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Gene List for Reporting Germline Findings in Cancer Patients

# Document History

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## Version History

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| Version | Date | Description |
| 1.3 | 19/01/2017 | This tumour type-specific gene list will be used for reporting germline findings in cancer patients |
| 1.4 | 04/04/2017 |  |
| 1.5 | 14/07/2017 | Added list of genes for Unknown tumours and Upper Gastrointestinal |
| 1.6 | 17/12/2017 | Added list of genes for Childhood tumours |
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## Reviewers

This document must be reviewed by the following:

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| Name | Title | Version |
| Ellen McDonagh | Lead Scientific Curator | 1.3 |
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## Approvers

This document must be approved by the following:

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| Name  | Responsibility | Date  | Version |
| Augusto Rendon | Director of Bioinformatics | 19/01/2017 | 1.3 |
| Clare Turnbull  | Clinical Lead for Cancer Data | 19/01/2017 | 1.3 |
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Table 1: List of genes for pertinent cancer findings per tumour type

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Gene Symbol | Ensembl Gene ID | Ensembl Transcript | Tumour Type | Variant Type |
| APC | ENSG00000134982 | ENST00000508376 | Adult Glioma | Loss-of-function |
| ATM\* | ENSG00000149311 | ENST00000278616 | Adult Glioma | Loss-of-function |
| MLH1 | ENSG00000076242 | ENST00000231790 | Adult Glioma | Loss-of-function |
| MSH2 | ENSG00000095002 | ENST00000233146 | Adult Glioma | Loss-of-function |
| MSH6 | ENSG00000116062 | ENST00000234420 | Adult Glioma | Loss-of-function |
| PMS2 | ENSG00000122512 | ENST00000265849 | Adult Glioma | Loss-of-function |
| TP53 | ENSG00000141510 | ENST00000269305 | Adult Glioma | Loss-of-function |
| BRCA1 | ENSG00000012048 | ENST00000357654 | Breast | Loss-of-function |
| BRCA2 | ENSG00000139618 | ENST00000544455 | Breast | Loss-of-function |
| PALB2 | ENSG00000083093 | ENST00000261584 | Breast | Loss-of-function |
| PTEN | ENSG00000171862 | ENST00000371953 | Breast | Loss-of-function |
| TP53 | ENSG00000141510 | ENST00000269305 | Breast | Loss-of-function |
| APC | ENSG00000134982 | ENST00000508376 | Colorectal | Loss-of-function |
| BMPR1A | ENSG00000107779 | ENST00000372037 | Colorectal | Loss-of-function |
| MLH1 | ENSG00000076242 | ENST00000231790 | Colorectal | Loss-of-function |
| MSH2 | ENSG00000095002 | ENST00000233146 | Colorectal | Loss-of-function |
| MSH6 | ENSG00000116062 | ENST00000234420 | Colorectal | Loss-of-function |
| MUTYH\* | ENSG00000132781 | ENST00000450313 | Colorectal | Loss-of-function |
| PMS2 | ENSG00000122512 | ENST00000265849 | Colorectal | Loss-of-function |
| POLD1 | ENSG00000062822 | ENST00000440232 | Colorectal | Loss-of-function |
| POLE | ENSG00000177084 | ENST00000320574 | Colorectal | Loss-of-function |
