# HEREDITARY CANCER GENE PANEL DGD20062014

| Gene | Median coverage | % covered > 10x | % covered > 20x | Associated Phenotype description and OMIM ID |
|---|---|---|---|---|
| ALK | 92,6 | 99% | 96% | {Neuroblastoma, susceptibility to, 3}, 613014 |
| APC | 142,8 | 100% | 99% | Adenomatous polyposis coli, 175100<br>Gastric cancer, somatic, 613659<br>Adenoma, periampullary, somatic<br>Hepatoblastoma, somatic, 114550<br>Desmoid disease, hereditary, 135290<br>Colorectal cancer, somatic, 114500<br>Brain tumor-polyposis syndrome 2, 175100 Gardner syndrome, 175100 |
| ATM | 115,2 | 100% | 99% | Ataxia-telangiectasia, 208900<br>Lymphoma, B-cell non-Hodgkin, somatic<br>{Breast cancer, susceptibility to}, 114480<br>Lymphoma, mantle cell<br>T-cell prolymphocytic leukemia, somatic |
| ATR | 119,6 | 100% | 99% | Seckel syndrome 1, 210600<br>Cutaneous telangiectasia and cancer syndrome, familial, 614564 |
| BAP1 | 85,5 | 100% | 96% | Tumor predisposition syndrome, 614327 |
| BARD1 | 111,3 | 100% | 99% | {Breast cancer, susceptibility to}, 114480 |
| BLM | 113,4 | 99% | 98% | Bloom syndrome, 210900 |
| BMPR1A | 54,9 | 77% | 62% | Polyposis, juvenile intestinal, 174900<br>Polyposis syndrome, hereditary mixed, 2, 610069<br>Juvenile polyposis syndrome, infantile form, 174900 |
| BRCA1 | 152,2 | 99% | 96% | {Breast-ovarian cancer, familial, 1}, 604370<br>{Pancreatic cancer, susceptibility to, 4}, 614320 |

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| BRCA2 | 145,6 | 100% | 99% | {Breast-ovarian cancer, familial, 2}, 612555<br>Fanconi anemia, complementation group D1, 605724<br>Prostate cancer, 176807<br>{Breast cancer, male, susceptibility to}, 114480<br>Wilms tumor, 194070<br>{Medulloblastoma}, 155255<br>{Glioblastoma 3}, 613029<br>{Pre-B-cell acute lymphoblastic leukemia}<br>Pancreatic cancer, 613347 |
| BRIP1 | 129,7 | 100% | 99% | ?Breast cancer, early-onset, 114480<br>Fanconi anemia, complementation group J, 609054 |
| BUB1 | 107,7 | 99% | 98% | Colorectal cancer with chromosomal instability |
| BUB1B | 113,7 | 100% | 98% | Colorectal cancer, somatic, 114500<br>Mosaic variegated aneuploidy syndrome 1, 257300<br>[Premature chromatid separation trait], 176430 |
| BUB3 | 116,2 | 100% | 100% | No OMIM phenotype<br>Variegated aneuploidy (de Voer (2013) Gastroenterology 145, 544) |
| CDC73 | 147,1 | 100% | 100% | Hyperparathyroidism, familial primary, 145000<br>Hyperparathyroidism-jaw tumor syndrome, 145001<br>Parathyroid adenoma with cystic changes, 145001<br>Parathyroid carcinoma, 608266 |
| CDH1 | 114,1 | 100% | 99% | Endometrial carcinoma, somatic, 608089<br>Ovarian carcinoma, somatic, 167000<br>{Breast cancer, lobular}, 114480<br>Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215<br>{Prostate cancer, susceptibility to}, 176807 |
| CDK4 | 116,8 | 98% | 93% | {Melanoma, cutaneous malignant, 3}, 609048 |
| CDKN2A | 111,9 | 100% | 100% | {Melanoma, cutaneous malignant, 2}, 155601<br>Melanoma and neural system tumor syndrome, 155755<br>Pancreatic cancer/melanoma syndrome, 606719<br>Orolaryngeal cancer, multiple, -3 |
| CENPJ | 130,6 | 100% | 100% | Microcephaly 6, primary, autosomal recessive, 608393<br>Seckel syndrome 4, 613676 |

