

BigY Plant Hg38 Novel Variants etc (Jan 2018)

Note Hg38 locations unless otherwise stated

Yfull and FTDNA ids..

P1a Plant YF04268 is 11830
P19a Plant YF04270 is 96105
P26a Plant YF04839 is 182593
PT3a Plant YF04841 is 235642
P40a Plant YF05854 is 436455
P43a Plant YF05858 is 453216
P5a .. is 11858
P28a .. is 230023

Branches A+D + P5a

Y22430 8908472 A-T Hg19=8776513 [Yfull: P1a+(0- 3+); P19a-(16- 0+); P26a-(17- 0+); PT3a-(3- 0+); P40a+(0- 3+); P43a+(0- 1+)] [YSEQ: P1a+; P1c+; P2a+; P19a-; P29a+; P40a+; PT1b-]

P1a (11830)

???20101718 only P1a A-G high Hg19=22263604 [P1a 8-31+ PT3a 2-,9+ P26a 14-5+ P19a 18-2+ P40a 7-,7+ P43a 6-,5+]

FGC22149 14382479 A-C Hg19=16494359 [YSEQ Sanger P1a+, P1c-, P33a-] [BigY: P1a+] [Yfull: P1a+(0-, 15+); P19a-(6-, 0+); P26a-(14-, 0+); PT3a-(26-, 0+)]

Yfull private, P19a no call

Yfull ambiguous or ?low, P19a no calls P26a- and PT3a-...

Hg19=8078641-G-T [P1a+ 0-, 2+; P19a nc; P26a- 4-, 0+; PT3a- 2-, 3+]

?Hg19=8072280-T-C [Yfull: P1a?+ 1-, 5+; P19a nc; P26a- 8- 0+; PT3a- 1- 0+]

Yfull ambiguous, P19a and P26a no calls PT3a-...

Hg19=13857166-A-G [P1a+ 0-, 2+; P19a no call; P26a no call; PT3a- 1- 0+]

Hg19=20126657-T-C [P1a+ 0-, 2+; P19a no call; P26a no call; PT3a- 1- 0+]

Not confirmed by Sanger

CTS11688 Hg19=23225633-A-C [YSEQ Sanger P1a-, P1c-] [** grey Big Tree] [BigY: all ?] [Yfull:

4*(1 read) P1a+ 0-, 1+; P19a no call; P26a- 1- 0+; PT3a- 1- 0+]

A10165 Hg19=21281530-C-T [YSEQ Sanger P1a-, P1c-, P33a] [P1a+ 0-, 2+; P19a nc; P26a- 2-

0+; PT3a- 1-, 0+]

PF374 Hg19=9943632-A-C [Yfull: P1a(0- 2+); P19a nc; P26a (1- 0+); PT3a (2- 0+)]

P5a (11858) awaiting YFull

14382564 only P5a G-A high Hg19=16494444 [P1a 69- P26a 105- PT3a 49- P19a 1+77- P40a 37- P43a 40-]

6942647 only P5a C-T high Hg19=6810688 [P1a 138- P26a 39- PT3a 50- P19a 45- P40a 58- P43a 57-]

7687461 only P5a C-T high Hg19=7555502 [P1a 134- P26a 52- PT3a 78- P19a 49- P40a 75- P43a 85-]

???56833081 only P5a T-C high Hg19=58979228 [P1a 2-8C1DEL P26a 3-7C1A PT3a 8-20C1G P19a 4-4+ P40a 5-13+ P43a 12-20C1G]

P19a (96105)

11055626 only P19a C-T high Hg19=13211302 [Yfull: P1a-(80-, 0+); P19a+(0-, 26+); P26a-(78-, 0+); PT3a-(77-,0+)]

11083546 only P19a T-A high Hg19=13239222 [Yfull: P1a-(44-, 0+); P19a+(0-, 44+); P26a-(51-, 0+); PT3a-(54-,0+)]

Y53440 15465552 only P19a A-C high Hg19=17577432 YFS529117 [Yfull: P1a-(110-, 0+); P19a+(4-, 70+); P26a-(114-,0+); PT3a-(114-, 0+)]

Y53441 15465553 only P19a C-A high Hg19=17577433 YFS529118 [Yfull: P1a-(110-, 0+); P19a+(4-, 70+); P26a-(113-,1+); PT3a-(114-, 0+)]

15568890 only P19a C-T high Hg19=17680770 YFS529119 [Yfull: P1a-(142-, 0+); P19a+(4-, 65+); P26a-(123-, 0+); PT3a-(106-, 0+)]

F3085 17454387 G-A Hg19=19566267 [Yfull: P1a-(18-, 0+); P19a+(0-, 16+); P26a-(56-, 0+); PT3a-(27-, 0+)]

?21426365 only P19a T-C high Hg19=23588251 [P19a 2-10+ P1a 37- P26a 21- PT3a 28- P40a 27- P43a 29-]

Yfull no calls for P1a and PT3a...

