|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **성명 / 성별** | $name$ / $gender$ | **검체 종류** | $clinicalSpecimen$ | **검사번호** | $checkNumber$ |
| **등록번호** | $registration$ | **검체 채취일** | $collectDate$ | **검사일** | $dateOfInspection$ |
| **생년월일** | $birth$ | **검체 접수일** | $dateOfReceipt$ | **예비 보고일** | $tempReportDate$ |
| **의뢰의사** | $doctor$ | **검체상태** | 적합/부적합 (이유: $reason$) | **최종 보고일** | $reportDate$ |

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| **검출 유전 변이 (Detected Genetic Variants)** |

NGS Panel for $panel$

|  |
| --- |
| **검출 유전 변이 (Detected Genetic Variants)** |

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Gene | Ref. Seq | Nucleotide | Amino acid | VAF\* | Depth† | SIGNIFICANCE‡ | Remark\*\* |
|  |  |  |  |  |  |  |  |

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Gene | Ref. Seq | Nucleotide | Amino acid | VAF\* | Depth† | SIGNIFICANCE‡ | Remark\*\* |
|  |  |  |  |  |  |  |  |

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Gene | Ref. Seq | Nucleotide | Amino acid | VAF\* | Depth† | SIGNIFICANCE‡ | Remark\*\* |
|  |  |  |  |  |  |  |  |

상기 결과는 임상적 중요성에서 Tier I, II, III에 해당하는 변이만 기술하였습니다. (J Mol Diagn. 2017;19;4-23)   
Tier I: 중요한 임상적 의의가 있는 변이, Tier II: 잠재적 임상적 의의가 있는 변이, Tier III: 임상적 의의가 알려지지 않은 변이  
\* VAF, Variant Allele Frequency; †DEPTH, Total Read Depth; ‡SIGNIFICANCE, clinical significance; §Low coverage, read depth < 100x  
\*\*\* WARNING : 100X 이하 지역

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| **소견 및 결과 해석** |

|  |
| --- |
| **매우 낮은 신뢰도로 생성된 설명** |

**화순전남대학교병원 진단검사의학과**

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **검사자** | $name1$ | / | $name2$ |  | **보고자** | $name3$ | / | $name4$ | (서명) |

|  |
| --- |
| **표적 치료제 선택 가이드 (Targeted Therapeutic Implications)** |

1. 임상적으로 중요한 변이 (Variants with Clinical Significance)

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Genomic  Alternations** | **Tier I\* (중요한 임상적 의의)** | | **Tier II\*\* (잠재적 임상적 의의)** | |
| **Level A** | **Level B** | **Level C** | **Level D** |
| **Total Count** | **$evidenceACount$** | **$evidenceBCount$** | **$evidenceCCount$** | **$evidenceDCount$** |
|  |  |  |  |  |

**\* Tier I : 중요한 임상적 의의**  
- Level A : 전문 가이드라인에 포함된 FDA-승인 약제  
- Level B : 전문가들의 합의가 이루어진, 근거가 충분한 연구를 통해 인정된 약제  
**\*\* Tier II : 잠재적 임상적 의의**  
- Level C : 다른 종류의 종양에서 FDA-승인을 얻은 약제 / 다양한 소규모 연구를 통해 약간의 합의가 이루어진 약제  
- Level D : 전임상시험 단계의 약제, 또는 전문가 합의는 없으나 몇몇 케이스 보고가 있는 약제

참고문헌 : J Mol Diagn. 2017;19;4-23

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| **검사 세부정보 (Test Details)** |

1. 검사 대상 유전자 (Genes in Panel) 및 결과 요약

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Gene** | **Tier I** | **Tier II** | **Tier III** | **Total** |  | **Gene** | **Tier I** | **Tier II** | **Tier III** | **Total** | |
|  |  |  |  |  |  |  |  |  |  |  |

