HEALTHSTRING™

report (annexure)

DESCRIPTION

HEALTH STRING is a preemptive genetic screening test that analyzes genes implicated in 205 life-limiting health conditions. This test utilizes next-generation DNA sequencing technology to detect variations/mutations in genes, aiding in identifying the underlying genetic causes of these health conditions and providing valuable health insights to the user.

The report enables physicians to develop specific actionable strategies for customers to prevent adverse health events, through lifestyle changes, medications, or any other suitable medical treatment. It also aids in early symptom identification and facilitates timely clinical interventions, unlocking the full potential of a healthy life.

METHODOLOGY

The protein-coding regions of genes within the DNA are sequenced to detect genetic variants or mutations. Proprietary AI/ML-based algorithms are employed to classify the genetic variants and identify genetic factors that may contribute to health conditions. With a 99% accuracy and reproducibility rate in detecting genetic variants, the results are reported based on the guidelines set forth by the American College of Medical Genetics and Genomics and the Association for Molecular Pathology (ACMG-AMP).

LIMITATION

Cancers and heart diseases are influenced by a combination of your inborn genetic factors and lifestyle choices. HEALTH STRING focuses solely on genetic factors known to contribute to these diseases and does not encompass risks attributed to lifestyle factors or any other unknown causes. Only variants located in the protein coding regions of genes are reported; intronic variants, repeat expansions, copy number variations, or chromosomal rearrangements are not assessed. However, it's important to note that due to limitations in DNA sequencing technology, certain genes may be inadequately covered, and variants may not be confidently detected, potentially impacting the test results.

DISCLAIMER

HEALTH STRING is developed and its performance validated by Precision Health Innovations Pvt Ltd (PHIPL). The processing of the sample and the genetic testing carried out by PHIPL are based on current scientific and analytical standards to the best of its knowledge. In rare cases, genetic tests may not yield the correct result due to the quality of the sample provided to PHIPL, or the test may fail due to unforeseeable and/or unknown reasons that cannot be influenced by PHIPL in advance. In such cases, PHIPL and/or any of its affiliates shall not be held responsible and/or liable.

HEREDITARY CANCERS (100)

