



ACMG Actionable Diseases

Precision Panel



Overview

The American College of Medical Genetics and Genomics (ACMG) Actionable Diseases Gene Panel shows a comprehensive selection of genes by ACMG. Mutations in these genes lead to disorders that have been scientifically proven to be actionable, meaning that early intervention improves prognosis, life expectancy, quality of life and overall well-being. Early identification before the onset of manifestations allows the prevention of symptoms thus initiating prompt multidisciplinary treatment. The main goal is to report the known or expected pathogenic variants in these genes while performing exome and genome sequencing, even if those are unrelated to the primary medical reason for testing.

The Igenomix ACMG Actionable Disorders Precision Panel can be used to identify those genes and perform a screening, whether the patient shows symptoms or not, to locate mutations and start an early treatment.

Indications

The Igenomix ACMG Actionable Disorders Precision Panel is indicated as a screening and diagnostic test in those cases where there are:

- Family history of cancer or cardiomyopathy
- Multiple relatives on the same side of the family with any form of cancer or cardiomyopathy.
- Asymptomatic patients who wish to check the chance of developing any of the reported diseases.





Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of multidisciplinary treatment including lifestyle modifications, early surveillance from malignancy, regular follow up with a specialist, and medical or surgical care if needed.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Family planning for adequate reproductive decisions, using available assisted reproduction technology.

Genes & Diseases

| GENE | OMIM DISEASES | INHERITANCE* | % GENE COVERAGE (20X) | HGMD** |
|---------|---|--------------|--------------------------|--------------|
| ACTA2 | Aortic Aneurysm, Moyamoya Disease, Multisystemic Smooth Muscle Dysfunction Syndrome | AD | 100 | 88 of 88 |
| ACTC1 | Atrial Septal Defect, Cardiomyopathy | AD | 99.93 | 72 of 74 |
| ΑΡϹ | Desmoid Disease, Familial Adenomatous Polyposis, Hepatocellular Carcinoma, Cenani-Lenz Syndrome, Gardner Syndrome, Turcot Syndrome, Colorectal Cancer, Gastric Cancer | AD | 98.92 | 1846 of 1882 |
| АРОВ | Hypercholesterolemia, Hypobetalipoproteinemia | AD,AR | 99.62 | 369 of 375 |
| ATP7B | Wilson Disease | AR | 99.97 | 989 of 1000 |
| BMPR1A | Polyposis Syndrome, Familial Colorectal Cancer, Hereditary Mixed Polyposis Syndrome | AD | 100 | 124 of 127 |
| BRCA1 | Breast And Ovarian Cancer, Fanconi Anemia, Familial Pancreatic Carcinoma, Peritoneal Carcinoma | AD,AR,MU | 98.97 | 2783 of 2894 |
| BRCA2 | Breast And Ovarian Cancer, Fanconi Anemia, Glioma, Medulloblastoma, Pancreatic Cancer, Prostate Cancer, Wilms Tumor, Nephroblastoma | AD,AR,MU | 98.51 | 3343 of 3451 |
| CACNA1S | Hypokalemic Periodic Paralysis, Malignant Hyperthermia, Thyrotoxic Periodic Paralysis | AD | 100 | 64 of 64 |
| COL3A1 | Ehlers-Danlos Syndrome, Polymicrogyria, Acrogeria, Cerebral Saccular Aneurysm | AD,AR | 100 | 676 of 676 |
| DSC2 | Arrhythmogenic Right Ventricular Dysplasia | AD,AR | 100 | 123 of 124 |
| DSG2 | Arrhythmogenic Right Ventricular Dysplasia, Cardiomyopathy | AD | 99.38 | 167 of 169 |
| DSP | Arrhythmogenic Right Ventricular Dysplasia, Cardiomyopathy, Epidermolysis Bullosa, Skin Fragility-Woolly Hair Syndrome, Carvajal Syndrome, Idiopathic Pulmonary Fibrosis, Palmoplantar Keratoderma | AD,AR | 99.91 | 366 of 369 |
| FBN1 | Acromicric Dysplasia, Ectopia Lentis, Geleophysic Dysplasia, Marfan Syndrome, Mass Syndrome, Stiff Skin Syndrome, Weill-Marchesani Syndrome, Familial Thoracic Aortic Aneurysm, Aortic Dissection, Glaucoma, Microspherophakia, Shprintzen-Goldberg Syndrome | AD | 100 | 2836 of 2845 |
| GLA | Fabry Disease | X,XR,G | 98 | - |
| KCNH2 | Long Qt Syndrome, Short Qt Syndrome, Romano-Ward Syndrome | AD | 98.69 | 908 of 930 |
| KCNQ1 | Atrial Fibrillation, Beckwith-Wiedemann Syndrome, Jervell And Lange- Nielsen Syndrome, Long Qt Syndrome, Short Qt Syndrome, Romano- Ward Syndrome | AD,AR | 93.23 | 600 of 624 |
| LDLR | Hypercholesterolemia | AD | 99.89 | 1921 of 1996 |
| LMNA | Cardiomyopathy, Charcot-Marie-Tooth Disease, Emery-Dreifuss Muscular Dystrophy, Heart-Hand Syndrome, Hutchinson-Gilford Progeria Syndrome, Lipodystrophy, Malouf Syndrome, Mandibuloacral Dysplasia, Werner Syndrome, Laminopathy, Hypergonadotropic Hypogonadism | AD,AR | 100 | 619 of 620 |





