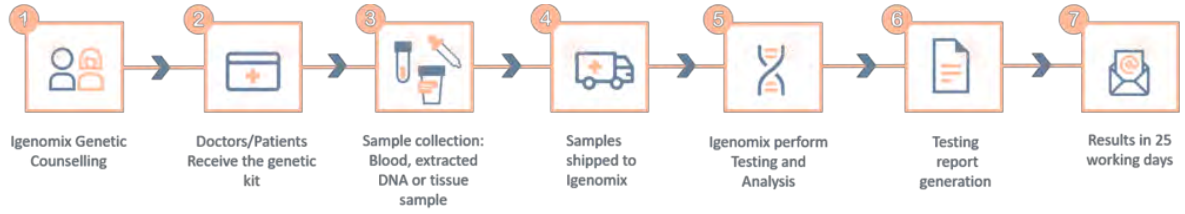
EPCAM		Hereditary Nonpolyposis Colorectal Cancer Type 8, Congenital Diarrhea With Tufting Enteropathy, Lynch Syndrome	AR	99.94%	52 of 70
FAM175A	Moderate risk	Ovarian Cancer, Breast Cancer, Fanconi Anemia Complementation Group A	-	94.81%	NA of NA
FANCC		Fanconi Anemia Complementation Group C	AR	100%	75 of 75
FANCM		Fanconi Anemia, Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation, Premature Ovarian Failure; Spermatogenic Failure	AR	99.73%	59 of 61
GEN1		Xeroderma Pigmentosum Complementation Group G	-	99.71%	6 of 6
MEN1		Familial Isolated Hyperparathyroidism, Insulinoma, Multiple Endocrine Neoplasia Type 1, Pituitary Gigantism, Prolactinoma	AD	99.90%	871 of 876
MLH1		Hereditary Nonpolyposis Colorectal Cancer Type 2, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.94%	1079 of 1118
MRE11	Moderate risk	Ataxia-Telangiectasia-Like Disorder, Hereditary Breast And Ovarian Cancer Syndrome	AR	99.95%	NA of NA
MSH2		Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.99%	1032 of 1057
MSH6		Hereditary Nonpolyposis Colorectal Cancer Type 5, Endometrial Carcinoma, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.28%	613 of 641
MUTYH		Familial Adenomatous Polyposis, Gastric Cancer, MUTYH-Related Attenuated Familial Adenomatous Polyposis	AR	100%	183 of 183
NBN	Moderate risk	Aplastic Anemia, Hereditary Breast And Ovarian Cancer Syndrome, Acute Lymphocytic Leukemia, Nijmegen Breakage Syndrome	AR,MU,P	100%	200 of 200
NF1		17q11.2 Microduplication Syndrome, Hereditary Pheochromocytoma-Paraganglioma, Juvenile Myelomonocytic Leukemia, Neurofibromatosis Type 1, Neurofibromatosis-Noonan Syndrome, Familial Spinal Neurofibromatosis Type I, Watson Syndrome	AD	97.97%	3082 of 3166
PALB2		Breast Cancer, Familial Pancreatic Carcinoma, Fanconi Anemia Complementation Group N, Hereditary Breast And Ovarian Cancer Syndrome	AD,AR	98.78%	601 of 617
PIK3CA		Breast Cancer, Capillary Malformation Of The Lower Lip, Lymphatic Malformation Of Face And Neck, Asymmetry Of Face And Limbs And Partial/Generalized Overgrowth, Colorectal Cancer, Congenital Lipomatous Overgrowth, Vascular Malformations And Epidermal Nevi, Cowden Syndrome, Gastric Cancer, Hemihyperplasia-Multiple Lipomatosis Syndrome, Hepatocellular Carcinoma, Seborrheic Keratosis, Lung Cancer, Lynch Syndrome, Macrocephaly-Capillary Malformation, Meningioma	AD	99.58%	54 of 58
PMS2		Hereditary Nonpolyposis Colorectal Cancer Type 4, Lynch Syndrome, Mismatch Repair Cancer Syndrome	AD,AR	97.17%	264 of 285
PTEN	High risk	Bannayan-Riley-Ruvalcaba Syndrome, Cowden Disease, Hereditary Breast And Ovarian Cancer Syndrome, Juvenile Polyposis Of Infancy, Lhermitte-Duclos Disease, Macrocephaly/Autism Syndrome, Familial Meningioma, Prostate Cancer, Proteus Syndrome, Proteus-Like Syndrome, Segmental Outgrowth-Lipomatosis-Arteriovenous Malformation-Epidermal Nevus Syndrome	AD	99.97%	609 of 629
RAD50	Moderate risk	Hereditary Breast And Ovarian Cancer Syndrome, Nijmegen Breakage Syndrome-like Disorder	AR	99.94%	117 of 120
RAD51C	Moderate risk	Familial Breast-Ovarian Cancer, Fanconi Anemia Complementation Group O, Hereditary Breast And Ovarian Cancer Syndrome	AR	100%	130 of 130
RAD51D	Moderate risk	Hereditary Breast And Ovarian Cancer Syndrome	-	100%	97 of 97
RECQL		Inherited Cancer-Predisposing Syndrome	-	99.71%	32 of 34
RINT1		Infantile Liver Failure Syndrome	AR	99.96%	16 of 16
STK11	High risk	Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular tumor	AD	81.99%	456 of 470
TP53	High risk	Adrenocortical Carcinoma, Basal Cell Carcinoma, Bone Marrow Failure Syndrome, Breast Cancer, Colorectal Cancer, Essential Thrombocythemia, Familial Pancreatic Carcinoma, Glioma, Hepatocellular Carcinoma, Hereditary Breast And Ovarian Cancer Syndrome, Li-Fraumeni Syndrome, Nasopharyngeal Carcinoma, Osteosarcoma, Pancreatic Cancer, Papilloma Of Choroid Plexus	AD,MU,P	98.92%	557 of 563
XRCC2		Fanconi Anemia Complementation Group U, Male Infertility With Azoospermia Or Oligozoospermia Due To Single Gene Mutation	AR	98.39%	28 of 28

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



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- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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