Ataxia Telangetasia, Ataxia Telangetasia-like Disorder, Bannayan-Riley-Ruvalcaba Syndrome, Basal Cell Nevus Syndrome, Birt-Hogg-Dube Syndrome, Bloom Syndrome, Breast cancer, Carney Complex, Carney-Stratakis Syndrome, Chediak-Higashi Syndrome, Costello Syndrome, Cowden Syndrome, Denys-Drash Syndrome, Endometrial cancer, Familial Monocytic Leukemia, Familial Multiple Glomus Tumors, Familial Multiple Trichoepithelioma, Familial myeloproliferative/lymphoproliferative neoplasms, Familial Osteosarcoma, Familial Pancreatic Cancer, Familial Uveal Melanoma, Fanconi Anemia, Ferguson-Smith Syndrome, Gardner syndrome, Glioma, Hemochromatosis, Hereditary Acute Myeloid Leukemia, Hereditary Adenomatous Polyposis, Hereditary Adrenal Hyperplasia, Hereditary Adrenal Pheochromocytoma, Hereditary Adrenocortical Cancer, Hereditary Barrett Esophagus/Esophageal Adenocarcinoma, Hereditary Bone Dysplasia with Malignant Fibrous Histiocytoma, Hereditary Breast-Ovarian Cancer, Hereditary Colorectal Cancer, Hereditary Desmoid Disease, Hereditary Epidermodysplasia Verruciformis, Hereditary Gastric Carcinoma, Hereditary GIST, Hereditary Hodgkin Lymphoma, Hereditary Infantile Hemangioma, Hereditary Isolated Pituitary Adenoma, Hereditary Leiomyomatosis & Renal Cell Cancer, Hereditary Lung Cancer, Hereditary Malignant Melanoma, Hereditary Medulloblastoma, Hereditary Melanoma & Renal Cancer, Hereditary Neuroblastoma, Hereditary Paragangliomas, Hereditary Pleuropulmonary Blastoma, Hereditary Primary Pigmented Nodular Adrenocortical Disease, Hereditary Prostate Cancer, Hereditary Thyroid Cancer, Hereditary Wilms' Tumor, Howel-Evans syndrome, Hyperparathyroidism-Jaw Tumor Syndrome, Juvenile Hyaline Fibromatosis, Juvenile Polyposis Syndrome, Lhermitte-Duclos syndrome, Li-Fraumeni Syndrome, LIG4 Syndrome, Lymphangioleiomyomatosis, Lynch Syndrome, Melanoma and neural system tumor syndrome, Melanoma-pancreatic cancer syndrome, Mismatch Repair Cancer Syndrome, Mosaic Variegated Aneuploidy, Muir-Torre Syndrome, Multiple Endocrine Neoplasia, Multiple Exostoses, Myelodysplastic syndrome, Neurofibromatosis, Nijmegen Breakage Syndrome, Nijmegen Breakage-like Syndrome, Noonan syndrome-like disorder with or without juvenile myelomonocytic AD 24 43 leukemia, Oligodontia-Colorectal Cancer Syndrome, Opitz Trigonocephaly Syndrome, Oral Cancer, Pallister-Hall Syndrome, Palmoplantar Keratoderma & Squamous Cell Carcinoma, Perlman Syndrome, Peutz-Jeghers Syndrome, POLD1 & POLE Associated Colorectal Adenomas, Porphyria Cutanea Tarda, Proteus Syndrome, Retinoblastoma, Rhabdoid Predisposition Syndrome, Rothmund-Thompson Syndrome, SC Phocomelia Syndrome, Schwachman-Diamond Syndrome, Schwannomatosis, Seckel Syndrome 1, Simpson-Golabi-Behmel Syndrome, TERT Mutation-Associated Haematological Disorders, Tumor predisposition syndrome, Tyrosinemia, von Hippel-Lindau syndrome, Werner Syndrome, Wiskott-Aldrich Syndrome, Xeroderma Pigmentosum

By tissue and organs

Adrenal gland, Blood, Blood vessels, Bone, Bone marrow, Brain, Breast, Colon, Connective tissue, Endocrine system, Endometrium, Esophagus, Eye, Gastrointestinal tract, Sebum gland, Gums, Hypothalamus, Kidney, Liver, Lung, Lymphoid tissue, Mouth, Nerves, Ovary, Pancreas, Perichondrium (heart), Pituitary gland, Prostate, Rectum, Skin, Soft tissue, Stomach, Throat, Thyroid gland.

HEART CONDITIONS (105)