CTS771 Hg19=7004177-G-T [BigY: only P19a unknown confidence] [Yfull: P1a no call;

P19a+ 0-, 2+; P26a- 2- 0+; PT3a no call]

P28a (230023) awaiting YFull

11155683 only P28a G-A high Hg19=13311359 [P1a 63- P26a 57- PT3a 60- P19a 46- P40a 73- P43a 74-]

?56825935 only P28a C-A high Hg19=58972082 DC537 [P1a 17- P26a 18-2+ PT3a 10-2+ P19a 15- P40a 13- P43a 25-4+]

PF6330 C-T high 19857632 Hg19=22019518 [P1a 47- P26a 166- PT3a 57- P19a 102- P40a 63- P43a 63-]

Invalid apparent Branch B shared by P26a+PT3a (P1a- with P19a no call) but low read :

CONFIRMED FOR PT3a but awaiting Yseq result for P26a

?PF6726 10112991 Hg19=9950600 C-T [Yfull: P1a-(4- 0+); P19a no call; P26a+(0- 2+); PT3a+(0- 1+); P40a no call P43a(4-1+)] Yseq test withdrawn (see below)

NOT CONFIRMED BY YSEQ FOR P26a OR PT3A

??PF428 Hg19=10018083-T-G [Yfull: P1a-(4- 0+); P19a no call; P26a+(0- 2+); PT3a?+(2-4+)] Yseq available

Withdrawal of PF6726 by YSEQ..

PF6726 Y+ is not a typo but it refers to the IUPAC base code Y = C or T. This means that we have found a C and a T base stacked above each other in the sequencing trace. This is similar to heteroplasmy at some mtDNA sequences.

PF6726 is unfortunately not such an easy to interpret marker. I trust you that you have seen T+ results in some NGS BAM files, but they are telling you just half the truth. NGS assemblers map the reads according to the best fit of a reference sequence. However the hg19 reference sequence doesn't cover much of the centromeric region of the Y chromosome. Most of this region isn't sequenced at all due to the complexity of the highly repetitive elements. PF6726 is in the flanking region of the centromere and is really a part of this complex repetitive section. All centromeric reads are mapping to this flanking region (because they have nothing to map to in the not contained centromere section) and this creates very misleading side effects.

P26a (182593)

8216356 only P26a C-A high Hg19=8084397 YFS1211490 [Yfull: P1a-(18-, 1+); P19a-(20-, 0+); P26a+(0-,20+); PT3a-(25-, 0+)] [+ Big Tree] Yseq available

???11543441 only P26a G-T high Hg19=13699117 [P1a 170T3C1360G, P26a 127T558G, PT3a 75T2C869G, P19a 75T366G, P40a 94T779G, P43a 87T2C893G]

???56830584 only P26a G-C high Hg19=58976731 DC613 [P1a 8-3+, P26a 1-6+, PT3a 18-21+, P19a 15-18+, P40a 2-9+, P43a 20-21+]

?CTS10386 17317634 Hg19=19429514-C-T [Yfull: P1a-(3-, 0+); P19a-(1-, 0+); P26a+(0-,1+); PT3a-(1-, 0+)] [** Big Tree] Only 1+ in Yfull

?YFS1211919 19422085 G-A Hg19=21583971 [Yfull: P1a nc; P19a 3-; P26a 2+; PT3a 2-; P40a 6-; P43a 2-] Yfull ambiguous with P1a no call; Yseq available

?YFS1211917 19100821 C-A Hg19=21262707 [Yfull: P1a-(3- 0+); P26a+(0- 2+); PT3a-(5- 0+); P19a nc; P40a nc; P43a 3-] Yfull with P19a and P40a no call; Yseq available

?YFS1211949 20949850 G-T Hg19=23111736 [P1a 3-, P26a 2+, PT3a 3-, P19a nc, P40a 1-, P43a 2-] Yfull novel, P19a nc, Yseq available

OLD HG19 findings..

Yfull novel, ambiguous for P1a

9462202 YFS1211499 [Yfull: P1a-(9- 3+); P19a-(3- 0+); P26a+(0-,2+); PT3a-(3- 0+)]

Yfull ambiguous with P1a and P19a no calls and P26a+(0- 2+) and PT3a-...

3151711 YFS1211462 [Yfull: PT3a- 3- 0+]

4501775 YFS1211472 [Yfull: PT3a- 1- 0+]

8079435 YFS1211489 [Yfull: PT3a- 1- 0+]

15536369 YFS1211868 [Yfull: PT3a- 2- 0+]

21269382 YFS1211918 [Yfull: PT3a- 1- 0+]

Yfull ambiguous with P1a and PT3a no calls and P26a+(0- 2+) and P19a-..