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| CHEK2 | 53,6 | 69% | 65% | Li-Fraumeni syndrome, 609265<br>Osteosarcoma, somatic, 259500<br>{Breast cancer, susceptibility to}, 114480<br>{Prostate cancer, familial, susceptibility to}, 176807<br>{Breast and colorectal cancer, susceptibility to} |
| CREBBP | 81,7 | 99% | 97% | Rubinstein-Taybi syndrome, 180849 |
| CTC1 | 90,7 | 100% | 97% | Cerebroretinal microangiopathy with calcifications and cysts, 612199 |
| CTNNA1 | 89,5 | 99% | 92% | No OMIM phenotype<br>Gastric cancer, diffuse (Majewski (2012) J Pathol epub) |
| CYLD | 112,6 | 100% | 99% | Cylindromatosis, familial, 132700<br>Brooke-Spiegler syndrome, 605041<br>Trichoepithelioma, multiple familial, 1, 601606 |
| DDB2 | 85,8 | 100% | 95% | Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 |
| DDX11 | 10,6 | 22% | 15% | Warsaw breakage syndrome, 613398 |
| DICER1 | 112,5 | 100% | 99% | Pleuropulmonary blastoma, 601200<br>Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 |
| DKC1 | 45,1 | 99% | 90% | Dyskeratosis congenita, X-linked, 305000 |
| EGFR | 93,2 | 100% | 99% | Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980<br>Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980<br>{Nonsmall cell lung cancer, susceptibility to}, 211980 |
| ELANE | 114,8 | 98% | 94% | Neutropenia, cyclic, 162800<br>Neutropenia, severe congenital 1, autosomal dominant, 202700 |
| ERCC1 | 79,1 | 100% | 97% | Cerebrooculofacioskeletal syndrome 4, 610758 |
| ERCC2 | 85,1 | 100% | 94% | Xeroderma pigmentosum, group D, 278730<br>Trichothiodystrophy, 601675<br>Cerebrooculofacioskeletal syndrome 2, 610756 |
| ERCC3 | 117,1 | 100% | 99% | Xeroderma pigmentosum, group B, 610651<br>Trichothiodystrophy, 601675 |
| ERCC4 | 146,1 | 98% | 95% | Xeroderma pigmentosum, group F, 278760<br>XFE progeroid syndrome, 610965 |

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| | | | | Fanconi anemia, complementation group Q, 615272<br>Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 |
| ERCC5 | 116,3 | 99% | 97% | Xeroderma pigmentosum, group G, 278780<br>Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 |
| ERCC6 | 147,6 | 98% | 97% | Cockayne syndrome, type B, 133540<br>Cerebrooculofacioskeletal syndrome 1, 214150<br>De Sanctis-Cacchione syndrome, 278800<br>{Macular degeneration, age-related, susceptibility to 5}, 613761<br>UV-sensitive syndrome 1, 600630<br>{Lung cancer, susceptibility to}, 211980 |
| EXO1 | 118,4 | 95% | 94% | No OMIM phenotype<br>Colorectal cancer, non-polyposis (Wu (2001) Gastroenterology 120,1580) |
| EXT1 | 95,2 | 99% | 98% | Exostoses, multiple, type 1, 133700<br>Chondrosarcoma, 215300 |
| EXT2 | 113,6 | 100% | 100% | Exostoses, multiple, type 2, 133701 |
| FANCA | 81,3 | 99% | 97% | Fanconi anemia, complementation group A, 227650 |
| FANCB | 63,4 | 96% | 89% | Fanconi anemia, complementation group B, 300514 |
| FANCC | 81,8 | 100% | 98% | Fanconi anemia, complementation group C, 227645 |
| FANCD2 | 98,7 | 88% | 86% | Fanconi anemia, complementation group D2, 227646 |
| FANCE | 85 | 94% | 88% | Fanconi anemia, complementation group E, 600901 |
| FANCF | 156,1 | 100% | 100% | Fanconi anemia, complementation group F, 603467 |
| FANCG | 120,4 | 100% | 96% | Fanconi anemia, complementation group G, 614082 |
| FANCI | 120,4 | 100% | 100% | Fanconi anemia, complementation group I, 609053 |
| FANCL | 96 | 100% | 99% | Fanconi anemia, complementation group L, 614083 |
| FANCM | 116,9 | 100% | 99% | Fanconi anemia, complementation group M, 614087 |