| MUH2 MUH2 Concert Lancer, Mismatch Repair Cancer Syndrome, Mult-Torre AD,ARAD,AR99.941079 of 1118MSH2 MSH2 Microme, Linch Syndrome, Mismatch Repair Cancer Syndrome, Mult-Torre Calerectal Cancer, Hareditary Nonpolyposis, Endometrial Carcinoma, Mult-Torre SyndromeAD,AR99.28613 of 641MM17Matematch Repair Cancer Syndrome, Mult-Torre SyndromeAD,AR99.251072 of 1079MM17Admonatious Polyposis, Esdatric CancerAR100163 of 138MYI2RC3 Carciomypathy, Left Ventricular NoncompactionAD,AR99.951072 of 1079MM112 MY12Aortic Aneuryan, Toracic Antic Aneuryan, Antic Dissection, ADAD10067 of 67MM12 Carciomypathy, Left Ventricular NoncompactionAD,AR99.951003 of 1054MV12 CarciomypathyAD,AR10066 of 68MV12 CarciomypathyAD,AR100350 of 302MV12 CarciomypathyNeurolitoronatosis, SchwannomatosisAD100306 of 307PK52 PK624Arrythythogen Storage Disease Of Heart, Wolff. AD,AR99.9861 of 61PK625 PK724Arrythythogen Storage Disease Of Heart, Wolff. AD,AR99.99.7c609 of 629PK745 PK746Noncer, Nismatch Repair Cancer Syndrome, Lynch AD,AR99.94941 of 995PK746 PK747Nopale Disease, Manayan-Biley-Kunakaba Syndrome, Bladder Cancer, Manayan-Biley-Kunakaba Syndrome, AD,ARAD90.97c609 of 629PK747 PK747Hirschaprung Disease, Natiope Disease, Natiope Singer Singe Disease, Natiope S | MEN1 | Endocrine Neoplasia, Hyperparathyroidism, Insulinoma, Pituitary Gigantism, Prolactinoma | AD | 99.9 | 871 of 876 |
|--|--------|---|----------|-------|--------------|
| MsHzLynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Colorectal Cancer, Hereditary Nopolyposs, Endometrial Carinoma, ADAR99.991032 of 1057MsH6Mismatch Repair Cancer Syndrome, Junch Syndrome, Muir-Torre SyndromeADAR99.28613 of 641MVT7MAdomatch Repair Cancer Syndrome, Muir-Torre | MLH1 | Colorectal Cancer, Mismatch Repair Cancer Syndrome, Muir-Torre | AD,AR | 99.94 | 1079 of 1118 |
| MHHColorectal Cancer, Hereditary Nonpolypois, Endomerial Carcinoma, Syndrome99.286.13 of 641MUTMHAdenomatous Polypois, Gastric CancerAR100613 of 641MUTRPGCardiomypoisty, Eth Ventricular NoncompactionAD,AR99.951072 of 1079MYHLMyrapstry, Thoracic Anti: Aneuryam, Actric Dissection, Myrapstry, Ebstein MalformationAD,AR99.951053 of 1054MYHZCardiomypoisty, Lett Ventricular NoncompactionAD,AR99.951053 of 1054MYHZCardiomypoisty, Distettinal Hypoperisalis SyndromeAD10067 of 67MYHZCardiomypoisty, Ebstein MalformationAD,AR100329 of 362OTCHypercholestrooleniaAD10039.90 of 362OTCHypercholestrooleniaAD100305 of 307PKR2Arridiomypathy, Elstein Malformatolis, SchwannomatosisAD100305 of 307PKR2Arrydingoma, Neurofiltoronatosis, SchwannomatosisAD100305 of 307PKR2Colorectal Cancer, Mismatch Repair Cancer Syndrome, LynchAD,AR97.17264 of 285PKR2Colorectal Cancer, Mismatch Repair Cancer Syndrome, LynchAD,AR99.91609 of 629PKR3Colorectal Cancer, Nemela Physics Storage Disage Structure StorageAD100453 of 454PKR402Cardemypathy, Elstein Misher Spectrum Disease, Meningiona, Prostate Cancer, Janenie Physics Storage Disage Of Heart, Wolff- Parkinsin White Syndrome, Meningiona, Prostate Cancer, Janenie Physics Storage Disage Of Marcer, Janenie Physics Storage Disage | MSH2 | Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre | AD,AR | 99.99 | 1032 of 1057 |
| MYBPC3Cardiomyopathy, Left Ventricular NoncompactionAD.AR99.951072 of 1079MYH1Mortic Aneuryam, Thorack Antric Aneuryam, Antric Dissection, MYH7AD10067 of 67MYH7Myopathy, Ebstein MalformationAD.AR99.951053 of 1054MYH2CardiomyopathyAD10067 of 67MY12CardiomyopathyAD10042 of 42ATMeningioma, Ruerofibromatosis, SchwannematosisAD100359 of 58DR7HypercholesterolemiaAD10096 of 98PR82Arrhythmogenic Right Ventricular Dyslasia, Brugada SyndromeAD100366 of 307PM52Oractal Cancer, Mismatch Repair Cancer Syndrome, Lynch SyndromeAD, AR97.17264 of 285PRKAG2Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff- Parkinson-While Syndrome, Segmental Outgrowth, Luconce Disease, Proteus Syndrome, Segmental Outgrowth, Luconce Disease, Proteus Syndrome, Segmental Outgrowth, Luconce Disease, Proteus Syndrome, Segmental Outgrowth, Luconce Disease, Metholbastoma, Shall Cell Cancer Of Headhomed, Disease, Multip Endocrine Neolasia, Pheochromocytoma-Paraganglioma, Reala AgenesiaAD90.91941 of 995RB1Hirschgrung Disease, Multip Endocrine Neolasia, Pheochromocytoma-Paraganglioma, Renal AgenesiaAD90.0453 of 454RF7Pheochromocytoma-Paraganglioma, Renal AgenesiaAD90.0261 of 261SOHA2Paraganglioma, Pheothremocytoma, Covden Syndrome, Mypathy-Disters Of Muscle, Mallagnatt Hyperthermia, Minicore Mypathy-Deren Mythomagenic Right AgenesiaA | MSH6 | Colorectal Cancer, Hereditary Nonpolyposis, Endometrial Carcinoma, Mismatch Repair Cancer Syndrome, Lynch Syndrome , Muir-Torre | AD,AR | 99.28 | 613 of 641 |
| MYH11Aprite Aneuryam, Thoracic Aortic Aneuryam, Aortic Dissection, Megacysis, Microcolon, Intestinal Hypperistals SyndromeAD10067 of 67MYH7Myopathy, Ebstel MalformationAD,AR99.951053 of 1054MY12CardiomyopathyAD10067 of 67MY12CardiomyopathyAD,AR10042 of 42AV2Meningioma, Neurofibronatosis, SchwannomatosisAD10095 of 582PCSK9HypercholesterolemiaAD10096 of 98PRP2Arrhythmogenic Right Ventricular Dysplasia, Brugada SyndromeAD100306 of 307PMS2Colorectal Cancer, Niematch Repair Cancer Syndrome, LynchAD,AR97.17264 of 285SyndromeSyndromeAD99.9361 of 61PRKACardiomyopathy, Glycagen Storage Disease Of Heart, Wolff- Parkinson-White Syndrome, Segmental Outgrowth, Ustantasis, Archiveneus Malformation, EpidormaticAD99.97609 of 629PTENDocks Disease, Macrocephaly, Autism Spectrum Disease, | МИТҮН | Adenomatous Polyposis, Gastric Cancer | AR | 100 | 183 of 183 |