| PTEN | ENSG00000171862 | ENST00000371953 | Colorectal | Loss-of-function |
| SMAD4 | ENSG00000141646 | ENST00000342988 | Colorectal | Loss-of-function |
| STK11 | ENSG00000118046 | ENST00000326873 | Colorectal | Loss-of-function |
| FH | ENSG00000091483 | ENST00000366560 | Endometrial Carcinoma | Loss-of-function |
| MLH1 | ENSG00000076242 | ENST00000231790 | Endometrial Carcinoma | Loss-of-function |
| MSH2 | ENSG00000095002 | ENST00000233146 | Endometrial Carcinoma | Loss-of-function |
| MSH6 | ENSG00000116062 | ENST00000234420 | Endometrial Carcinoma | Loss-of-function |
| PMS2 | ENSG00000122512 | ENST00000265849 | Endometrial Carcinoma | Loss-of-function |
| PTEN | ENSG00000171862 | ENST00000371953 | Endometrial Carcinoma | Loss-of-function |
| MLH1 | ENSG00000076242 | ENST00000231790 | Hepatopancreatobiliary/ Upper Gastrointestinal | Loss-of-function |
| MSH2 | ENSG00000095002 | ENST00000233146 | Hepatopancreatobiliary/ Upper Gastrointestinal | Loss-of-function |
| MSH6 | ENSG00000116062 | ENST00000234420 | Hepatopancreatobiliary/ Upper Gastrointestinal | Loss-of-function |
| PMS2 | ENSG00000122512 | ENST00000265849 | Hepatopancreatobiliary/ Upper Gastrointestinal | Loss-of-function |
| BAP1 | ENSG00000163930 | ENST00000460680 | Malignant Melanoma | Loss-of-function |
| CDK4 | ENSG00000135446 | ENST00000257904 | Malignant Melanoma | Activating |
| CDKN2A | ENSG00000147889 | ENST00000304494 | Malignant Melanoma | Loss-of-function |
| BRCA1 | ENSG00000012048 | ENST00000357654 | Ovarian | Loss-of-function |
| BRCA2 | ENSG00000139618 | ENST00000544455 | Ovarian | Loss-of-function |
| MLH1 | ENSG00000076242 | ENST00000231790 | Ovarian | Loss-of-function |
| MSH2 | ENSG00000095002 | ENST00000233146 | Ovarian | Loss-of-function |
| MSH6 | ENSG00000116062 | ENST00000234420 | Ovarian | Loss-of-function |
| PMS2 | ENSG00000122512 | ENST00000265849 | Ovarian | Loss-of-function |
| RAD51C | ENSG00000108384 | ENST00000337432 | Ovarian | Loss-of-function |
| RAD51D | ENSG00000185379 | ENST00000345365 | Ovarian | Loss-of-function |
| BRCA2 | ENSG00000139618 | ENST00000544455 | Prostate | Loss-of-function |
| FH | ENSG00000091483 | ENST00000366560 | Renal | Loss-of-function |
| FLCN | ENSG00000154803 | ENST00000285071 | Renal | Loss-of-function |
| MET | ENSG00000105976 | ENST00000318493 | Renal | Activating |
| PTEN | ENSG00000171862 | ENST00000371953 | Renal | Loss-of-function |
| SDHB | ENSG00000117118 | ENST00000375499 | Renal | Loss-of-function |
| VHL | ENSG00000134086 | ENST00000256474 | Renal | Loss-of-function |
| TP53 | ENSG00000141510 | ENST00000269305 | Sarcoma | Loss-of-function |
| APC | ENSG00000134982 | ENST00000508376 | Unknown | Loss-of-function |
| ATM | ENSG00000149311 | ENST00000278616 | Unknown | Loss-of-function |
| BAP1 | ENSG00000163930 | ENST00000460680 | Unknown | Loss-of-function |
| BMPR1A | ENSG00000107779 | ENST00000372037 | Unknown | Loss-of-function |
| BRCA1 | ENSG00000012048 | ENST00000357654 | Unknown | Loss-of-function |