6624538 YFS1211486 [Yfull: P19a- 1- 0+]

Yfull ambiguous with P19a and PT3a no calls and P26a+(0- 2+) and P1a-...

4166089 YFS1211469 [Yfull: P1a-(2- 0+)]

17492163 YFS1211876 [Yfull: P1a-(1- 0+)]

Indicated by BigY or Big Tree but extremely doubtful...

PF682 S3569 14624294-C-T [Yfull: P1a?(3-, 5+); P19a-(1-, 0+); P26a?(3-, 4+); PT3a?+(2-, 4+)]

CTS7391 17455962-C-T [Yfull: P1a-(4-, 0+); P19a-(1-, 0+); P26a?(1-, 1+); PT3a-(7-, 0+)]

PT3a (235642)

?8937075 only PT3a T-A high Hg19=8805116 YFS1212707 [Yfull: P1a-(46-, 0+); P19a-(26-, 0+); P26a-(32-,0+); PT3a+(2-,36+)] [+ Big Tree] Yseq available
15912529 only PT3a C-T high Hg19=18024409 YFS1213002 [Yfull: P1a-(37-, 0+); P19a-(8-, 0+); P26a-(14-,0+); PT3a+(0-,21+)] [** Big Tree] Yseq available
13125955 Hg19=15237869 G-T YFS1212991 [Yfull: P1a-(91-, 0+); P19a-(84-, 0+); P26a-(155-,0+); PT3a+(0-,99+)] [+ Big Tree] Yseq available

OLD HG19 findings...

Indicated by BigY or Big Tree but weak...

CTS2782 14480060-A-C [BigY: PT3a+ medium confidence] [Yfull: P1a?-(3-,1+); P19-(3-,0+); P26a?-(4-, 2+); PT3a?+(2-, 4+)]d

16410949-G-T [Big Tree: **] [Yfull: P1a no call; P19a-(1-, 0+); P26a no call; PT3a+(0-,1+)]

Branch D P40a+P43a (and also P2a by YSEQ)

A10650 20052196 only P40a and P43a A-T high (branch D) Hg19=22214082 [Yfull: P1a-(140-, 0+); P19a-(117-,1+); P26a-(165-,0+); PT3a-(74-,0+); P40a+(0-,63+); P43a-(0-,68+)] Yseq (\$17.50)

?BY26225 2997177 C-G [Big Y: +ve P40a P43a; -ve P1a; ? P5a P19a P26a P28a PT3a]

P40a (436455) and P2a (for A10964 and A10965 by YSEQ)

A10964 7279376 only P40a A-G high Hg19=7147417 YFS176235 G [Yfull: P1a-(91-, 0+); P19a-(119-,1+); P26a-(206-,0+); PT3a-(58-,0+); P40a+(2-,75+); P43a(42-,0+)] [YSEQ; P2a+]

A10965 19983594 only P40a A-G high Hg19=22145480 YFS1768457 [Yfull: P1a-(72-, 0+); P19a-(53-,1+); P26a-(118-,0+); PT3a-(55-,0+); P40a+(1-,50+), P43a(74-,0+)] [YSEQ; P2a+; P40a+]

OLD HG19 findings...

Acceptables only mainly no call

3751927-C-A YFS1768223 M [Yfull:P1a nc; P19a nc; P26a(0+,1-); PT3a nc; P40a(3+,0-); P43a nc]

Shared up to L617 in Acceptables only (Branches B and D but others are no call)

24435456-G-C YFS1768457 MC [Yfull:P1a nc; P19a nc; P26a (1+,0-); PT3a nc; P40a(3+,0-); P43a nc]

P43a (453216)

A10960 12835138 only P43a C-A high Hg19=14947064 YFS1770183 G [Yfull: P1a-(16-, 0+); P19a-(41-,1+); P26a-(16-,0+); PT3a-(32-,0+); P40a(15-,0+); P43a(0-,20+)] BigY High and Big Tree + and Yfull Best

A10961 14532117 only P43a A-T high Hg19=16643997 YFS1770190 G [Yfull: P1a-(185-, 0+); P19a-(62-,0+); P26a-(109-,0+); PT3a-(149-,0+); P40a(143-,0+); P43a+(2-,110+)] BigY High and Big Tree + and Yfull Acceptable

A10962 16908600 only P43a A-C high Hg19=19020480 YFS1770197 [Yfull: P1a-(39-, 0+); P19a-(32-,0+); P26a-(66-,0+); PT3a-(42-,0+); P40a(46-,0+); P43a(0-,58+)] BigY High and Big Tree + and Yfull Best

A10963 19498923 only P43a C-A high Hg19=21660809 YFS1770220 G [Yfull: P1a-(124-, 0+); P19a-(74-,0+); P26a-(103-,0+); PT3a-(103-,0+); P40a(95-,0+); P43a+(1-,68+)] BigY High and Big Tree + and Yfull Acceptable

?Y68911 C-A 13679140 Hg19=15791020-C-A YFS1770187 [Yfull: P1a (1-,0+); P19a(1-,0+); P26a(4-,0+); PT3a(5-,0+) P40a(3-,0+); P43a(0-,3+)] Yfull Acceptable only (ok but low calls)

OLD HG19 findings...

