

Tabella 1: OncoScreening® - Elenco dei geni analizzati e delle patologie neoplastiche investigate

	DISEASE NAME	PhenoMIM	GENE
1	Pituitary adenoma, ACTH-secreting	219090	AIP
	Pituitary adenoma, growth hormone-secreting	102200	
	Pituitary adenoma, prolactin-secreting	600634	
2	Neuroblastoma, susceptibility to, 3	613014	ALK
3	Adenomatous polyposis coli (Colorectal, duodenal, liver, thyroid, pancreatic, CNS)	175100	APC
4	Breast cancer, susceptibility to (Breast, pancreatic)	114480	ATM
5	Tumor predisposition syndrome	614327	BAP1
6	Breast cancer, susceptibility to (Breast, ovarian)	114480	BARD1
7	Polyposis syndrome (Colorectal, stomach, pancreatic)	174900 610069	BMPRIA
8	Breast-ovarian cancer, familial, 1	604370	BRCA1
	Pancreatic cancer, susceptibility to, 4 (Breast, ovarian, male breast, prostate, pancreatic)	614320	
9	Breast-ovarian cancer, familial, 2	612555	BRCA2
	Pancreatic cancer	613347	
	Prostate cancer (Breast, ovarian, male breast, prostate, pancreatic)	176807	
10	Breast cancer, early-onset (Breast, ovarian)	114480	BRIPI
11	Colorectal cancer, somatic	114500	BUB1B
12	Parathyroid carcinoma	608266	CDC73
13	Endometrial carcinoma, somatic	608089	CDH1
	Gastric cancer, familial diffuse, with or without cleft lip and/or palate	137215	
	Ovarian carcinoma, somatic	167000	
	Breast cancer, lobular	114480	
	Prostate cancer, susceptibility to (Breast, gastric, colorectal)	176807	
14	Melanoma, cutaneous malignant, 3	609048	CDK4
15	Melanoma and neural system tumor syndrome	155755	CDKN2A
	Pancreatic cancer/melanoma syndrome	606719	
	Melanoma, cutaneous malignant, 2 Melanoma, pancreatic	155601	
16	Leukemia, acute myeloid	601626	CEBPA
17	Li-Fraumeni syndrome	609265	CHEK2
	Breast cancer, susceptibility to	114480	
	Prostate cancer, familial, susceptibility to	176807	
	Osteosarcoma, somatic Breast, colorectal	259500	
18	Pleuropulmonary blastoma	601200	DICER1
	Rhabdomyosarcoma, embryonal, 2	180295	
19	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in	211980	EGFR

	Nonsmall cell lung cancer, susceptibility to		
20	Colorectal cancer, hereditary nonpolyposis, type 8 Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	613244	EPCAM
21	Chondrosarcoma Exostoses, multiple, type 1	215300 133700	EXT1
22	Exostoses, multiple, type 2	133701	EXT2
23	Leiomyomatosis and renal cell cancer Kidney, leiomyomas	150800	FH
24	Colorectal cancer, somatic Renal carcinoma, chromophobe, somatic Kidney	114500 144700	FLCN
25	Leukemia, acute myeloid, susceptibility to Myelodysplastic syndrome, susceptibility to	601626 614286	GATA2
26	Wilms tumor, somatic	194070	GPC3
27	Hepatic adenoma, somatic Renal cell carcinoma	142330 144700	HNF1A
28	Bladder cancer, somatic Thyroid carcinoma, follicular, somatic	109800 188470	HRAS
29	Gastrointestinal stromal tumor, familial Germ cell tumors Leukemia, acute myeloid	606764 273300 601626	KIT
30	Pheochromocytoma, susceptibility to	171300	MAX
31	Multiple endocrine neoplasia 1	131100	MEN1
32	Hepatocellular carcinoma, childhood type Renal cell carcinoma, papillary, 1, familial and somatic Kidney	114550 605074	MET
33	Melanoma, cutaneous malignant, susceptibility to, 8 Kidney	614456	MITF
34	Colorectal cancer, hereditary nonpolyposis, type 2 Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	609310	MLH1
35	Breast, ovarian		MRE11A
36	Colorectal cancer, hereditary nonpolyposis, type 1 Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	120435	MSH2
37	Colorectal cancer, hereditary nonpolyposis, type 5 Endometrial cancer, familial Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	614350 608089	MSH6
38	Adenomas, multiple colorectal Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas Gastric cancer, somatic Breast, colorectal	608456 132600 613659	MUTYH
39	Leukemia, acute lymphoblastic breast cancer prostate cancer Breast, ovarian	613065	NBN
40	Neurofibromatosis, type 1 Paranglioma / Pheochromocytoma, neurofibromas, Gastrointestinal stromal	162200	NF1