\*보건복지부 필수유전자

2. 기준 염기서열 (Accession number of Reference Sequence)

|  |  |
| --- | --- |
| **Gene** | **DB Accession number** |
| ABL1 | NM\_007313 NM\_005157 |
| ABL2 | NM\_007314 NM\_001168238 NM\_001168237 NM\_001168236 NM\_001168239 NM\_005158, NM\_001136000 |
| AFF1 | NM\_001166693 NM\_001313959 NM\_001313960 NM\_005935 |
| ANKRD26 | NM\_014915 NM\_001256053 |
| ARHGAP26 | NM\_015071 NM\_001135608 NM\_001349547 |
| ARID5B | NM\_032199 NM\_001244638 |
| ASXL1 | NM\_001164603 NM\_015338 |
| ATG2B | NM\_018036 |
| BCOR | NM\_001123384 NM\_017745 NM\_001123385 NM\_001123383　DB |
| BCR | NM\_021574 NM\_004327 |
| BMI1 | NM\_005180 |
| BRAF | NM\_001354609 NM\_004333 |
| CALR | NM\_004343 |
| CBFB | NM\_022845 NM\_001755 |
| CBL | NM\_005188 |
| CDKN2A | NM\_058195 NM\_058197 NM\_001195132 NM\_000077 |
| CDKN2B | NM\_004936 NM\_078487 |
| CEBPA | NM\_001287435 NM\_001287424 NM\_001285829 NM\_004364 |
| CEP72 | NM\_018140 |
| CREBBP | NM\_004380 NM\_001079846 |
| CRLF2 | NM\_022148 NM\_022148\_2 NM\_001012288 NM\_001012288\_2 |
| CSF1R | NM\_005211 NM\_001349736 NM\_001288705 |
| CSF3R | NM\_000760 NM\_156039 NM\_172313 |
| CYP2D6 | NM\_001025161 NM\_000106 |
| DDX41 | NM\_001321732 NM\_001321830 NM\_016222 |
| DEK | NM\_003472 NM\_001134709 |
| DHX15 | NM\_001358 |
| DKC1 | NM\_001363 NM\_001142463 NM\_001288747 |
| DNM2 | NM\_001190716 NM\_004945 NM\_001005362 NM\_001005361 NM\_001005360 |
| DNMT3A | NM\_175630 NM\_175629 NM\_022552 NM\_153759 NM\_001320893 NM\_001320892 |
| EBF1 | NM\_001324101 NM\_001324107 NM\_001324108 NM\_001324109 NM\_001324111 NM\_001290360 NM\_024007 NM\_001324103 NM\_001324106 NM\_182708 |
| ECT2L | NM\_001077706 NM\_001195037 |
| EGFR | NM\_001346898 NM\_001346897 NM\_005228 NM\_001346899 NM\_001346941 NM\_001346900 NM\_201284 NM\_201283 NM\_201282 |
| EP300 | NM\_001429 |
| EPOR | NM\_000121 |
| **Gene** | **DB Accession number** |
| ETNK1 | NM\_001039481 NM\_018638 |
| ETV6 | NM\_001987 |
| EZH2 | NM\_152998 NM\_004456 NM\_001203249 NM\_001203248 NM\_001203247 |
| FBXW7 | NM\_033632 NM\_001257069 NM\_018315 NM\_001349798 NM\_001013415 |
| FGFR1 | NM\_023105 NM\_001174064 NM\_001174065 NM\_001174066 NM\_001174067 NM\_001354370 NM\_015850 NM\_001354367 NM\_023106 NM\_023110 NM\_001354368 NM\_001354369 NM\_001174063 |
| FIP1L1 | NM\_001134938 NM\_001134937 NM\_030917 |
| FLT3 | NM\_004119 |
| GATA1 | NM\_002049 |
| GATA2 | NM\_001145662 NM\_032638 NM\_001145661 |
| GATA3 | NM\_002051 NM\_001002295 |
| GSKIP | NM\_001271905 NM\_001271904 NM\_016472 NM\_001271906 |
| IDH1 | NM\_001282387 NM\_001282386 NM\_005896 |
| IDH2 | NM\_001290114 NM\_002168 NM\_001289910 |
| IKZF1 | NM\_001291837 NM\_006060 NM\_001220768 NM\_001220767 NM\_001291847 NM\_001291846 |
| NM\_001291845 NM\_001291844 NM\_001291843 NM\_001291842 NM\_001220765 NM\_001220771 |
