

Table 1: ColonScreen® - List of analyzed genes and neoplastic diseases investigated

	DISEASE NAME	PhenoMIM	GENE
1	Adenomatous polyposis coli (Colorectal, duodenal, liver, thyroid, pancreatic, CNS)	175100	<i>APC</i>
2	Polyposis syndrome (Colorectal, stomach, pancreatic)	174900 610069	<i>BMPRIA</i>
3	Colorectal cancer, hereditary nonpolyposis, type 8 Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	613244	<i>EPCAM</i>
4	Colorectal cancer, hereditary nonpolyposis, type 2 Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	609310	<i>MLH1</i>
5	Colorectal cancer, hereditary nonpolyposis, type 1 Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	120435	<i>MSH2</i>
6	Colorectal cancer, hereditary nonpolyposis, type 5 Endometrial cancer, familial Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	614350 608089	<i>MSH6</i>
7	Adenomas, multiple colorectal Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas Gastric cancer, somatic Breast, colorectal	608456 132600 613659	<i>MUTYH</i>
8	Mismatch repair cancer syndrome	276300	<i>PMS1</i>
9	Colorectal cancer, hereditary nonpolyposis, type 4 Colorectal, uterine, stomach, ovarian, small bowel, hepatobiliary, urinary tract, brain, pancreatic, sebaceous	614337	<i>PMS2</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	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome Pancreatic cancer, somatic Polyposis, juvenile intestinal	175050 260350 174900	<i>SMAD4</i>
12	Peutz-Jeghers syndrome Pancreatic cancer Testicular tumor, somatic Small bowel, colorectal, pancreatic, breast, ovarian	175200 260350 273300	<i>STK11</i>