| MTH21Megacyatis, Microcolon, Intestinal Hypoperistalsis SyndromeADI.006.07 of 57MYH71Myopathy, Ebstein MalformationAD,AR9901053 of 1054MY12CardiomyopathyADAD10042 of 42M72CardiomyopathyAD,AR10042 of 42M72Meningioma, Neurofibromatosis, SchwannomatosisAD100956 of 98PCK89Hyperanmonemia, Ornithine Transcarbamyase DeficiencyX,R,G99.97-PCK89HypercholesterolemiaAD100936 of 98PRR2Arrhythmogenic Right Ventricular Dysplasia, Brugada SyndromeAD100936 of 98PRR2Colorectal Cancer, Mismatch Repair Cancer Syndrome, LynchAD,AR97.17264 of 285PRKACZCordiomyopathy, Giycagen Storage Disease Of Heart, Wolff- Parkinson-White Syndrome. Segmental Outgrowth, Duolos Disease, Macrocephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Jamagan-Riley-Roualcaba Syndrome, Duolos Disease, Macrocephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Jamagan-Riley-Roualcaba Syndrome, Duolos Disease, Macrocephaly, Autism Spectrum Disease, Meningioma Jistjät100453 of 454RE1Theochromocytoma - Sagmental Outgrowth, Lung, Monsoawa JistjätAD,AR97.63733 of 746RFR4Reochromocytoma - Sagmental Outgrowth, Lung, Monsoawa JistjätAD,AR96.788 of 64RFR4Prechromocytoma - Sagade Syndrome, Prechromocytoma - Sagade Syndrome, Syndrome, Progressive Familia Heart Block, Sick Sinus Syndrome, AD,ARMU99.41941 of 995 <td< td=""><td>МҮВРСЗ</td><td>Cardiomyopathy, Left Ventricular Noncompaction</td><td>AD,AR</td><td>99.95</td><td>1072 of 1079</td></td<> | МҮВРСЗ | Cardiomyopathy, Left Ventricular Noncompaction | AD,AR | 99.95 | 1072 of 1079 |
| MYL2CardiomyopathyAD10067 of 67MYL3CardiomyopathyAD,AR10042 of 42NF2Meningioma, Neurofibromatosis, SchwannomatosisAD100359 of 362OTCHyperammonemia, Ornithine Transcarbamylase DeficiencyX,R,G99.97-OTSHypercholesterolemiaAD100366 of 88PR2Arrhythmogenic Right Ventricular Dysplasia, Brugada SyndromeAD100366 of 88PR2Colorectal Cancer, Mismatch Repair Cancer Syndrome, LynchAD,AR97.17264 of 285PRKAGZCardiomyopathy, Glycogen Storage Disease Of Heart, Wolff- Romingioma, Prostera Cancer, SyndromeAD99.9861 of 61Colorectal Cancer, Mismatch Repair Cancer Syndrome, Loponabry, Glycogen Storage Disease, Ottoraga Riley-Rwindas Syndrome, Loponabry, Glycogen Storage Disease, Syndrome, SyndromeAD99.97609 of 629PTENBidder Cancer, Ostesarcoma, Retinoblastoma, Small Cell Cancer Of The Lung, Monsoany 13g14AD99.41941 of 995RFTPheochromocytoma, Provid Carcinoma, Haddad Syndrome, Pheochromocytoma, Paragangioma, Renal AgenesisAD100453 of 454RFR2Syndrome, Contral Carcel Syndrome, Cardiomyopathy, Long QLAD,AR97.63733 of 746Arrythmogenic Ring ads Syndrome, Carcel Syndrome, Carcel Syndrome, Syndr | MYH11 | | AD | 100 | 67 of 67 |
| MYL3CardiomyopathyAD,AR10042 of 42N/2Meningioma, Neurofibromatosis, SchwannomatosisAD100359 of 362OTCHyperanmonemia, Ornithine Transcarbamylase DeficiencyX,XR,G99.97PCS9HypercholesterolemiaAD10096 of 98PK92Arrhythmogenic Right Ventricular Dysplasia, Brugada SyndromeAD100306 of 307PM32Colorectal Cancer, Mismath Repair Cancer Syndrome, LynchAD,AR97.17264 of 283PRKAG2Colorectal Cancer, Mismatch Repair Cancer Syndrome, LynchAD,AR99.9361 of 61Colorectal Cancer, Wancroephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Banayan-Riley-Rivalcaba Syndrome, Brakmacn, Americaneur, Juveniano, Edgemania, Cancer, Juveniano, Segmental Outgrowth, Lipomatosis, Artenovenous Mafformation, Epiderma Neusu99.97609 of 629Ducks Disease, Proteus Syndrome, Segmental Outgrowth, Hirschsprung Disease, Multiple Endocrine Neoplasia, Pheochromocytoma, Thyroid Carcinoma, Renal AgencisAD100453 of 454RETPheochromocytoma, Thyroid Carcinoma, Haddad Syndrome, Pheochromocytoma, Thyroid Carcinoma, Cardiomyopathy, Long Qt Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, Dopathy, External DophalmogiejaAD99.4399.43941 of 995SUNASSudden Infanz Death Syndrome, Cardiomyopathy, Long Qt Yaria Fibrillation, Rrugada Syndrome, Cardiomyopathy, Long Qt Sudden Infanz Death Syndrome, Cardiomyopathy, Carcinate-Cong Reductase Deficiency Cong Reductase Deficiency Cong Reductase DeficiencyAD90.56620 of 63SDHAF2Pa | MYH7 | Myopathy, Ebstein Malformation | AD,AR | 99.95 | 1053 of 1054 |
| NF2Meningiona, Neurofibromatosis, SchwannomatosisAD100359 of 362OTCHypercholesterolemiaAD10096 of 98PR2Arrhythmogenic Right Ventricular Dysplasia, Brugada SyndromeAD100306 of 307PM52Colorectal Cancer, Mismatch Repair Cancer Syndrome, LynchAD,AR97.17264 of 285PRK462Parkinson-White SyndromeAD99.9861 of 61Colorectal Cancer, Mismatch Repair Cancer Syndrome, LynchAD99.9861 of 61Courden Disease, Macrocephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Banayan-Riley-Rvukalaba Syndrome, Breast And Ovarian Cancer, Juvenile Polyposis Of Infanzy, Linermitte- Lupomatosis, Arteriovenoux Malformation, Epidermal NevusAD99.91609 of 629R81Bidedr Cancer, Otseosarcoma, Retinobiastoma, Small Cell Cancer Of Myopathy, External OphthalmoplegiaAD99.41941 of 995RVR1Central Core Disease Of Muscle, Malginant Hyperthermia, Minicore Myopathy, External OphthalmoplegiaAD,AR97.63733 of 746RVR1Central Core Disease Of Muscle, Malginant Hyperthermia, Minicore Myopathy, External OphthalmoplegiaAD,AR99.45929 of 942ScNsASyndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Syndrome, CardiomyopathyAD,AR,MU99.45929 of 942ScNsASyndrome, Nerdical Stromal Tumor, Paraganglioma, Pheochromocytoma, Cardiamyopathy, Long QUAD,AR,MU99.9562 of 63ScNsASondrome, Nardone, Stromal Tumor, Paraganglioma, Pheochromocytoma, Cardiam Syndrome, Succinate- Cog Reductas | MYL2 | Cardiomyopathy | AD | 100 | 67 of 67 |
| OTCHyperammonemia, Ornithine Transcarbamylase DeficiencyX,XR,G99.97-PCSK9HypercholesterolemiaAD10096 of 98PKP2Arrhythmogenic Right Ventricular Dysplasia, Brugada SyndromeAD100306 of 307PMS2Colorectal Cancer, Mismatch Repair Cancer Syndrome, LynchAD,AR97.17264 of 285PRKAG2Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff- Cowden Disease, Macrocephaly, Autism Spectrum Disease, Menigoina, Protate Cancer, Sundamayan-Riley, Alvukabab Syndrome, Dudos Disease, Ateriovenous Malformation, Epidermal Nevus99.97609 of 629PTENBreast And Ovarian Cancer, Juvenile Polyposis Of Infancy, Ihermitte- Dudos Disease, Ateriovenous Malformation, Epidermal NevusAD99.41941 of 995RB1Bladder Cancer, Osteosarcoma, Retinoblastoma, Small Cell Cancer Of Pheochromocytoma-Paraganglioma, Renal AgenesisAD100453 of 454RVR1Carclinomy, Haddad Syndrome, Pheochromocytoma-Paraganglioma, Renal AgenesisAD99.2466 of 472SCNSASyndrome, Rugada Syndrome, Cardiomypathy, Long Qt Syndrome, Paraganglioma, Renal AgenesisAD99.45929 of 942SCNSASyndrome, PheochromocytomaGastorintestinal Stromal Tumor, Paraganglioma, PheochromocytomaAD99.9562 of 63SCNSASyndrome, Reast Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, PheochromocytomaAD99.9562 of 63SDHAFZParaganglioma, Pheochromocytoma, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Gastroi | MYL3 | Cardiomyopathy | AD,AR | 100 | 42 of 42 |