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| FH | 86,7 | 99% | 91% | Fumarase deficiency, 606812<br>Leiomyomatosis and renal cell cancer, 150800 |
| FLCN | 98,7 | 99% | 96% | Birt-Hogg-Dube syndrome, 135150<br>Pneumothorax, primary spontaneous, 173600<br>Renal carcinoma, chromophobe, somatic, 144700<br>Colorectal cancer, somatic, 114500 |
| GDNF | 156,7 | 100% | 100% | Central hypoventilation syndrome, 209880<br>{Pheochromocytoma, modifier of}, 171300<br>{Hirschsprung disease, susceptibility to, 3}, 613711 |
| GFI1 | 68,6 | 100% | 100% | Neutropenia, severe congenital 2, autosomal dominant, 613107<br>Neutropenia, nonimmune chronic idiopathic, of adults, 607847 |
| GPC3 | 46,1 | 100% | 96% | Simpson-Golabi-Behmel syndrome, type 1, 312870<br>Wilms tumor, somatic, 194070 |
| GREM1 | 147,4 | 100% | 100% | No OMIM phenotype<br>{Colorectal cancer, increased risk, association with}(Peters (2012) Hum Genet 131,217)<br>Oligosyndactyly of the hands, Cenani-Linz-like (Dimitrov (2010) J Med Genet 47,569)<br>Mixed polyposis syndrome (Jaeger (2012) Nat Genet 44,699) |
| HAX1 | 131,5 | 100% | 100% | Neutropenia, severe congenital 3, autosomal recessive, 610738 |
| HNF1A | 79,8 | 99% | 96% | MODY, type III, 600496<br>{Diabetes mellitus, noninsulin-dependent, 2}, 125853<br>{Diabetes mellitus, insulin-dependent}, 222100<br>Hepatic adenoma, somatic, 142330<br>Renal cell carcinoma, 144700<br>Diabetes mellitus, insulin-dependent, 20, 612520 |
| KIF1B | 114,8 | 100% | 98% | Charcot-Marie-Tooth disease, type 2A1, 118210<br>Pheochromocytoma, 171300<br>{Neuroblastoma, susceptibility to, 1}, 256700 |
| KIT | 108,8 | 99% | 95% | Piebaldism, 172800<br>Gastrointestinal stromal tumor, familial, 606764<br>Mast cell disease, 154800<br>Leukemia, acute myeloid, 601626<br>Germ cell tumors, 273300 |

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| KLLN | 101 | 100% | 100% | Cowden syndrome 4, 615107 |
| MAX | 73,9 | 90% | 86% | {Pheochromocytoma, susceptibility to}, 171300 |

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| MEN1 | 108,4 | 99% | 95% | Multiple endocrine neoplasia 1, 131100<br>Carcinoid tumor of lung<br>Parathyroid adenoma, somatic<br>Lipoma, somatic<br>Angiofibroma, somatic<br>Adrenal adenoma, somatic |
| MLH1 | 99,4 | 100% | 98% | Colorectal cancer, hereditary nonpolyposis, type 2, 609310<br>Mismatch repair cancer syndrome, 276300<br>Muir-Torre syndrome, 158320 |
| MLH3 | 161,5 | 99% | 98% | Colorectal cancer, somatic, 114500<br>Colorectal cancer, hereditary nonpolyposis, type 7, 614385<br>Endometrial cancer, 608089 |
| MPL | 108 | 100% | 93% | Thrombocytopenia, congenital amegakaryocytic, 604498<br>Thrombocythemia 2, 601977<br>Myelofibrosis with myeloid metaplasia, somatic, 254450 |
| MRE11A | 90,7 | 100% | 100% | Ataxia-telangiectasia-like disorder, 604391 |
| MSH2 | 100,5 | 99% | 97% | Colorectal cancer, hereditary nonpolyposis, type 1, 120435<br>Muir-Torre syndrome, 158320<br>Mismatch repair cancer syndrome, 276300 |
| MSH6 | 145,2 | 100% | 100% | Colorectal cancer, hereditary nonpolyposis, type 5, 614350<br>Endometrial cancer, familial, 608089<br>Mismatch repair cancer syndrome, 276300 |
| MUTYH | 109,5 | 100% | 100% | Adenomas, multiple colorectal, 608456<br>Gastric cancer, somatic, 613659<br>Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 |