| BRCA2 | ENSG00000139618 | ENST00000544455 | Unknown | Loss-of-function |
| BRIP1 | ENSG00000136492 | ENST00000259008 | Unknown | Loss-of-function |
| CDC73 | ENSG00000134371 | ENST00000367435 | Unknown | Loss-of-function |
| CDH1 | ENSG00000039068 | ENST00000261769 | Unknown | Loss-of-function |
| CDK4 | ENSG00000135446 | ENST00000257904 | Unknown | Activating |
| CDKN2A | ENSG00000147889 | ENST00000304494 | Unknown | Loss-of-function |
| CHEK2 | ENSG00000183765 | ENST00000328354 | Unknown | Loss-of-function |
| DICER1 | ENSG00000100697 | ENST00000343455 | Unknown | Loss-of-function |
| EPCAM | ENSG00000119888 | ENST00000263735 | Unknown | Loss-of-function |
| FH | ENSG00000091483 | ENST00000366560 | Unknown | Loss-of-function |
| FLCN | ENSG00000154803 | ENST00000285071 | Unknown | Loss-of-function |
| KIT | ENSG00000157404 | ENST00000288135 | Unknown | Activating |
| MAX | ENSG00000125952 | ENST00000358664 | Unknown | Loss-of-function |
| MEN1 | ENSG00000133895 | ENST00000394374 | Unknown | Loss-of-function |
| MET | ENSG00000105976 | ENST00000318493 | Unknown | Activating |
| MLH1 | ENSG00000076242 | ENST00000231790 | Unknown | Loss-of-function |
| MSH2 | ENSG00000095002 | ENST00000233146 | Unknown | Loss-of-function |
| MSH6 | ENSG00000116062 | ENST00000234420 | Unknown | Loss-of-function |
| MUTYH\* | ENSG00000132781 | ENST00000450313 | Unknown | Loss-of-function |
| NF1 | ENSG00000196712 | ENST00000358273 | Unknown | Loss-of-function |
| NF2 | ENSG00000186575 | ENST00000338641 | Unknown | Loss-of-function |
| NTHL1\* | ENSG00000065057 | ENST00000219066 | Unknown | Loss-of-function |
| PALB2 | ENSG00000083093 | ENST00000261584 | Unknown | Loss-of-function |
| PMS2 | ENSG00000122512 | ENST00000265849 | Unknown | Loss-of-function |
| POLD1 | ENSG00000062822 | ENST00000440232 | Unknown | Loss-of-function |
| POLE | ENSG00000177084 | ENST00000320574 | Unknown | Loss-of-function |
| PTCH1 | ENSG00000185920 | ENST00000331920 | Unknown | Loss-of-function |
| PTEN | ENSG00000171862 | ENST00000371953 | Unknown | Loss-of-function |
| RAD51C | ENSG00000108384 | ENST00000337432 | Unknown | Loss-of-function |
| RAD51D | ENSG00000185379 | ENST00000345365 | Unknown | Loss-of-function |
| RB1 | ENSG00000139687 | ENST00000267163 | Unknown | Loss-of-function |
| RET | ENSG00000165731 | ENST00000355710 | Unknown | Activating |
| SDHA | ENSG00000073578 | ENST00000264932 | Unknown | Loss-of-function |
| SDHAF2 | ENSG00000167985 | ENST00000301761 | Unknown | Loss-of-function |
| SDHB | ENSG00000117118 | ENST00000375499 | Unknown | Loss-of-function |
| SDHC | ENSG00000143252 | ENST00000367975 | Unknown | Loss-of-function |
| SDHD | ENSG00000204370 | ENST00000375549 | Unknown | Loss-of-function |
| SMAD4 | ENSG00000141646 | ENST00000342988 | Unknown | Loss-of-function |
| SMARCA4 | ENSG00000127616 | ENST00000450717 | Unknown | Loss-of-function |
| SMARCB1 | ENSG00000099956 | ENST00000263121 | Unknown | Loss-of-function |
| STK11 | ENSG00000118046 | ENST00000326873 | Unknown | Loss-of-function |
| SUFU | ENSG00000107882 | ENST00000369902 | Unknown | Loss-of-function |