| PCSK9HypercholesterolemiaAD10096 of 98PKP2Arrhythmogenic Right Ventricular Dysplasia, Brugada SyndromeAD100306 of 307PMS2Colorectal Cancer, Mismatch Repair Cancer Syndrome, Lynch SyndromeAD,AR97.17264 of 285PRKAGZCardiomyopathy, Glycogen Storage Disease Of Heart, Wolff- Parkinson-White SyndromeAD99.9861 of 61Colorectal Cancer, Mismatch Repair Cancer, Sundrome, Lynch Meningioma, Prostate Cancer, Bannayan-Riley-Ruvalcaba Syndrome, Barest And Ovarian Cancer, Juennel Polyposis Of Infancy, Lhermitte- Duckos Disease, Proteus Syndrome, Segmental Outgrowth, Lipomatosis, Arteriovenous Malformation, Epidemmal NewusAD99.97609 of 629RB1Bladder Cancer, Osteoararoma, Retinoblastoma, Small Cell Cancer Of The Lung, Monosomy 13q14AD99.41941 of 995Hirschaprung Disease, Muitiple Endocrine Neoplasia, Pheachromocytoma, Thyroid Carcinoma, Haddad Syndrome, Central Core Disease Of Muscle, Malignant Hyperthermia, Minicore Mypathy, External OphthalmologiajaAD99.4397.63733 of 746RVR2Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, Dilated Carciomy, Progressve Familial Heart Block, Sick Sinus Syndrome, Syndrome, Progressve Familial Heart Block, Sick Sinus Syndrome, Syndrome, Progressve Familial Heart Block, Sick Sinus Syndrome, AD, RP 99.9390.945929 of 942ScNSASyndrome, PheochromocytomaAD90.02261 of 264Corney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cardiomy Syndrome, Surdrame, Paraganglioma, Pheochromocytoma, Cardiol Syndrome, Covden <td>NF2</td> <td>Meningioma, Neurofibromatosis, Schwannomatosis</td> <td>AD</td> <td>100</td> <td>359 of 362</td> | NF2 | Meningioma, Neurofibromatosis, Schwannomatosis | AD | 100 | 359 of 362 |
| PRP2 Arrhythmogenic Right Ventricular Dysplasia, Brugada SyndromeAD100306 of 307PMS2 Colorectal Cancer, Mismatch Repair Cancer Syndrome, LynchAD,AR97.17264 of 285PRKAG2 Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff- Parkinson-White SyndromeAD99.9861 of 61PRKMG2 Coviden Disease, Macrocephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Bannayan-Riley-Ruvalcaba Syndrome, Ducks Disease, Proteus Syndrome, Segmental Outgrowth, Lipomatasis, Arteriovenous Malformation, Epidermal NewsAD99.97609 of 629RB1Bladder Cancer, Joscoarcoma, Retinoblastoma, Small Cell Cancer Of The Lung, Monosomy 13414AD99.41941 of 995RF2 Pheochromocytoma, Thyroid Carcinoma, Haddad Syndrome, Lipomatosis, Arteriovenous Malforman, Renal AgenesisAD100453 of 554RVR1 Central Core Disease Of Muscle, Malignant Hyperthermia, Minicore Mypathy, Etrana DophtalmologiajaAD,AR97.63733 of 746RVR2 Syndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Syndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, AD,AR99.45929 of 942SDHAE2 Paraganglioma, Pheochromocytoma Cardiomyopathy, Long Qt Syndrome, Redistrost Distribuilistion, Romano- Ward Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cardiomyopathy, Long Qt Syndrome, Redistrost DeficiencyAD90.91261 of 264SDHAE2 Paraganglioma, Pheochromocytoma, Cardiomyopathy, Long Qt Carney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cardiom | отс | Hyperammonemia, Ornithine Transcarbamylase Deficiency | X,XR,G | 99.97 | - |
| PMS2Colorectal Cancer, Mismatch Repair Cancer Syndrome, Lynch SyndromeAD,AR97.17264 of 285PRKAG2Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff- Parkinson-White Syndrome Cowden Disease, Macrocephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Juvenile Polyposis Of Infancy, Lhermitte- ADAD99.9861 of 61PTENBreast And Ovarian Cancer, Juvenile Polyposis Of Infancy, Lhermitte- Lipomatosis, Arteriovenous Malformation, Epidermal NevusAD99.97609 of 629RB1Bladder Cancer, Osteosarcoma, Retinoblastoma, Small Cell Cancer Of The Lung, Monosomy 13q14AD99.41941 of 995RE7Pheochromocytoma, Thyroid Carcinoma, Haddad Syndrome, Pheochromocytoma, Thyroid Carcinoma, Haddad Syndrome, A DAD100453 of 454RYR1Central Core Disease, Multiple Endocrine Neoplasia, Pheochromocytoma, Thyroid Carcinoma, Haddad Syndrome, Syndrome, Carcinoma, Thyroid Carcinoma, Haddad Syndrome, And AD99.2466 of 472RYR2Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, Dilated Cardiomyopathy Atrial Fibrillation, Brugada Syndrome, Cardiomyopathy, Long Qt Syndrome, Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma AD, 99.9562 of 63SDHAF2Paraganglioma, Pheochromocytoma Paraganglioma, Pheochromocytoma, Cardiomy Syndrome, Succinate- Coq Reductase DeficiencyAD99.9562 of 63SDHAF2Paraganglioma, Pheochromocytoma Paraganglioma, Pheochromocytoma, Cardiomy Syndrome, Succinate- Coq Reductase DeficiencyAD99.9562 of 63SDHACarney-Stratakis Syndrome, Macrome, Phe | РСЅК9 | Hypercholesterolemia | AD | 100 | 96 of 98 |