Dilated Cardiomyopathy 1a, Dilated Cardiomyopathy 1b, Dilated Cardiomyopathy 1c With Or Without Left Ventricular Noncompaction, Dilated Cardiomyopathy 1d, Dilated Cardiomyopathy 1e, Dilated Cardiomyopathy 1g, Dilated Cardiomyopathy 1i, Dilated Cardiomyopathy 1j, Dilated Cardiomyopathy 1l, Dilated Cardiomyopathy 1k, Dilated Cardiomyopathy 1m, Dilated Cardiomyopathy 1o, Dilated Cardiomyopathy 1p, Dilated Cardiomyopathy 1q, Dilated Cardiomyopathy 1r, Dilated Cardiomyopathy 1s, Dilated Cardiomyopathy 1u, Dilated Cardiomyopathy 1v, Dilated Cardiomyopathy 1w, Dilated Cardiomyopathy 1x, Dilated Cardiomyopathy 1y, Dilated Cardiomyopathy 1z, Dilated Cardiomyopathy 1aa With Or Without Left Ventricular Noncompaction, Dilated Cardiomyopathy 1bb, Dilated Cardiomyopathy 1cc, Dilated Cardiomyopathy 1dd, Dilated Cardiomyopathy 1ee, Dilated Cardiomyopathy 1ff, Dilated Cardiomyopathy 1gg, Dilated Cardiomyopathy 1hh, Dilated Cardiomyopathy 1ii, Dilated Cardiomyopathy 1jj, Dilated Cardiomyopathy 1kk, Dilated Cardiomyopathy 1ll, Dilated Cardiomyopathy 1mm, Dilated Cardiomyopathy 1nn, Dilated Cardiomyopathy 2a, Dilated Cardiomyopathy 2b, Dilated Cardiomyopathy 2c, Dilated Cardiomyopathy 2d, Dilated Cardiomyopathy 2e, Dilated Cardiomyopathy 2f, Dilated Cardiomyopathy 3b, Left Ventricular Noncompaction 1, Left Ventricular Noncompaction 2, Left Ventricular Noncompaction 3, Left Ventricular Noncompaction 4, Left Ventricular Noncompaction 5, Left Ventricular Noncompaction 6, Left Ventricular Noncompaction 7, Left Ventricular Noncompaction 8, Left Ventricular Noncompaction 9, Left Ventricular Noncompaction 10, Familial Restrictive Cardiomyopathy 1, Familial Restrictive Cardiomyopathy 2, Familial Restrictive Cardiomyopathy 3, Familial Restrictive Cardiomyopathy 4, Familial Restrictive Cardiomyopathy 5, Familial Restrictive Cardiomyopathy 6, Familial Arrhythmogenic Right Ventricular Dysplasia 1, Familial Arrhythmogenic Right Ventricular Dysplasia 2, Familial Arrhythmogenic Right Ventricular Dysplasia 3, Familial Arrhythmogenic Right Ventricular Dysplasia 4, Familial Arrhythmogenic Right Ventricular Dysplasia 5, Familial Arrhythmogenic Right Ventricular Dysplasia 6, Familial Arrhythmogenic Right Ventricular Dysplasia 8, Familial Arrhythmogenic Right Ventricular Dysplasia 9, Familial Arrhythmogenic Right Ventricular Dysplasia 10, Familial Arrhythmogenic Right Ventricular Dysplasia 11, Familial Arrhythmogenic Right Ventricular Dysplasia 12, Familial Arrhythmogenic Right Ventricular Dysplasia 13, Familial Arrhythmogenic Right Ventricular Dysplasia 14, Familial Hypertrophic Cardiomyopathy 1, Familial Hypertrophic Cardiomyopathy 2, Familial Hypertrophic Cardiomyopathy 3, Familial Hypertrophic Cardiomyopathy 4, Familial Hypertrophic Cardiomyopathy 6, Familial Hypertrophic Cardiomyopathy 7, Familial Hypertrophic Cardiomyopathy 8, Familial Hypertrophic Cardiomyopathy 9, Familial Hypertrophic Cardiomyopathy 10, Familial Hypertrophic Cardiomyopathy 11, Familial Hypertrophic Cardiomyopathy 12, Familial Hypertrophic Cardiomyopathy 13, Familial Hypertrophic Cardiomyopathy 14, Familial Hypertrophic Cardiomyopathy 15, Familial Hypertrophic Cardiomyopathy 16, Familial Hypertrophic Cardiomyopathy 17, Familial Hypertrophic Cardiomyopathy 18, Familial Hypertrophic Cardiomyopathy 20, Familial Hypertrophic Cardiomyopathy 21, Familial Hypertrophic Cardiomyopathy 22, Familial Hypertrophic Cardiomyopathy 23 with Or without Ventricular Noncompaction, Familial Hypertrophic Cardiomyopathy 24, Familial Hypertrophic Cardiomyopathy 25, Familial Hypertrophic Cardiomyopathy 26, Familial Hypertrophic Cardiomyopathy 27, Familial Hypertrophic Cardiomyopathy 28, Arrhythmogenic Right Ventricular Cardiomyopathy 1, Arrhythmogenic Right Ventricular Cardiomyopathy 2, Arrhythmogenic Right Ventricular Cardiomyopathy 3, Brugada syndrome, Long QT Syndrome, Short QT Syndrome, Polymorphic Ventricular Tachycardia.