| TMEM127 | ENSG00000135956 | ENST00000258439 | Unknown | Loss-of-function |
| TP53 | ENSG00000141510 | ENST00000269305 | Unknown | Loss-of-function |
| TSC1 | ENSG00000165699 | ENST00000298552 | Unknown | Loss-of-function |
| TSC2 | ENSG00000103197 | ENST00000219476 | Unknown | Loss-of-function |
| VHL | ENSG00000134086 | ENST00000256474 | Unknown | Loss-of-function |
| WT1 | ENSG00000184937 | ENST00000332351 | Unknown | Loss-of-function |
| CDKN1B | ENSG00000111276 | ENST00000228872 | Unknown | Loss-of-function |
| ALK\* | ENSG00000171094 | ENST00000389048 | Childhood | Activating |
| APC\* | ENSG00000134982 | ENST00000508376 | Childhood | Loss-of-function |
| ATM | ENSG00000149311 | ENST00000278616 | Childhood | Loss-of-function |
| BLM | ENSG00000197299 | ENST00000355112 | Childhood | Loss-of-function |
| BMPR1A\* | ENSG00000107779 | ENST00000372037 | Childhood | Loss-of-function |
| BRAF\* | ENSG00000157764 | ENST00000288602 | Childhood | Activating |
| BRCA1 | ENSG00000012048 | ENST00000357654 | Childhood | Loss-of-function |
| BRCA2 | ENSG00000139618 | ENST00000544455 | Childhood | Loss-of-function |
| BRIP1 | ENSG00000136492 | ENST00000259008 | Childhood | Loss-of-function |
| CBL\* | ENSG00000110395 | ENST00000264033 | Childhood | Loss-of-function |
| CDKN1C\* | ENSG00000129757 | ENST00000414822 | Childhood | Loss-of-function |
| DDB2 | ENSG00000134574 | ENST00000256996 | Childhood | Loss-of-function |
| DICER1\* | ENSG00000100697 | ENST00000343455 | Childhood | Loss-of-function |
| ERCC1 | ENSG00000012061 | ENST00000300853 | Childhood | Loss-of-function |
| ERCC2 | ENSG00000104884 | ENST00000391945 | Childhood | Loss-of-function |
| ERCC3 | ENSG00000163161 | ENST00000285398 | Childhood | Loss-of-function |
| ERCC4 | ENSG00000175595 | ENST00000311895 | Childhood | Loss-of-function |
| ERCC5 | ENSG00000134899 | ENST00000355739 | Childhood | Loss-of-function |
| FANCA | ENSG00000187741 | ENST00000389301 | Childhood | Loss-of-function |
| FANCC | ENSG00000158169 | ENST00000289081 | Childhood | Loss-of-function |
| FANCD2 | ENSG00000144554 | ENST00000287647 | Childhood | Loss-of-function |
| FANCE | ENSG00000112039 | ENST00000229769 | Childhood | Loss-of-function |
| FANCF | ENSG00000183161 | ENST00000327470 | Childhood | Loss-of-function |
| FANCG | ENSG00000221829 | ENST00000378643 | Childhood | Loss-of-function |
| FANCI | ENSG00000140525 | ENST00000310775 | Childhood | Loss-of-function |
| FANCL | ENSG00000115392 | ENST00000402135 | Childhood | Loss-of-function |
| FANCM | ENSG00000187790 | ENST00000267430 | Childhood | Loss-of-function |
| HRAS\* | ENSG00000174775 | ENST00000451590 | Childhood | Loss-of-function |
| KRAS\* | ENSG00000133703 | ENST00000256078 | Childhood | Loss-of-function |
| MAP2K1\* | ENSG00000169032 | ENST00000307102 | Childhood | Activating |
| MAP2K2\* | ENSG00000126934 | ENST00000262948 | Childhood | Activating |
| MEN1\* | ENSG00000133895 | ENST00000394374 | Childhood | Loss-of-function |
| MLH1 | ENSG00000076242 | ENST00000231790 | Childhood | Loss-of-function |