| PMS2SyndromeAD/AR97.17Z64 07.28PRKAG2Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff- Parkinson-White SyndromeAD99.9861 of 61PTENCordiomyopathy, Glycogen Storage Disease Of Heart, Wolff- Parkinson-White SyndromeAD99.97609 of 629PTENBreast And Ovarian Cancer, Juvenile Polyposis Of Infancy, Lhermitte- Lujconstois, Arteriovenous Malformation, Epidermal NevusAD99.9199.91609 of 629R81Bladder Cancer, Osteosarcoma, Retinoblastoma, Small Cell Cancer Of Pheochromocytoma-Thyroid Carcinoma, Haddad Syndrome, Pheochromocytoma-Thyroid Carcinoma, Haddad Syndrome, Pheochromocytoma-Thyroid Carcinoma, Haddad Syndrome, ADAD100453 of 454Pheochromocytoma-Thyroid Carcinoma, Haddad Syndrome, Pheochromocytoma-Thyroid Carcinoma, Haddad Syndrome, Cartial Core Disease Of Muscle, Malignant Hyperthermia, Minicore Myopathy, External OphthalmoplegiaAD,AR97.63733 of 746RYR2Arrhythmogenic Right Ventricular Dysplasi, Ventricular Tachycardia, Sudden Infant Death Syndrome, Cardiomyopathy, Long Qt Syndrome, Progressive Familla Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Succinate- Qar Reductase DeficiencyAD99.9562 of 63SDHAR2Carney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Carcino Syndrome, Succinate- ADAD99.98164 of 166SDHAParaganglioma, PheochromocytomaAD90.56136 of 136SDHAParaganglioma, Pheochromocytoma, Carcino Syndrome, Paccetasia ADAD99.9562 of 63SDHAR2< | PKP2 | Arrhythmogenic Right Ventricular Dysplasia, Brugada Syndrome | AD | 100 | 306 of 307 |
| PRKAG2Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff- Parkinson-White SyndromeAD99.9861 of 61Cowden Disease, Macrocephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Bannayan-Riley-Ruvalcaba Syndrome, Duclos Disease, Proteus Syndrome, Segmental Outgrowth, Lipomatosis, Arteriovenous Malformation, Epidermal Nevus Bladder Cancer, Osteosarcoma, Retinoblastoma, Small Cell Cancer Of The Lung, Monscomy 13q14AD99.91941 of 995RETPreochromocytoma, Thyroid Carcinoma, Haddad Syndrome, AD100453 of 454Preochromocytoma-Paraganglioma, Renal AgenesisAD100453 of 454RYR1Central Core Disease Of Muscle, Malignant Hyperthermia, Minicore Myopathy, External OphthalmoplegiaAD, AR, MJ97.63733 of 746RYR2Arthythmogen Right Ventricular Displaia, Ventricular Tachycardia, Dilated Cardiomyopathy, Long QtAD99.22466 of 472SCNSASyndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Cardiomyopathy, Long QtAD96.788 of 8SDHAF2Paraganglioma, PheochromocytomaAD96.788 of 4SDHACarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cardiomyopathy, CurodenAD, AR, MU99.9562 of 63SDHACarney-Stratakis Syndrome, Ratroital Syndrome, Succinate- Cong Reductase Deficiency, Paraganglioma, Pheochromocytoma, Carden Syndrome, Toroaic AndAD100128 of 128SDHACarney-Stratakis Syndrome, Ratrocytoma, Carden Syndrome, Rouden Syndrome, Succinate- Cong Reductase Deficiency, Paraganglioma, Phe | PMS2 | | AD,AR | 97.17 | 264 of 285 |
| PTENCowden Disease, Macrocephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Bannayan-Riley-Kuvalcaba Syndrome, Berest And Ovarian Cancer, Unevnile Polyposis Of Infancy, Lhermitte- Duclos Disease, Proteus Syndrome, Segmental Outgrowth, Lipomatosis, Arteriovenous Malformation, Epidermal NevusAD99.97609 of 629RB1Bladder Cancer, Osteoarcoma, Retinoblastoma, Small Cell Cancer Of The Lung, Monosomy 13q14AD99.41941 of 995RETPheochromocytoma, Thyroid Carcinona, Haddad Syndrome, Pheochromocytoma, Thyroid Carcinona, Haddad Syndrome, Pheochromocytoma-Paragangiloma, Renal AgenesisAD100453 of 454RYR1Central Core Disease Of Muscle, Malignant Hyperthermia, Minicore Myopathy, External OphthalmoplegiaAD,AR97.63733 of 746RYR2Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, Sudden Infant Death Syndrome, Cardiomyopathy, Long Ot Sudden Infant Death Syndrome, Cardiomyopathy, Long Ot Ward SyndromeAD,AR,MU99.45929 of 942SDHAF2Paraganglioma, PheochromocytomaAD96.788 of 8Carney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cardinod Syndrome, Succinate- Cog Reductase Deficiency, Paraganglioma, Cowden Syndrome, PheochromocytomaAD99.9562 of 63SDHACarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Cowden Syndrome, Pacretaic Cancer, Thoracic Aortic Aneurysm, Aortic DissectionAD90.9562 of 63SDHAParaganglioma, Pheochromocytoma, Cardinod Syndrome, Cowden Syndrome, Succinate-Cog Reductase Deficiency, Paraganglioma, Pheochromocytoma, Cardina Co | PRKAG2 | Cardiomyopathy, Glycogen Storage Disease Of Heart, Wolff- | AD | 99.98 | 61 of 61 |
| RB1Biadder Cancer, Osteosarcoma, Retinoblastoma, Small Cell Cancer Of The Lung, Monosomy 13q14AD99.41941 of 995RETPheochromocytoma, Thyroid Carcinoma, Haddad Syndrome, Pheochromocytoma, Thyroid Carcinoma, Haddad Syndrome, Pheochromocytoma, Paraganglioma, Renal AgenesisAD100453 of 454RYR1Central Core Disease Of Muscle, Malignant Hyperthermia, Minicore Myopathy, External OphthalmoplegiaAD, AR97.63733 of 746RYR2Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, Dilated Cardiomyopathy Atrial Fibrillation, Brugada Syndrome, Cardiomyopathy, Long Ot Syndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Cardiomyopathy, Long Ot SyndromeAD, AR, MU99.45929 of 942SDHAF2Paraganglioma, PheochromocytomaAD96.788 of 8SDHAF2Carney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cowden Syndrome, Succinate- Coq Reductase DeficiencyAD100261 of 264SDH0Carney-Stratakis Syndrome, Mitochondrial Complex II Deficiency, Paraganglioma, Pheochromocytoma, Cardinal Syndrome, CowdenAD,AR99.98164 of 166SMAD3Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome, CowdenAD,AR99.56136 of 136SMAD3Loeys-Dietz Syndrome, Self-Healing Squarome, Paraganglioma, PheochromocytomaAD99.95136 of 136SDH0Paraganglioma, Pheochromocytoma, Carcinoid Syndrome, CowdenAD,AR99.98164 of 166Syndrome, Succinate-Coq Reductase DeficiencyAD100128 of 128 <td>PTEN</td> <td>Cowden Disease, Macrocephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Bannayan-Riley-Ruvalcaba Syndrome, Breast And Ovarian Cancer, Juvenile Polyposis Of Infancy, Lhermitte- Duclos Disease, Proteus Syndrome, Segmental Outgrowth,</td> <td>AD</td> <td>99.97</td> <td>609 of 629</td> | PTEN | Cowden Disease, Macrocephaly, Autism Spectrum Disease, Meningioma, Prostate Cancer, Bannayan-Riley-Ruvalcaba Syndrome, Breast And Ovarian Cancer, Juvenile Polyposis Of Infancy, Lhermitte- Duclos Disease, Proteus Syndrome, Segmental Outgrowth, | AD | 99.97 | 609 of 629 |
