Wider view on mutation spectrum

Autosomal Dominant Retinitis Pigmentosa Analysis Kit

– all you need in one box –

Quick facts

- 385 disease related genetic variations from 16 genes detected simultaneously in a single run
- Developed with the leading scientists in the Autosomal Dominant Retinitis Pigmentosa (AD-RP) research community backed by the latest scientific and medical studies
- Suitable for isolated familial cases of retinitis pigmentosa in succeeding generations

Easy to use

- Everything you need is in one box
- Analysis software comes with integrated guidance for mutation detection
- Barcoded arrays to ensure precise tracking
- Consultations from day-to-day users

Autosomal Dominant Retinitis Pigmentosa

- Genorama reliable AD-RP Analysis Kit is based on flexible and cost-effective Arrayed Primer EXtension (APEX) genotyping technology
- The kit enables to determine 385 genetic variations in 16 genes - CA4, FSCN2, IMPDH1, NRL, PRPF3, PRPF31, PRPF8, RDS, RHO, ROM1, RP1, RP9, CRX, TOPORS and PNR
- Genorama AD-RP test has been developed as up to date as current discoveries enable and is always open for scientifically proved updates

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Reliable and up to date

- List of relevant publications is available on Genorama website
- Intensively used throughout years and hundreds of screened samples
- Based on established and validated APEX technology
- Genorama is keeping a close eye on new discoveries in the field

Genorama AD-RP Analysis KIT includes all consumables for successful analysis process and provides all necessary reagents, PCR primers and barcoded arrays for 25 tests

- PCR primers and purification Kit
- APEX Template Preparation Kit
- APEX Reaction Mixture Kit
- AD-RP microarrays

APEX technology

Arrayed Primer EXtension (APEX) genotyping is based upon an array of oligonucleotides, immobilized on glass surface via their 5' end. Patient's DNA is amplified by PCR, digested enzymatically and annealed to the immobilized primers, which promote sites for template-dependent DNA polymerase extension reactions using four unique fluorescently labelled dideoxy nucleotides. The mutation is detected by the change in primer sites colour code using the previously installed Genorama genotyping platform.

Ordering information

Cat #	Product name
G60106	Autosomal Dominant Retinitis Pigmentosa Analysis KIT for 25 reactions

