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| --- | --- | --- | --- |
|  |  | Document Record ID Key | |
| Work stream | Project B |  | |
| Programme Director | Augusto Rendon | **Status** |  |
| Document Owner | Ellen McDonagh | **Version** | 1.4 |
| Document Author | ­­­Ellen McDonagh | **Version Date** | 04/04/2017 |

Gene List for Reporting Germline Findings in Cancer Patients

# Document History

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## Version History

|  |  |  |
| --- | --- | --- |
| 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 |  |
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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

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| --- | --- | --- | --- | --- |
| 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 | Loss-of-function |
| MSH2 | ENSG00000095002 | ENST00000233146 | Hepatopancreatobiliary | Loss-of-function |
| MSH6 | ENSG00000116062 | ENST00000234420 | Hepatopancreatobiliary | Loss-of-function |
| PMS2 | ENSG00000122512 | ENST00000265849 | Hepatopancreatobiliary | 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 |

\* Only biallelic germline mutations are reported for ATM and MUTYH genes