

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

	DISEASE NAME	PhenoMIM	GENE
1	Breast cancer, susceptibility to (Breast, pancreatic)	114480	<i>ATM</i>
2	Breast cancer, susceptibility to (Breast, ovarian)	114480	<i>BARD1</i>
3	Breast-ovarian cancer, familial, 1 Pancreatic cancer, susceptibility to, 4 (Breast, ovarian, male breast, prostate, pancreatic)	604370 614320	<i>BRCA1</i>
4	Breast-ovarian cancer, familial, 2 Pancreatic cancer Prostate cancer (Breast, ovarian, male breast, prostate, pancreatic)	612555 613347 176807	<i>BRCA2</i>
5	Breast cancer, early-onset (Breast, ovarian)	114480	<i>BRIP1</i>
6	Endometrial carcinoma, somatic Gastric cancer, familial diffuse, with or without cleft lip and/or palate Ovarian carcinoma, somatic Breast cancer, lobular Prostate cancer, susceptibility to (Breast, gastric, colorectal)	608089 137215 167000 114480 176807	<i>CDH1</i>
7	Li-Fraumeni syndrome Breast cancer, susceptibility to Prostate cancer, familial, susceptibility to Osteosarcoma, somatic Breast, colorectal	609265 114480 176807 259500	<i>CHEK2</i>
8	Breast, ovarian		<i>MRE11A</i>
9	Leukemia, acute lymphoblastic breast cancer prostate cancer Breast, ovarian	613065	<i>NBN</i>
10	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	<i>PTEN</i>
11	Breast-ovarian cancer, familial, susceptibility to, 3 Breast, ovarian	613399	<i>RAD51C</i>
12	Adrenal cortical carcinoma Breast cancer Colorectal cancer Hepatocellular carcinoma Li-Fraumeni syndrome Nasopharyngeal carcinoma Osteosarcoma Pancreatic cancer Basal cell carcinoma 7 Glioma susceptibility 1 Breast, sarcoma, brain, adrenocortical, leukemia, gastrointestinal, genitourinary	202300 114480 114500 114550 151623 607107 259500 260350 614740 137800	<i>TP53</i>