3.1]

3.13

分 sequence alignment

[GB/T 29859 2013 2.2.1]

3.14

分 reference genome sequence

[GB/T 35890 2018 3.11]

4

AIMs:	Ancestry Informative Markers
AISNPs:	SNP Ancestry Informative SNPs
bp:	Base Pair
DNA:	Deoxyribonucleic Acid
Fst:	Fixation index between subpopulation and total population
InDel:	Insertion Deletion
IISNPs:	SNP Individual Identification SNPs
MAF:	Minor Allele Frequency
nt	Nucleotide
PIC:	Polymorphism Information Content
SNP:	Single Nucleotide Polymorphisms
STR:	Short Tandem Repeat
SOP	Standard Operating Procedure

- a) 100 Mb >20, 99.9%
- b) 0 99 99
- c) 16000 r/min
- d)
- e)
- f) -20 4

5.2.2 ↑

- a) DNA
- b) PCR PCR PCR DNA
- c) PCR dNTP PCR DNA PCR
- d)

6

6.1

1

1

	IISNP STR	ABO Rh HLA	AISNPs

6.2

6.2.1

STR SNP

- a)
- b)

c) HLA A B C DQB1 DRB1
B C DQB1 DRB1

HLA A

6.2.3

SNP

a)

Hardy Weinberg H W

P>0.001

b)

MAF>0.01 $\delta > 0.5$

c) Fst>0.3

)

A

7

7.1

5

1

1

7.2

2

2

7.5.2

<30

7.5.3 分

7.5.4

SNP

PCA

7.6

7.6.1 DNA

7.6.1.1

20 ng/ μ L DNA 1 μ g OD 260/280 1.7 1.9

DNA

DNA

DNA

7.6.1.2

DNA 10 ng

DNA

7.6.2

DNA

DNA

7.6.3

DNA >2.5

7.6.4 DNA

DNA

7.6.5

) Q30 % 0.1 % Q30 %>80 SplitRate %>90 30 SplitRate%

7.6.6

reads

2

90%

2

reads ^a X	^b X
5000 7000	300
2500	250
1000	50
500	40
200	10
100	5
50	5
^a ^b reads	

7.6.7

7.6.7.1

7.6.7.2

SOP

7.7

3

3

A

DNA

A. 1

A. 1. 1 STR

D21S11 D18S51 D5S818 D7S820 D13S317 D16S539 FGA D8S1179 D3S1358 TPOX Penta E
Penta D D2S1338 D19S433 D12S391 D6S1043 D1S1656 D2S441 D3S1744 D3S3045 D4S2366
D5S2500 D6S477 D7S1517 D7S3048 D8S1132 D10S1248 D10S1435 D10S2325 D11S2368 D13S325
D14S608 D15S659 D17S1290 D18S535 D19S253 D21S2055 D22 GATA198B05

A. 1. 2 X STR

DXS6789 DXS6795 DXS6803 DXS6809 DXS7132 DXS7133 DXS7423 DXS8377 DXS8378 DXS9895
DXS9898 DXS10101 DXS10134 DXS10135 DXS10074 GATA172D05 HPRTB

A. 1. 3 Y STR

DYS456 DYS389I DYS390 DYS389II DYS458 DYS19 DYS385 a/b DYS393 DYS391 DYS439
DYS635 DYS392 Y GATA H4 DYS437 DYS438 DYS448

A. 1. 4 SNP

A. 1. 4. 1 SNP

rs740910 rs1490413 rs1335873 rs1979255 rs1493232 rs2040411 rs1528460 rs717302
rs251934 rs8037429 rs891700 rs901398 rs873196 rs964681 rs737681 rs1463729 rs1360288
rs1382387 rs1413212 rs2056277 rs2107612 rs1015250 rs1005533 rs729172 rs10495407
rs1357617 rs719366 rs1031825 rs733164 rs938283 rs2111980 rs1886510 rs914165 rs354439
rs763869 rs2076848 rs1024116 rs1355366 rs735155 rs1454361 rs727811 rs917118
rs2831700 rs907100 rs1029047 rs2046361 rs722098 rs876724 rs2016276 rs826472
rs2830795 rs1028528

A. 1. 4. 2 SNP

rs1630312 rs3091244 rs2069945 rs6001030 rs140676 rs356167 rs941454 rs10045 rs3743842
rs2298556 rs3816662 rs2307223 rs10811897 rs17287498 rs385780 rs11141033 rs4540055
rs3812847 rs2032582 rs2278786

A. 1. 5 X SNP

rs2056688 rs2128519 rs1534285 rs763056 rs1373592 rs993010 rs119794 rs124792
rs925178 rs1207480 rs1936313 rs1977719 rs1372687 rs1557602 rs985125 rs983315
rs2190288 rs1991961 rs1931662 rs149910 rs1573704 rs1340718 rs1930674 rs1339597
rs1981452

A. 1. 6 Y SNP

rs11096433 M145 rs9306845 rs9786479 rs17276358 rs2075640 M134 M88 M95 rs10980425
rs17323322 M122 rs13447354 M89 rs9786707 M15 rs16980711 M9 rs17316592 rs17276345

A. 1. 7 SNP

709 1719 1736 3010 3394 3970 4216 4883 5147 5417 5460 6392 6455 8584 8701
9090 10397 10398 11914 12705 13708 13928 14318 14783 15487 16519
) SNP SNP

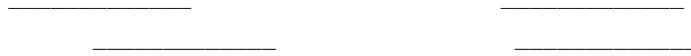
A. 2

SNP
rs2814778 rs3737576 rs7554936 rs10497191 rs1834619 rs1876482 rs200690 rs207770
rs6754311 rs798443 rs121498138 rs1919550 rs1220984 rs3811801 rs483103 rs7557739
rs7722456 rs870347 rs16891982 rs192655 rs2826159 rs317115 rs1402906 rs1871534
rs2196051

B

B. 1

B. 1



				ABO	Rh	HLA	1. 2. 3.	1. 2. 3. 4. DNA 5.	
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[1] Dean, M., et al. (1994). "Polymorphic admixture typing in human ethnic populations." *American Journal of Human Genetics* 55(4): 788–808.

[2] Edwards, A., et al. (1992). "Genetic variation at five trimeric and tetrameric tandem repeat loci in four human population groups." *Genomics* 12(2): 241–253.

[3] (2014). SNP , .

[4] Holsinger, K. E. and B. S. Weir (2009). "Genetics in geographically structured populations: defining, estimating and interpreting F_{ST} ." *Nature Reviews Genetics* 10(9): 639