917 GENES

(Hereditary Cancers and Heart Conditions)

AAGAB, ABCA5, AP… …C6, ABCC8, ABCD1, ABL… …ACVR1, ACVRL1,
ADA, ADA2, AD… …AIP, AKT1, ALAD, AL… …R1, ANTXR2,
AP2S1, AP3… …ARID1A, ARID1B, … …ASXL1,
ATM, AT… …KA, AXIN1, AXIN2, … …BCL10,
BC… …BMPR1B, BRAF, … …BUB1B,
… …ASP8, CASR, CAT, CE…
… …79A, CD79B, CD81, CD96, … …23, CDK4,
… …D7, CHEK2, CHIC2, CHRNG, CI… …LCNKB,
…A1, COL1A1, CO… …6, COL7A1, CPLANE1, CPLX1, CPOX, CR2, CREB1,
…CTC1, CTHRC1, … …S1, CTRC, CTSA, CTSC, CXCR4, CYLD, CYP11B1, CYP11B2,
…CC, DCLR… …DX41, DDX59, DEF6, DHCR24, DHCR7, DHH, DHX37,
…LST, … …DNASE1L3, DNM2, DNMT3A, DOCK8, DPF2, DPM1, DVL1,
… …EDNRB, EFL1, EGFR, EIF2AK4, EI… …MO2, ELP1, ENG,
EN… …3, ERCC2, ERCC3, ERCC4, ER… …CO2, ESR1,
ETV6, E… …B, F5, FAH, FAM149B1, FA… …B,
FANCC, FAN… …NCM, FAS, FASLG, FA…
…GF8, FGFR… …FLI1, FLNA, FLT3, …
…7D2, G6P… …ATA4, GBA, G…
…5, GINS1, … …GJC2, GLI…
…PC4, GP… …E2, GT…
… …DQB1, HL… …MR, … …PGL…
… …IDH2, IFIT… …140, IK… …KBKG, IKZF1,
… …7R, ING1, INPP5E, INS, IN… …IVNS1ABP, JAG
KA… …, KCNJ11, KCNN3, KCN… …AA0753, K
KIF1B, K… …KLLN, KRAS, KRIT1, KRT1, … …KRT
L2HGDH, LA… …03, LETM1, LIG4, LMNA, LMO…
MAD1L1, MAD2L2, … …1, MAP2K1, MAP2K2, MAP3K1, MAP…
MC2R, MCC, MCM4, MI… …, MDM4, MEFV, MEN1, MET, MFN2, MGAT2, …
MMP1, MN1, MNX1, MPL, MPLKIP, MRAP, MRE11, MS4A1, MSH2, MSH3, MSH6, MSL3, …
MTMR14, MTOR, MUC5B, MUTYH, MVD, MVK, MXI1, MYC, MYCN, MY… …MYH8, M…
NBEAL2, NBN, ND5, N… …A, NEK1, NEK9, NEUROD1, NF1… …NFKB2, N…
NOD2, NODAL, N… …H3, NPM1, NQO2, NR0B… …SD1, NSD2, …
NUMA1, NUP2… …OPCML, PALB2, P… …PAX7, PB…
PCNA, PD… …DGFRA, PDG… …JB1, PHF2…
PHKA… …PIGL, P… …PG2A, PLA…
PI… …POLD1, … …POR,
… …C, PRDM1… …RKN,
… …PN12, PTPRJ, PU… …0,
… …RAD54B, … …, RARA, RASA1, RASG… …, RELA,
…03, RHBDF2, RHO… …RNASEH2B, RNASEH2C, R… …139,
…PIL, RPL10, RI… …L26, RPL27, RPL35, RPL35A, RP… …PS14, RPS15A,
…S27, RP… …AS2, RSPO1, RSPRY1, RTEL1, RUNX1, RYR1, SAMD9,
… …10 …CN9A, SDHA, SDHAF2, SDHB, SDHC, SDHD,SEC23A,
… …SFTPC, SH2B3, SH2D1A, SH3GL1, SH3KBP1, SHH, SHOC2,
SHO… …2A18, SLC25A11, SLC25A13, SLC26A2, SLC26A4, SLC2A2,
SLC37A4, … …4C, SMAD7, SMARCA4, SMARCAD1, SMARCAL1,
SMARCB1, SMA… …SMPD1, SNAI2, SOCS1, SOS1, SOX… …OX2, SOX4, SOX6,
…N, SPINK1, … …SRD5A3, SREBF1, SRGAP1, SR… …SSX1,
…TAG3, STA… …STS, SUFU, SYK, TAF1, TA… …TD24,
…CF3, TCF4, … …TERC, TERT, TET2, TFA…
…R2, TMC… …M127, TMEM216, TM…
…F4, TNPO3, … …, TP53, TP63, TP…
…IS1, TRNS2, … …, TRPV3, TSC1, TS…
…ROD, UROS, USB1, USP8, US…
WD… …53, WRN, WT1, WWOX, … …RCC
ZFHX3, ZI… …BCC9, ACTC1, ACTN2, … …2, CRYAB, DE…
CTNNA3, DES, … …, DTNA, EYA4, FHOD3, … …KIF20A, LAMA4,
MIB1, MYBPC3, MYH… …MYL3, MYLK2, MYOZ2, MY… …PRKAG2,
PSEN2, RAF1, RBM20, RP… …R2, SCN5A, SDHA, SGCD, TCAP, … …TPM1
VCL, OBSCN, CACNA2D1, MYO6, CAP2, PNPLA2, LAMA2, NPPA, NE…