| RET Pheochromocytoma, Thyroid Carcinoma, Haddad Syndrome, Pheochromocytoma-Paraganglioma, Renal AgenesisAD100453 of 454RYR2Central Core Disease Of Muscle, Malignant Hyperthermia, Minicore Myopathy, External OphthalmoplegiaAD,AR97.63733 of 746RYR2Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, Dilated CardiomyopathyAD99.2466 of 472SCN5ASyndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Ventricular Fibrillation, Romano- Ward SyndromeAD,AR,MU99.45929 of 942SDHAF2Paraganglioma, PheochromocytomaAD96.78& of 8SDHAF2Paraganglioma, Pheochromocytoma, Cowden Syndrome, Succinate- Coq Reductase DeficiencyAD90.262 of 63SDHCCarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cardio Syndrome, Succinate- Coq Reductase DeficiencyAD99.9562 of 63SDHDParaganglioma, Pheochromocytoma, Cardinal Complex II Deficiency, | RB1 | Bladder Cancer, Osteosarcoma, Retinoblastoma, Small Cell Cancer Of The Lung, Monosomy 13q14 | AD | 99.41 | 941 of 995 |
| KYR1Myopathy, External OphthalmoplegiaAD97.63733 of 746RVR2Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, Dilated CardiomyopathyAD99.2466 of 472RVR2Arrial Fibrillation, Brugada Syndrome, Cardiomyopathy, Long Qt Syndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Ventricular Fibrillation, Romano- Ward SyndromeAD,AR,MU99.45929 of 942SCN5ASphAF2Paraganglioma, PheochromocytomaAD96.788 of 8SDHAF2Paraganglioma, PheochromocytomaAD90.783261 of 264Carney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cowden Syndrome, Succinate- Coq Reductase DeficiencyAD99.9562 of 63SDHDCarney-Stratakis Syndrome, Mitochondrial Complex Ii Deficiency, Paraganglioma, Pheochromocytoma, Carcinoid Syndrome, Cowden Syndrome, Succinate-Coq Reductase DeficiencyAD,AR99.98164 of 166SMAD3Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection, Hemorrhagic TelangiectasiaAD90.56136 of 136STK11Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular Tumor Aortic Aneurysm, Aortic Dissection, Hemorrhagic TelangiectasiaAD9496 of 100TGFBR1 Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic Aortic Aneurysm, Aortic Dissection, Lynch Syndrome, Andric Aneurysm, Aortic DissectionAD99.99165 of 166TMEM43Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss Muscular Arribysplasia, Emery-Dreifuss Muscul | RET | Pheochromocytoma, Thyroid Carcinoma, Haddad Syndrome, | AD | 100 | 453 of 454 |
| RYR2Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, Dilated CardiomyopathyAD99.2466 of 472SCN5AAtrial Fibrillation, Brugada Syndrome, Cardiomyopathy, Long Qt Syndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Ventricular Fibrillation, Romano- Ward SyndromeAD, AR, MU99.45929 of 942SDHAF2Paraganglioma, PheochromocytomaAD96.788 of 8SDHACarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cowden Syndrome, Succinate- Coq Reductase DeficiencyAD90.9562 of 63SDHCCarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Cowden Syndrome, PheochromocytomaAD99.9562 of 63SDHCCarney-Stratakis Syndrome, Mitochondrial Complex Ii Deficiency, Paraganglioma, Pheochromocytoma, Carcinoid Syndrome, Cowden Syndrome, Succinate-Coq Reductase DeficiencyAD90.9562 of 63SDHDCarney-Stratakis Syndrome, Aneurysm-Osteoarthritis Syndrome, Thoracic Aortic Aneurysm, Aortic DissectionAD100128 of 128SMAD3Loeys-Dietz Syndrome, Sudrome, Pancreatic Cancer, Thoracic Aortic Aneurysm, Aortic Dissection, Hemorrhagic Telangiectasia Arti Aneurysm, Aortic Dissection, Hemorrhagic Telangiectasia Arti Aneurysm, Aortic Dissection, Jennerhagic Telangiectasia Arti Aneurysm, Aortic Dissection, Jennerhagic Telangiectasia Arti Aneurysm, Aortic Dissection, Lynch Syndrome, Aortic Aneurysm, Aortic Dissection, Lynch Syndrome, Arti Aneurysm, Aortic Dissection, Lynch Syndrome, Arti Aneurysm, Aortic Dissection, Lynch Syndrome, Arti Arti Arti Arti Arti Aneurysm, Artic Dissection, Jennerh | RYR1 | | AD,AR | 97.63 | 733 of 746 |
| Atrial Fibrillation, Brugada Syndrome, Cardiomyopathy, Long Qt Syndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Ventricular Fibrillation, Romano- Ward SyndromeAD,AR,MU99.45929 of 942SDHAF2Paraganglioma, PheochromocytomaADAD96.788 of 8SDHBCarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cowden Syndrome, Succinate- Coq Reductase DeficiencyAD100261 of 264SDHCCarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Cowden Syndrome, PheochromocytomaAD99.9562 of 63SDHCCarney-Stratakis Syndrome, Mitochondrial Complex II Deficiency, Paraganglioma, Pheochromocytoma, Carcinoid Syndrome, Cowden Syndrome, Succinate-Coq Reductase DeficiencyAD99.9562 of 63SDHDParaganglioma, Pheochromocytoma, Carcinoid Syndrome, Floricic Aortic Aneurysm, Aortic DissectionAD99.98164 of 166SMAD3Loeys-Dietz Syndrome, Mitochondrial Complex II Deficiency, Polyposis Syndrome, Myhre Syndrome, Pancreatic Cancer, Thoracic Aortic Aneurysm, Aortic Dissection, Hemorrhagic Telangiectasia ATTAD99.56136 of 136STK11Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular Tumor Aortic Aneurysm, Aortic DissectionAD9496 of 100TGFBR1Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic Aortic Aneurysm, Aortic Dissection99.91165 of 166TMEM43Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss Muscular AD99.93165 of 166 | RYR2 | Arrhythmogenic Right Ventricular Dysplasia, Ventricular Tachycardia, | AD | 99.2 | 466 of 472 |
| SDHBCarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Pheochromocytoma, Cowden Syndrome, Succinate- Coq Reductase DeficiencyAD100261 of 264SDHCCarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Cowden Syndrome, PheochromocytomaAD99.9562 of 63SDHCCarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Cowden Syndrome, PheochromocytomaAD99.9562 of 63SDHDCarney-Stratakis Syndrome, Mitochondrial Complex Ii Deficiency, Paraganglioma, Pheochromocytoma, Carcinoid Syndrome, Cowden Syndrome, Succinate-Coq Reductase Deficiency Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome, Thoracic Aortic Aneurysm, Aortic DissectionAD100128 of 128SMAD3SMAD4Polyposis Syndrome, Myhre Syndrome, Pancreatic Cancer, Thoracic Aortic Aneurysm, Aortic Dissection, Hemorrhagic Telangiectasia Aortic Aneurysm, Aortic Dissection, Hemorrhagic Telangiectasia Aortic Aneurysm, Aortic Dissection81.99456 of 470TGFBR1Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic Aortic Aneurysm, Aortic Dissection, Lynch Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection, Lynch Syndrome, Anoric Aneurysm, Aortic Dissection, Lynch Syndrome, Anoric Aneurysm, Aortic Dissection, Lynch Syndrome, AndbAD99.98165 of 166TMEM43Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss MuscularAD99.9826 of 26 | SCN5A | Atrial Fibrillation, Brugada Syndrome, Cardiomyopathy, Long Qt Syndrome, Progressive Familial Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Ventricular Fibrillation, Romano- | AD,AR,MU | 99.45 | 929 of 942 |