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| NF1 | 80 | 83% | 80% | Neurofibromatosis, type 1, 162200<br>Leukemia, juvenile myelomonocytic, 607785<br>Melanoma, desmoplastic neurotrophic (2)<br>Neurofibromatosis, familial spinal, 162210<br>Neurofibromatosis-Noonan syndrome, 601321<br>Watson syndrome, 193520 |
| NF2 | 81,5 | 99% | 94% | Neurofibromatosis, type 2, 101000<br>Meningioma, NF2-related, somatic, 607174<br>Schwannomatosis, 162091 |
| NHP2 | 47,6 | 100% | 92% | Dyskeratosis congenita, autosomal recessive 2, 613987 |
| NOP10 | 160,8 | 100% | 100% | Dyskeratosis congenita, autosomal recessive 1, 224230 |
| NSD1 | 117,3 | 100% | 99% | Sotos syndrome 1, 117550<br>Leukemia, acute myeloid, 601626 (1)<br>Beckwith-Wiedemann syndrome, 130650 |
| OGG1 | 88,5 | 99% | 95% | Renal cell carcinoma, clear cell, somatic, 144700 |
| PALB2 | 128 | 99% | 98% | Fanconi anemia, complementation group N, 610832<br>{Breast cancer, susceptibility to}, 114480<br>{Pancreatic cancer, susceptibility to, 3}, 613348 |
| PHOX2B | 59,7 | 100% | 88% | Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880<br>{Neuroblastoma, susceptibility to, 2}, 613013<br>Neuroblastoma with Hirschsprung disease, 613013 |
| PMS1 | 115,1 | 100% | 100% | No OMIM phenotype<br>Colorectal cancer, non-polyposis (Nicolaides (1994) Nature 371,75) |
| PMS2 | 62,2 | 56% | 54% | Mismatch repair cancer syndrome, 276300<br>Colorectal cancer, hereditary nonpolyposis, type 4, 614337 |
| POLD1 | 78,5 | 95% | 90% | {Colorectal cancer, susceptibility to, 10}, 612591<br>Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 |
| POLE | 99,5 | 100% | 98% | {Colorectal cancer, susceptibility to, 12}, 615083<br>FILS syndrome, 615139 |
| PRF1 | 85,5 | 100% | 99% | Hemophagocytic lymphohistiocytosis, familial, 2, 603553<br>Lymphoma, non-Hodgkin, 605027 |

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| PRKAR1A | 101,1 | 97% | 92% | Carney complex, type 1, 160980<br>Myxoma, intracardiac, 255960<br>Thyroid carcinoma, papillary, somatic, 188550<br>Pigmented nodular adrenocortical disease, primary, 1, 610489<br>Adrenocortical tumor, somatic,<br>Acrodysostosis 1, with or without hormone resistance, 101800 |
| PTCH1 | 83,8 | 99% | 94% | Basal cell nevus syndrome, 109400<br>Basal cell carcinoma, somatic, 605462<br>Holoprosencephaly-7, 610828 |
| PTEN | 121,7 | 100% | 97% | Cowden syndrome 1, 158350<br>Lhermitte-Duclos syndrome, 158350<br>Bannayan-Riley-Ruvalcaba syndrome, 153480<br>{Meningioma}, 607174<br>{Glioma susceptibility 2}, 613028<br>Macrocephaly/autism syndrome, 605309<br>PTEN hamartoma tumor syndrome<br>VATER association with macrocephaly and ventriculomegaly, 276950<br>{Prostate cancer, somatic}, 176807<br>Thyroid carcinoma, follicular, somatic, 188470<br>Malignant melanoma, somatic, 155600<br>Endometrial carcinoma, somatic, 608089<br>Squamous cell carcinoma, head and neck, somatic, 275355 |
| PTPRJ | 111,4 | 97% | 96% | Colon cancer, somatic, 114500 |
| RAD50 | 110,8 | 100% | 99% | Nijmegen breakage syndrome-like disorder, 613078 |
| RAD51C | 99 | 100% | 100% | Fanconi anemia, complementation group O, 613390<br>{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 |
| RAD51D | 64,7 | 98% | 90% | {Breast-ovarian cancer, familial, susceptibility to, 4}, 614291 |
| RB1 | 112,8 | 99% | 98% | Retinoblastoma, 180200<br>Osteosarcoma, somatic, 259500<br>Bladder cancer, somatic, 109800<br>Small cell cancer of the lung, somatic, 182280<br>Retinoblastoma, trilateral, 180200 |