| NM\_001220770 NM\_001291841 NM\_001291840 NM\_001291839 NM\_001291838 |
| IKZF2 | NM\_016260 NM\_001079526 |
| IKZF3 | NM\_001257413 NM\_001257414 NM\_001284515 NM\_001284514 NM\_001257409 NM\_001257408 |
| NM\_001257410 NM\_001257411 NM\_001257412 NM\_012481 NM\_001284516 NM\_183232 |
| NM\_183231 NM\_183230 NM\_183229 NM\_183228 |
| IL3 | NM\_000588 |
| IL7R | NM\_002185 |
| JAK1 | NM\_001321857 NM\_001321856 NM\_001321852 NM\_001321854 NM\_001321855 NM\_002227, NM\_001320923 NM\_001321853 |
| JAK2 | NM\_001322195 NM\_001322194 NM\_001322196 NM\_001322198 NM\_001322199 NM\_001322204 NM\_004972 |
| JAK3 | NM\_000215 |
| KDM6A | NM\_001291416 NM\_021140 NM\_001291417 NM\_001291418 NM\_001291421 NM\_001291415 |
| KIT | NM\_001093772 NM\_000222 |
| KMT2A | NM\_005933 NM\_001197104 |
| KRAS | NM\_033360 NM\_004985 |
| LMO2 | NM\_001142315 NM\_005574 NM\_001142316 |
| MECOM | NM\_004991 NM\_001105077 NM\_001105078 NM\_005241 NM\_001163999 NM\_001164000 NM\_001205194 |
| MGA | NM\_001080541 NM\_001164273 |
| MKL1 | NM\_001318139 NM\_001282661 NM\_020831 NM\_001282662 NM\_001282660 |
| **Gene** | **DB Accession number** |
| MLLT3 | NM\_001286691 NM\_004529 |
| MPL | NM\_005373 |
| MTHFR | NM\_005957 NM\_001330358 |
| MYC | NM\_001354870 NM\_002467 |
| MYH11 | NM\_022844 NM\_002474 NM\_001040114 NM\_001040113 |
| NF1 | NM\_001128147 NM\_001042492 NM\_000267 |
| NPM1 | NM\_002520 NM\_001355007 NM\_001355010 NM\_001037738 NM\_001355009 NM\_001355006 NM\_199185 |
| NRAS | NM\_002524 |
| NSD1 | NM\_172349 NM\_022455 |
| NTRK3 | NM\_001320135 NM\_001320134 NM\_001243101 NM\_002530 NM\_001012338 NM\_001007156 |
| NUDT15 | NM\_001304745 NM\_018283 |
| NUP214 | NM\_001318324 NM\_005085 NM\_001318325 |
| NUP98 | NM\_016320 NM\_139132 NM\_005387 NM\_139131 |
| P2RY8 | NM\_178129 NM\_178129\_2 |
| PAX5 | NM\_001280555 NM\_001280554 NM\_001280553 NM\_001280551 NM\_001280552 NM\_001280550 |
| NM\_001280556 NM\_016734 NM\_001280547 NM\_001280548 NM\_001280549 |
| PBX1 | NM\_001204961 NM\_001204963 NM\_001353131 NM\_001353130 NM\_002585 |
| PCM1 | NM\_001352637 NM\_001352639 NM\_006197 NM\_001352633 NM\_001352650 NM\_001352649 |
| NM\_001352640 NM\_001352658 NM\_001352646 NM\_001352659 NM\_001315508 NM\_001352654 NM\_001352657 NM\_001315507 NM\_001352636 NM\_001352638 NM\_001352641 |
| NM\_001352660 NM\_001352643 NM\_001352655 NM\_001352656 NM\_001352634 NM\_001352635 |
| NM\_001352642 NM\_001352644 NM\_001352645 NM\_001352647 NM\_001352648 NM\_001352651 |
| NM\_001352652 NM\_001352653 NM\_001352632 |
| PDGFRA | NM\_001347830 NM\_001347829 NM\_006206 NM\_001347827 NM\_001347828 |
| PDGFRB | NM\_001355016 NM\_002609 NM\_001355017 |
| PHF6 | NM\_032335 NM\_001015877 NM\_032458 |
| PIP4K2A | NM\_001330062 NM\_005028 |
| PML | NM\_033239 NM\_033240 NM\_033244 NM\_033246 NM\_033249 NM\_033247 NM\_033238 |
| NM\_033250 NM\_002675 |