For further information please contact Genorama representatives

Genetic variations in test

CA4 40 C>T; 206 G>A; 655C>A
FSCN2 72 delG
IMPDH1 313 C>T; 594 T>A/G; 671 G>C; 676 G>A; 680 T>C; 692 G>C; 713 A>G; 802 G>A
NRL 148 T>A ; 149 C>T; 151 C>A/T; 152 C>T; 224_225 insC; 288 G>A; 365 G>A ; 479 T>C
PRPF3 1477 C>T ; 1466 C>A; 1481 C>T
PRPF31 IVS1+G>T; 79 G>T; IVS2+1 G>A; 220 C>T; 331-342 del12bp; 357_359 del 2bp (AA); 387_390 delC; 413 C>A; IVS5-1 G>A ; 421 G>T; 522_527+10 del16bp; IVS6+1 G>T ; IVS6-1 G>A; IVS6+3 A>G ; IVS6-3 -45del 43bp; 541G>T; 548-580 dup33bp; 581C>A; 636 delG; 646 G>C; 670 G>A; 732-737 del6bp/ins20bp; 757 G>C; 758_767 del; 768-771 insA; 785delT; 828-829 delCA; IVS8+1 G>C ; 871 G>C; 895 T>C; 973 G>T; 1049_IVS10+20del/insCCCCT; IVS10+1 G>A; 1115-1125 del11bp; 1141/1142 delG; IVS11+2 T>C; 1155_1159 del/insAGGGATT; 1374+654C>G
PRPF8 6353 C>T; IVS41-4 G>A; 6893-6896 del4bp/ins8bp; 6901 C>A/T; 6912 C>G; 6926 A>C/G; 6928 A>G; 6929 G>A; 6942 C>A; 6942-6943 delC; 6972-6977 del6bp/ins11bp; 6974-6994 del21bp; 6983 C>T; 6991 delG; 7000 T>A; 7006 T>C
RDS 2 T>C ; 37 C>T; 63 G>A; 73-74 delTG; 80 C>T; 94 A>G; 99-100 insC; 112-113 delG; 133 C>T; 136 C>T; 163 delT; 166_177 del; 198-202 del5bp/ins8bp; 199-201 del3bp; 202 G>C; 232G>C; 232_233insT; 260-267 del8bp; del265_268/insAGGGCC; 282 G>A; 352-354 del3bp; 367 C>T; 368-371 delG; 374 C>T; 377 T>G; 393_394 delC; 410 G>A; 420 421 ins4bp; 421 T>C; 422 A>G; 424 C>T; 433_434 delGA; 441 delT; 457-459 del3bp; 458 A>G; 469 G>A; 483 C>G; 484 G>C; 494 G>A; 499 G>A; 500 G>A; 505-507 del3bp; 514 C>G/T; 514_515 insT; 515 G>A; 518 A>T; 533 A>G; 535 T>C; 551 A>C; 554 T>C; 557 A>T; 577-579 delAA; IVS1+1 G>C; IVS1 +5_+6 insA; 584 G>T; 589 A>G; 594 C>G; 599 T>A; 607_620 del14bp(CGGTACCTGGTGGA); 609-625 del17bp; 616-627 del12bp; 621-624 insG; 622 G>C; 623 G>A; 625 G>C; 628 C>T; 629 C>G; 633 C>A; 634 A>G; 635 G>C; 637 T>C; 638 G>A/T; 639 C>T; 641 G>C/A; 646 C>T; 647 C>T; 655-657 del3bp; 656 C>G; 658 delC; 658 C>T; 659 G>A; 662C>T; 676 C>G; 700-701 insT; 711_722 del; 715 C>T; 730 A>C; 732 C>A/G; 736 T>C; 738_741 dup; 745G>A; 749 G>T; 774 C>A; 797 G>A; 802 G>A; 811_813del; 824-825 delTC; IVS2+2 T>C; IVS2+3 A>T; 855 C>A; 866 C>T; 897-898 delTG; 904 G>T; 914 G>A; 914 922 del9bp; 920 delT; 946T>G; 947 G>A; 975_978 dupGCTG; 991 C>T; 946 T>G; 1015 G>A
RHO 11 C>A; 44 A>G; 50 C>T; 67 C>G; 68 C>A/T; 84 G>T; 116 T>G; 119 T>G; 131 T>C; 133 T>C; 137 T>G; 151 G>C; 152 G>C/T; 155 T>A; 158 C>G; 165 C>A; 170 T>G; 173 C>G; 190 C>T; 202-213 del12bp; 217_219 del; 236 T>C; 260 T>A; 266 G>A; 269 G>A; 281 C>T; 310 G>A; 316 G>A/T; 325 G>A; 329 G>T/A; 341 G>A/T; 362 G>T; 374 T>G; 380 C>T; 392 T>C; 403 C>G/T; 404 G>T/C; 408 C>A; 409 G>A; 419 G>C; 448 G>A; 491 C>A/T; 501 C>G; 509 C>G; 511 C>T; 512 C>A/T; 520 G>A; IVS2-2 A>G; 532 T>A; 533 A>G; 535 A>T; 538 C>G; 541 G>A; 544 G>A; 551 A>C; 553 T>C; 556 T>C; 557 C>G; 560 G>A; 562 G>A; 563 G>A; 568 G>A/T; 569 A>G; 578 C>T; 620 T>G; 625 G>A; 632 A>G/C; 644 C>T; 647 T>G/A; 659 T>G; 664 T>C; 743 A>G; 745 G>T; 755 G>C; 759 G>A; 763-765 del3bp; 790-792 del3bp; 800 C>G/T; 810 C>A; 811 G>A; 850 G>A; 865 A>C; 875 C>A; 884C>T; 886 A>G; 887 A>T; 888 G>T; 891 C>A; 934 C>T; 936G>A; IVS4+1 G>T; IVS4-1 G>A/T; 938-946 del9bp; 953-955 del3bp; 979_982delCCAC; 983 T>C; 995-1011 del17bp; 998 C>T; 998-999 insAGGC; 1003 delG; 1019 delC; 1021-1028 del8bp; 1021 G>T; 1025 C>T; 1030 C>T; 1033 G>C/A; 1034 T>G; 1036 G>C; 1039 C>G/T/A; 1040 C>G/A/T; 1045 T>G
ROM1 178 C>A; 224 G>A; 236-240 insG; 323 C>T; 338 G>A; 725 G>A; 758 G>A
RP1 1118 C>T; 1458-1461 dup; 1498-1499 insGT; 1973-1980 insA; 1989 G>T; 2005 G>A; 2029 C>T/delC; 2035 C>T; 2056 C>T; 2098 G>T; 2112_2115 delA; 2165_2166 delAA/insG; 2167 G>T; 2168-2181 del14bp; 2169-2173 insG; 2171-2186 del16bp; 2184-2185 delG; 2206-2207 insT; 2232 T>A; 2236-2239 delA; 2255 C>T; 2285-2289 delTAAAT; 2287-2290 delAAATA; 2303-2304 delC; 2332 A>T; 2336-2337 delCT; 2590_2599 del; 2607-2614 insA; 2732 C>A; 2847delT; 2951 A>G; 2953 A>T; 3155-3157 delT; 4550-4555 delA; 5249-5252 delA; 5423 T>C
RP9 410 A>T
CRX 20-25 insG; 121 C>T; 122 G>A; 166 G>A; 196 G>A; 238 G>A; 239 A>C; 268 C>T; 365 G>A; 425 A>G; 436-447 del12bp; 443-447 insC; 455-458 delC; 472 G>A; 502 delG; 502-503 delGA; 510-512 delT; 519-520 delG; 527-529 delG; 540-541 delG; 571 delT; 586-591 insC; 587-590 delCCCC; 611-615 delC; 649-650 delG; 705-709 delC; 724 G>A; 720-742 dupl23bp; 752-753 delC; 786-789 delC; 816_818 del_insAA; 818 C>T
TOPORS 1205A>C; 2422 G>T; 2474_2475insA; 2552_2553delGA; 2569del
PNR 166 G>A
KLHL7 449G>A; 457G>A; 458C>T

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