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| RECQL4 | 90,1 | 98% | 97% | Rothmund-Thomson syndrome, 268400<br>RAPADILINO syndrome, 266280<br>Baller-Gerold syndrome, 218600 |
| RET | 87,3 | 96% | 92% | Multiple endocrine neoplasia IIA, 171400<br>Medullary thyroid carcinoma, 155240<br>Multiple endocrine neoplasia IIB, 162300<br>Central hypoventilation syndrome, congenital, 209880<br>Pheochromocytoma, 171300<br>Renal agenesis, 191830<br>{Hirschsprung disease, susceptibility to, 1}, 142623 |
| RPL11 | 70,9 | 96% | 89% | Diamond-Blackfan anemia 7, 612562 |
| RPL35A | 29,8 | 84% | 60% | Diamond-Blackfan anemia 5, 612528 |
| RPL5 | 32,7 | 72% | 57% | Diamond-Blackfan anemia 6, 612561 |
| RPS10 | 33 | 88% | 72% | Diamond-Blackfan anemia 9, 613308 |
| RPS17 | 0 | 0% | 0% | Diamond-Blackfan anemia 4, 612527 |
| RPS19 | 33,7 | 48% | 42% | Diamond-Blackfan anemia 1, 105650 |
| RPS24 | 87,5 | 100% | 92% | Diamond-blackfan anemia 3, 610629 |
| RPS26 | 37 | 73% | 57% | Diamond-Blackfan anemia 10, 613309 |
| RPS7 | 19,7 | 74% | 43% | Diamond-Blackfan anemia 8, 612563 |
| RTEL1 | 84,5 | 99% | 94% | Dyskeratosis congenita, autosomal recessive 5, 615190<br>Dyskeratosis congenita, autosomal dominant 4, 615190 |
| RUNX1 | 66,3 | 96% | 86% | Leukemia, acute myeloid, 601626<br>Platelet disorder, familial, with associated myeloid malignancy, 601399 |
| SBDS | 71,4 | 99% | 91% | Shwachman-Bodian-Diamond syndrome, 260400 |

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| SDHA | 10,9 | 29% | 17% | Leigh syndrome, 256000<br>Mitochondrial respiratory chain complex II deficiency, 252011<br>Cardiomyopathy, dilated, 1GG, 613642<br>Paragangliomas 5, 614165 |
| SDHAF2 | 92,8 | 100% | 100% | Paragangliomas 2, 601650 |
| SDHB | 96,5 | 100% | 100% | Paragangliomas 4, 115310<br>Pheochromocytoma, 171300<br>Paraganglioma and gastric stromal sarcoma, 606864<br>Cowden syndrome 2, 612359<br>Gastrointestinal stromal tumor, 606764 |
| SDHC | 22,8 | 46% | 31% | Paragangliomas 3, 605373<br>Paraganglioma and gastric stromal sarcoma, 606864<br>Gastrointestinal stromal tumor, 606764 |
| SDHD | 14,5 | 42% | 20% | Paragangliomas 1, with or without deafness, 168000<br>Pheochromocytoma, 171300<br>Carcinoid tumors, intestinal, 114900<br>Merkel cell carcinoma, somatic<br>Paraganglioma and gastric stromal sarcoma, 606864<br>Cowden syndrome 3, 615106 |
| SLX4 | 128,2 | 97% | 94% | Fanconi anemia, complementation group P, 613951 |
| SMAD4 | 129,6 | 100% | 93% | Pancreatic cancer<br>Polyposis, juvenile intestinal, 174900<br>Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050<br>Myhre syndrome, 139210 |
| SMARCA4 | 81,6 | 97% | 91% | Rhabdoid tumor predisposition syndrome 2, 613325<br>Mental retardation, autosomal dominant 16, 614609 |
| SMARCB1 | 117 | 100% | 100% | Rhabdoid tumors, somatic, 609322<br>Rhabdoid predisposition syndrome 1, 609322<br>Mental retardation, autosomal dominant 15, 614608 |
| SQSTM1 | 79,4 | 99% | 97% | Paget disease of bone, 602080 |