| PRPF8 | NM\_006445 |
| PTK2B | NM\_004103 NM\_173176 NM\_173175 NM\_173174 |
| PTPN11 | NM\_002834 NM\_080601 NM\_001330437 |
| RAD21 | NM\_006265 |
| RARA | NM\_001145302 NM\_001145301 NM\_001024809 NM\_000964 |
| RB1 | NM\_000321 |
| **Gene** | **DB Accession number** |
| RBM15 | NM\_001201545 NM\_022768 |
| RELN | NM\_005045 NM\_173054 |
| RPL10 | NM\_001256577 NM\_001256580 NM\_001303625 NM\_006013 NM\_001303624 NM\_001303626 |
| RUNX1 | NM\_001122607 NM\_001754 NM\_001001890 |
| RUNX1T1 | NM\_175636 NM\_004349 NM\_001198629 NM\_001198630 NM\_001198632 NM\_175635 |
| NM\_001198625 NM\_001198626 NM\_001198627 NM\_001198631 NM\_001198634 NM\_001198679 |
| NM\_001198628 NM\_001198633 NM\_175634 |
| SETBP1 | NM\_015559 NM\_001130110 |
| SF3B1 | NM\_001308824 NM\_012433 NM\_001005526 |
| SH2B3 | NM\_005475 NM\_001291424 |
| SLCO1B1 | NM\_006446 |
| SMC1A | NM\_006306 NM\_001281463 |
| SMC3 | NM\_005445 |
| SRP72 | NM\_006947 NM\_001267722 |
| SRSF2 | NM\_001195427 NM\_003016 |
| STAG2 | NM\_001042750 NM\_001042751 NM\_001042749 NM\_006603 NM\_001282418 |
| STAT5A | NM\_003152 NM\_001288718 NM\_001288719 NM\_001288720 |
| TAL1 | NM\_001290406 NM\_001290405 NM\_001290404 NM\_001290403 NM\_001287347 NM\_003189 |
| TCF3 | NM\_003200 NM\_001136139 NM\_001351778 NM\_001351779 |
| TERC | NR\_001566 |
| TERT | NM\_001193376 NM\_198253 |
| TET1 | NM\_030625 |
| TET2 | NM\_001127208 NM\_017628 |
| TLX1 | NM\_001195517 NM\_005521 |
| TLX3 | NM\_021025 |
| TP53 | NM\_001276699 NM\_001276698 NM\_001276761 NM\_001276760 NM\_001126117 NM\_001126118 |
| NM\_001126116 NM\_001126113 NM\_001126112 NM\_001126115 NM\_001126114 NM\_001276695 |
| NM\_001276697 NM\_001276696 NM\_000546 |
| TPMT | NM\_000367 NM\_001346817 NM\_001346818 |
| TSLP | NM\_138551 NM\_033035 |
| TYK2 | NM\_003331 |
| U2AF1 | NM\_006758 NM\_001025204 NM\_001025203 |
| WT1 | NM\_024426 NM\_024424 NM\_001198551 NM\_001198552 NM\_000378 |
| ZBTB7A | NM\_001317990 NM\_015898 |
| ZRSR2 | NM\_005089 |

3. 검사 및 정도 관리 정보 (Experiment Information and QC Data)

|  |  |  |  |
| --- | --- | --- | --- |
| **Q metric** | **Acceptable range** | **Parameters** | **Results** |
| DNA quality | >=3 | DIN | $zdnaQuality$ |
| Input DNA | >=200 | ng | $zinputDNA$ |
| PCR cycle | >=7 | cycles | $zpcrCycle$ |
| Total Hyb DNA | 500 ~ 1000 | ng | $ztotalHybDNA$ |
| Cluster density | 1000 ~ 1200 | K/mm2 | $zclusterDensity$ |
| Data throughput | >=200Mb | Mb | 390 |
| base >Q30 | >=90 | % | 95 |
| Mapped % | >=70 | % | 93 |
| Duplication rate | <=70 | % | 5 |
| on-target rate | >=20 | % | 46 |
| on-target coverage | >=100 | X | 387 |

4. 제품정보 (Panel Material Information)

|  |  |  |
| --- | --- | --- |
| **Panel** | **Reagent Kit** | **Sequencer** |
| $panel$ | Reagent Kit v2 (300 cycles) | $sequencer$ |

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