

Table 1: BreastScreen®

	DISEASE NAME	PhenoMIM	GENE
1	Breast cancer, susceptibility to (Breast, pancreatic)	114480	ATM
2	Breast cancer, susceptibility to (Breast, ovarian)	114480	BARD1
3	Breast-ovarian cancer, familial, 1 Pancreatic cancer, susceptibility to, 4 (Breast, ovarian, male breast, prostate, pancreatic)	604370 614320	BRCA1
4	Breast-ovarian cancer, familial, 2 Pancreatic cancer Prostate cancer (Breast, ovarian, male breast, prostate, pancreatic)	612555 613347 176807	BRCA2
5	Breast cancer, early-onset (Breast, ovarian)	114480	BRIP1
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	CDH1
7	Li-Fraumeni syndrome Breast cancer, susceptibility to Prostate cancer, familial, susceptibility to Osteosarcoma, somatic Breast, colorectal	609265 114480 176807 259500	CHEK2
8	Breast, ovarian		MRE11A
9	Leukemia, acute lymphoblastic breast cancer prostate cancer Breast, ovarian	613065	NBN
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	PTEN
11	Breast cancer, susceptibility to Pancreatic cancer, susceptibility to	114480 613348	PALB2
12	Breast-ovarian cancer, familial, susceptibility to, 3 Breast, ovarian	613399	RAD51C
13	Adrenal cortical carcinoma Breast cancer Colorectal cancer	202300 114480 114500	TP53

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		