Not acceptable for testing at YSEQ

Y2576 FGC21589 4567324-A-C [Yfull: P1a-(13-, 0+); P19a-(41-,0+); P26a-(62-,0+); PT3a-(18-,0+)]

Not acceptable Y-SNPs according to YSEQ:

• Apparent Branch B shared by P26a+PT3a (P1a- with P19a no call):

CTS12427/PF1324 28586775-C-T [Yfull: P1a-(1- 0+); P19a no call; P26a+(0- 3+); PT3a+(0- 1+)]

CTS12428 28586776-A-G [Yfull: P1a-(1- 0+); P19a no call; P26a+(0- 3+); PT3a+(0- 1+)]

CTS12429 28586782-G-A [Yfull: P1a-(1- 0+); P19a no call; P26a+(0- 3+); PT3a+(0- 1+)]

• Possible Branch A+B shared by P1a+P26a+PT3a (with P19a-):

?PF521 13403332-C-T [Yfull: P1a+(0- 4+); P19a-(2- 0+); P26a?+(2- 9+); PT3a?+(1- 4+)]

Position 13403332 isn't recommended because its sequence is 96.9% similar to chromosome 1 (142585753..142586753)

• Possible Branch E+?B+D shared by P19a+?P26a+?PT3a+P40a (with P1a-):

13323548-A-G [BigY: P40a+ High] [Yfull: P1a-(12- 0+); P19a?+(6- 13+); P26a?+(11- 20+);

PT3a?+(11- 24+)]

Position 13323548 isn't recommended. It is >96.8% similar to other segments in the genome and it is flanked by a homopolymer (poly A region). The position has been named Z33816 in haplogroup J2a.

The sequence isn't recommended for phylogeny of a specific haplogroup or family.

Y-STRs:

Summary of Yfull Y-STR results (available as csv downloads) :

P1a has 422 reliable, including 96 (omitting 15 n/a) of the standard Y-DNA111 Y-STRs

P19a has 419 reliable, including 91 (omitting 14 n/a and 5 unreliable) of standard 111

P26a has 454 reliable, including 98 (omitting 13 n/a and 0 unreliable) of standard 111

PT3a has 445 reliable, including 101 (omitting 8 n/a and 2 unreliable) of standard 111

P40a has 419 reliable, including 91 (omitting 16 n/a and 4 unreliable) of standard 111

P43a has 422 reliable, including 91 (omitting 17 n/a and 3 unreliable) of standard 111

Nearest R-L617 match to a non-Plant: P1a-Rogers has 93 different of 397 compared (0.234)

P1a-P19a has 31 different of the 403 which can be compared (0.077) (i.e. many more to differentiate between these two Plant branches than with a standard Y-DNA111 test)

8 of the extra were already known (2 at Y-DNA12; 1 at 13 to 25 level; 3 at 16 to 37; 2 at 68 to 111)

4 more already known are not reliable in BigY (2 at Y-DNA25; 1 at 26 to 37, 1 at 68 to 111)

This makes a total of 35 known differences, 5 of which differ by 2

1 already known: DYS570 (33, 35) (at 68 to 111 level)

4 new: DYS526A (12, 14); DYS547 (45, 47); DYS84 (17, 15); DYS612 (31, 29)

P1a-P26a has 40 different of the 417 that can be compared (0.096)

P1a-PT3a has 39 different of the 413 that can be compared (0.094)

P1a-P40a has 29 different of the 406 that can be compared (0.071)

P1a-P43a has 28 different of the 402 that can be compared (0.072)

P19a-P1a has 31 different of the 403 that can be compared (0.077)

P19a-P26a has 29 different of the 421 that can be compared (0.069)

P19a-PT3a has 30 different of the 408 that can be compared (0.074)

P19a-P40a has 31 different of the 394 that can be compared (0.079)

P19a-P43a has 32 different of the 395 that can be compared (0.081)

P26a-PT3a has 19 different of the 432 that can be compared (0.044)

P40a-P43a has 25 different of the 402 that can be compared (0.062)

P40a-P26a has 35 different of the 414 that can be compared (0.085)

P40a-PT3a has 34 different of the 407 that can be compared (0.074)

P43a-P26a has 35 different of the 416 that can be compared (0.084)

P43a-PT3a has 34 different of the 409 that can be compared (0.083)