| SDHBParaganglioma, Pheochromocytoma, Cowden Syndrome, Succinate- Coq Reductase DeficiencyAD100261 of 264SDHCCarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Cowden Syndrome, PheochromocytomaAD99.9562 of 63SDHCCarney-Stratakis Syndrome, Mitochondrial Complex Ii Deficiency, Paraganglioma, Pheochromocytoma, Carcinoid Syndrome, Cowden Syndrome, Succinate-Coq Reductase DeficiencyAD, AR99.98164 of 166SMAD3Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome, Thoracic Aortic Aneurysm, Aortic DissectionAD100128 of 128SMAD4Polyposis Syndrome, Myhre Syndrome, Pancreatic Cancer, Thoracic Aortic Aneurysm, Aortic Dissection, Hemorrhagic TelangiectasiaAD99.56136 of 136STK11Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular TumorAD81.99456 of 470TGFBR1Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic | SDHAF2 | Paraganglioma, Pheochromocytoma | AD | 96.78 | 8 of 8 |
| SDHCCarney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Cowden Syndrome, PheochromocytomaAD99.9562 of 63SDHDCarney-Stratakis Syndrome, Mitochondrial Complex Ii Deficiency, Paraganglioma, Pheochromocytoma, Carcinoid Syndrome, Cowden Syndrome, Succinate-Coq Reductase DeficiencyAD, AR99.98164 of 166SMAD3Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome, Thoracic Aortic Aneurysm, Aortic DissectionAD100128 of 128SMAD4Polyposis Syndrome, Myhre Syndrome, Pancreatic Cancer, Thoracic Aortic Aneurysm, Aortic Dissection, Hemorrhagic TelangiectasiaAD99.96136 of 136STK11Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular TumorAD81.99456 of 470TGFBR1Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic Aortic Aneurysm, Aortic DissectionAD99.99165 of 166TMEM43Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss MuscularAD99.9826 of 26 | SDHB | Paraganglioma, Pheochromocytoma, Cowden Syndrome, Succinate- | AD | 100 | 261 of 264 |
| SDHDParaganglioma, Pheochromocytoma, Carcinoid Syndrome, Cowden Syndrome, Succinate-Coq Reductase DeficiencyAD,AR99.98164 of 166SMAD3Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome, Thoracic Aortic Aneurysm, Aortic DissectionAD100128 of 128SMAD4Polyposis Syndrome, Myhre Syndrome, Pancreatic Cancer, Thoracic Aortic Aneurysm, Aortic Dissection, Hemorrhagic TelangiectasiaAD99.56136 of 136STK11Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular TumorAD81.99456 of 470TGFBR1Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic Aortic Aneurysm, Aortic DissectionAD9496 of 100TGFBR2Colorectal Cancer, Esophageal Cancer, Loeys-Dietz Syndrome Thoracic Aortic Aneurysm, Aortic Dissection, Lynch SyndromeAD99.9826 of 26TMEM43Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss Muscular AppAD99.9826 of 26 | SDHC | Carney-Stratakis Syndrome, Gastrointestinal Stromal Tumor, Paraganglioma, Cowden Syndrome, Pheochromocytoma | AD | 99.95 | 62 of 63 |
| SMAD3Aortic Aneurysm, Aortic DissectionAD100128 of 128SMAD4Polyposis Syndrome, Myhre Syndrome, Pancreatic Cancer, Thoracic Aortic Aneurysm, Aortic Dissection, Hemorrhagic TelangiectasiaAD99.56136 of 136STK11Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular TumorAD81.99456 of 470TGFBR1Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic Aortic Aneurysm, Aortic DissectionAD9496 of 100TGFBR2Colorectal Cancer, Esophageal Cancer, Loeys-Dietz Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection, Lynch SyndromeAD99.98165 of 166TMEM43Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss MuscularAD99.9826 of 26 | SDHD | Paraganglioma, Pheochromocytoma, Carcinoid Syndrome, Cowden | AD,AR | 99.98 | 164 of 166 |
| SMAD4Aortic Aneurysm, Aortic Dissection, Hemorrhagic TelangiectasiaAD99.56136 of 136STK11Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular TumorAD81.99456 of 470TGFBR1Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic Aortic Aneurysm, Aortic DissectionAD9496 of 100TGFBR2Colorectal Cancer, Esophageal Cancer, Loeys-Dietz Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection, Lynch SyndromeAD99.9165 of 166TMEM43Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss MuscularAD99.9826 of 26 | SMAD3 | Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome, Thoracic | AD | 100 | 128 of 128 |
| TGFBR1Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic Aortic Aneurysm, Aortic DissectionAD9496 of 100TGFBR2Colorectal Cancer, Esophageal Cancer, Loeys-Dietz Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection, Lynch SyndromeAD99.9165 of 166TMEM43Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss MuscularAD99.9826 of 26 | SMAD4 | | AD | 99.56 | 136 of 136 |
| I GFBR1AD9496 of 100Aortic Aneurysm, Aortic DissectionAD9496 of 100TGFBR2Colorectal Cancer, Esophageal Cancer, Loeys-Dietz Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection, Lynch SyndromeAD99.9165 of 166TMEM43Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss MuscularAD99.9826 of 26 | STK11 | Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular Tumor | AD | 81.99 | 456 of 470 |
| TIGFBR2 AD 99.9 165 of 166 Thoracic Aortic Aneurysm, Aortic Dissection, Lynch Syndrome AD 99.9 165 of 166 TMEM43 Arrhythmogenic Right Ventricular Dysplasia, Emery-Dreifuss Muscular AD 99.98 26 of 26 | TGFBR1 | Aortic Aneurysm, Aortic Dissection | AD | 94 | 96 of 100 |
| AD 99.98 26 0T 26 | TGFBR2 | | AD | 99.9 | 165 of 166 |
| | TMEM43 | | AD | 99.98 | 26 of 26 |