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| STK11 | 74,8 | 100% | 99% | Peutz-Jeghers syndrome, 175200<br>Melanoma, malignant, somatic<br>Pancreatic cancer, 260350<br>Testicular tumor, somatic, 273300 |
| SUFU | 86,2 | 94% | 85% | Medulloblastoma, desmoplastic, 155255<br>{Meningioma, familial, susceptibility to}, 607174 |
| TERT | 106,7 | 100% | 98% | {Bone marrow failure, telomere-related, 1}, 614742<br>{Dyskeratosis congenita, autosomal recessive 4}, 613989<br>{Dyskeratosis congenita, autosomal dominant 2}, 613989<br>{Coronary artery disease}<br>{Pulmonary fibrosis, telomere-related, 1}, 614742<br>{Leukemia, acute myeloid}, 601626<br>{Melanoma, cutaneous malignant, 9}, 615134 |
| TINF2 | 165 | 100% | 100% | Dyskeratosis congenita, autosomal dominant 3, 613990<br>Revesz syndrome, 268130 |
| TMEM127 | 67,4 | 100% | 94% | {Pheochromocytoma, susceptibility to}, 171300 |
| TNFRSF11A | 97,2 | 95% | 92% | Osteolysis, familial expansile, 174810<br>Paget disease of bone, 602080<br>Osteopetrosis, autosomal recessive 7, 612301 |
| TP53 | 79,3 | 100% | 100% | Colorectal cancer, 114500<br>Li-Fraumeni syndrome, 151623<br>Hepatocellular carcinoma, 114550<br>Osteosarcoma, 259500<br>Choroid plexus papilloma, 260500<br>Nasopharyngeal carcinoma, 607107<br>Pancreatic cancer, 260350<br>Adrenal cortical carcinoma, 202300<br>Breast cancer, 114480<br>{Basal cell carcinoma 7}, 614740<br>{Glioma susceptibility 1}, 137800 |
| TSC1 | 85,1 | 99% | 96% | Tuberous sclerosis-1, 191100<br>Lymphangioleiomyomatosis, 606690<br>Focal cortical dysplasia, Taylor balloon cell type, 607341 |

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| TSC2 | 81,7 | 99% | 95% | Tuberous sclerosis-2, 613254<br>Lymphangioleiomyomatosis, somatic, 606690 |
| VHL | 115,6 | 100% | 100% | von Hippel-Lindau syndrome, 193300<br>Renal cell carcinoma, somatic, 144700<br>Pheochromocytoma, 171300<br>Hemangioblastoma, cerebellar, somatic<br>Erythrocytosis, familial, 2, 263400 |
| WAS | 30,2 | 91% | 72% | Wiskott-Aldrich syndrome, 301000<br>Thrombocytopenia, X-linked, 313900<br>Neutropenia, severe congenital, X-linked, 300299<br>Thrombocytopenia, X-linked, intermittent, 313900 |
| WRAP53 | 131,3 | 100% | 98% | Dyskeratosis congenita, autosomal recessive 3, 613988 |
| WRN | 138,5 | 100% | 100% | Werner syndrome, 277700 |
| WT1 | 53,1 | 100% | 95% | Wilms tumor, type 1, 194070<br>Denys-Drash syndrome, 194080<br>Nephrotic syndrome, type 4, 256370<br>Frasier syndrome, 136680<br>Meacham syndrome, 608978<br>Mesothelioma, somatic, 156240 |
| XPA | 81,5 | 100% | 90% | Xeroderma pigmentosum, group A, 278700 |
| XPC | 114 | 100% | 98% | Xeroderma pigmentosum, group C, 278720 |
| XRCC2 | 182 | 100% | 100% | No OMIM phenotype<br>Breast cancer (Park (2012) Am J Hum Genet 90, 734) |

*Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013*
*Median Coverage describes the average number of reads seen across 50 exomes*
*% Covered 10x describes the percentage of a gene's coding region that is covered at least 10x*
*% Covered 20x describes the percentage of a gene's coding region that is covered at least 20x*
*OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013*
*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*