	tumor, breast, CNS, optic glioma		
41	Neurofibromatosis, type 2	101000	NF2
42	Leukemia, acute myeloid	601626	NSD1
43	Breast cancer, susceptibility to Pancreatic cancer, susceptibility to, 3 Breast, pancreatic	114480 613348	PALB2
44	Neuroblastoma, susceptibility to, 2	613013	PHOX2B
45	Mismatch repair cancer syndrome	276300	PMS1
46	Colorectal cancer, hereditary nonpolyposis, type 4 Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	614337	PMS2
47	Lymphoma, non-Hodgkin	605027	PRF1
48	Thyroid carcinoma, papillary, somatic	188550	PRKARIA
49	Basal cell carcinoma, somatic	605462	PTCH1
50	Endometrial carcinoma, somatic Malignant melanoma, somatic Squamous cell carcinoma, head and neck, somatic Thyroid carcinoma, follicular, somatic Glioma susceptibility 2 Meningioma Prostate cancer, somatic Breast, uterine, kidney, thyroid, colorectal	608089 155600 275355 188470 613028 607174 176807	PTEN
51	Breast, ovarian		RAD50
52	Breast-ovarian cancer, familial, susceptibility to, 3 Breast, ovarian	613399	RAD51C
53	Breast-ovarian cancer, familial, susceptibility to, 4 Breast, ovarian	614291	RAD51D
54	Retinoblastoma Bladder cancer, somatic Osteosarcoma, somatic Small cell cancer of the lung, somatic	180200 109800 259500 182280	RBI
55	Medullary thyroid carcinoma Multiple endocrine neoplasia IIA Multiple endocrine neoplasia IIB Pheochromocytoma Hirschsprung disease, susceptibility to, 1	155240 171400 162300 171300 142623	RET
56	Tylosis with esophageal cancer	148500	RHBDF2
57	Leukemia, acute myeloid	601626	RUNX1
58	Leukemia, acute myeloid	601626	SBDS
59	Parangliomas 2	601650	SDHAF2
60	Parangliomas 4 Gastrointestinal stromal tumor Paranglioma and gastric stromal sarcoma	115310 606764 606864	SDHB

	Pheochromocytoma	171300	
61	Parangliomas 3	605373	SDHC
	Gastrointestinal stromal tumor	606764	
	Paranglioma and gastric stromal sarcoma	606864	
62	Carcinoid tumors, intestinal	114900	SDHD
	Paranglioma and gastric stromal sarcoma	606864	
	Parangliomas 1, with or without deafness	168000	
	Pheochromocytoma	171300	
63	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	175050	SMAD4
	Pancreatic cancer, somatic	260350	
	Polyposis, juvenile intestinal	174900	
64	Rhabdoid predisposition syndrome 1	609322	SMARCB1
	Schwannomatosis-1, susceptibility to	162091	
65	Peutz-Jeghers syndrome	175200	STK11
	Pancreatic cancer	260350	
	Testicular tumor, somatic	273300	
	Small bowel, colorectal, pancreatic, breast, ovarian		
66	Meningioma, familial, susceptibility to	607174	SUFU
	Medulloblastoma, desmoplastic	155255	
67	Pheochromocytoma, susceptibility to	171300	TMEM127
68	Adrenal cortical carcinoma	202300	TP53
	Breast cancer	114480	
	Colorectal cancer	114500	
	Hepatocellular carcinoma	114550	
	Li-Fraumeni syndrome	151623	
	Nasopharyngeal carcinoma	607107	
	Osteosarcoma	259500	
	Pancreatic cancer	260350	
	Basal cell carcinoma 7	614740	
	Glioma susceptibility 1	137800	
Breast, sarcoma, brain, adrenocortical, leukemia, gastrointestinal, genitourinary			
69	Tuberous sclerosis-1	191100	TSC1
70	Tuberous sclerosis-2	613254	TSC2
71	von Hippel-Lindau syndrome	193300	VHL
	Erythrocytosis, familial, 2	263400	
	Pheochromocytoma	171300	
	Renal cell carcinoma, somatic	144700	
72	Wilms tumor, type 1	194070	WT1
	Mesothelioma, somatic	156240	