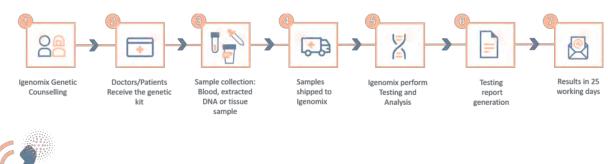




| TNNI3 | Cardiomyopathy | AD,AR | 100 | 139 of 139 |
|-------|---|---------|-------|--------------|
| TNNT2 | Cardiomyopathy | AD | 100 | 169 of 169 |
| TP53 | Adrenocortical Carcinoma, Osteosarcoma, Li-Fraumeni Syndrome, Basal Cell Carcinoma, Bone Marrow Failure Syndrome, Glioma Susceptibility, Papilloma Of Choroid Plexus, Thrombocythemia | AD,MU,P | 98.92 | 557 of 563 |
| TPM1 | Cardiomyopathy | AD | 100 | 108 of 108 |
| TSC1 | Focal Cortical Dysplasia Of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis | AD | 99.86 | 390 of 406 |
| TSC2 | Focal Cortical Dysplasia Of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis | AD | 100 | 1157 of 1159 |
| VHL | Erythrocytosis, Pheochromocytoma, Renal Cell Carcinoma, Von Hippel-Lindau Syndrome, Paraganglioma | AD,AR | 100 | 511 of 544 |
| WT1 | Aniridia, Denys-Drash Syndrome, Frasier Syndrome, Mesothelioma, Nephrotic Syndrome, Wilms Tumor, Gonadal Dysgenesis, Meacham Syndrome, Nephroblastoma, Wagr Syndrome | AD | 98.92 | 178 of 185 |

*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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