| MSH2 | ENSG00000095002 | ENST00000233146 | Childhood | Loss-of-function |
|  |  |  |  |  |
| MSH6 | ENSG00000116062 | ENST00000234420 | Childhood | Loss-of-function |
| NBN | ENSG00000104320 | ENST00000265433 | Childhood | Loss-of-function |
| NF1\* | ENSG00000196712 | ENST00000358273 | Childhood | Loss-of-function |
| NF2\* | ENSG00000186575 | ENST00000338641 | Childhood | Loss-of-function |
| NHP2 | ENSG00000145912 | ENST00000274606 | Childhood | Loss-of-function |
| NOP10 | ENSG00000182117 | ENST00000328848 | Childhood | Loss-of-function |
| NRAS\* | ENSG00000213281 | ENST00000369535 | Childhood | Loss-of-function |
| PALB2 | ENSG00000083093 | ENST00000261584 | Childhood | Loss-of-function |
| PAX5\* | ENSG00000196092 | ENST00000358127 | Childhood | Loss-of-function |
| PHOX2B\* | ENSG00000109132 | ENST00000226382 | Childhood | Loss-of-function |
| PMS2 | ENSG00000122512 | ENST00000265849 | Childhood | Loss-of-function |
| PRKAR1A\* | ENSG00000108946 | ENST00000392711 | Childhood | Loss-of-function |
| PTCH1\* | ENSG00000185920 | ENST00000331920 | Childhood | Loss-of-function |
| PTEN\* | ENSG00000171862 | ENST00000371953 | Childhood | Loss-of-function |
| PTPN11\* | ENSG00000179295 | ENST00000351677 | Childhood | Loss-of-function |
| RAD51C | ENSG00000108384 | ENST00000337432 | Childhood | Loss-of-function |
| RAF1\* | ENSG00000132155 | ENST00000251849 | Childhood | Loss-of-function |
| RB1\* | ENSG00000139687 | ENST00000267163 | Childhood | Loss-of-function |
| RECQL4 | ENSG00000160957 | ENST00000617875 | Childhood | Loss-of-function |
| RET\* | ENSG00000165731 | ENST00000355710 | Childhood | Activating |
| SH2B3 | ENSG00000111252 | ENST00000341259 | Childhood | Loss-of-function |
| SHOC2\* | ENSG00000108061 | ENST00000369452 | Childhood | Loss-of-function |
| SMAD4\* | ENSG00000141646 | ENST00000342988 | Childhood | Loss-of-function |
| SMARCA4\* | ENSG00000127616 | ENST00000450717 | Childhood | Loss-of-function |
| SMARCB1\* | ENSG00000099956 | ENST00000263121 | Childhood | Loss-of-function |
| SOS1\* | ENSG00000115904 | ENST00000402219 | Childhood | Loss-of-function |
| STK11\* | ENSG00000118046 | ENST00000326873 | Childhood | Loss-of-function |
| SUFU\* | ENSG00000107882 | ENST00000369902 | Childhood | Loss-of-function |
| TERT | ENSG00000164362 | ENST00000310581 | Childhood | Loss-of-function |
| TP53\* | ENSG00000141510 | ENST00000269305 | Childhood | Loss-of-function |
| TSC1\* | ENSG00000165699 | ENST00000298552 | Childhood | Loss-of-function |
| TSC2\* | ENSG00000103197 | ENST00000219476 | Childhood | Loss-of-function |
| VHL | ENSG00000134086 | ENST00000256474 | Childhood | Loss-of-function |
| WRN | ENSG00000165392 | ENST00000298139 | Childhood | Loss-of-function |
| WT1\* | ENSG00000184937 | ENST00000332351 | Childhood | Loss-of-function |
|  |  |  |  |  |
| XPA | ENSG00000136936 | ENST00000375128 | Childhood | Loss-of-function |
| XPC | ENSG00000154767 | ENST00000285021 | Childhood | Loss-of-function |

\* Only biallelic germline mutations are reported for these genes