DRUGS (307)

3,4-methylenedioxymethamphetamine, Abacavir, Ace Inhibitors, Acenocoumarol, Acetazolamide, Acetylcholine, Adalimumab, Alkylating Agents, Allopurinol, Alprazolam, Amikacin, Aminoglycoside Antibacterials, Amitriptyline, Amlodipine, Amodiaquine, Amoxicillin, Angiotensin Ii Antagonists, Anthracyclines And Related Substances, Antiepileptics, Antiinflammatory Agents Non-steroids, Antineoplastic Agents, Antipsychotics, Antithyroid Preparations, Antivirals For Treatment Of HIV Infections Combinations, Artesunate, Asparaginase, Aspirin, Atazanavir, Atenolol, Atomoxetine, Atorvastatin, Azathioprine, Benazepril, Beta Blocking Agents, Bevacizumab, Bleomycin, Boceprevir, Bumetanide, Buprenorphine, Bupropion, Capecitabine, Captopril, Carbamazepine, Carbimazole, Carboplatin, Carvedilol, Cavosonstat, Celecoxib, Cerivastatin, Cetuximab, Chloramphenicol, Chlorthalidone, Ciprofloxacin, Cisplatin, Citalopram, Clavulanate, Clindamycin, Clobazam, Clomipramine, Clopidogrel, Clozapine, Cocaine, Codeine, Corticosteroids, Curcumin, Cyanocobalamin, Cyclophosphamide, Cyclosporine, Cysteamine, Cytarabine, Dabigatran, Daclatasvir, Dapsone, Daunorubicin, Deferasirox, Deferiprone, Deleobuvir, Desflurane, Desipramine, Diclofenac, Digoxin, Dihydrostreptomycin, Dimercaprol, Direct Acting Antivirals, Disulfiram, Diuretics, Docetaxel, Doxepin, Doxorubicin, Drugs For Treatment Of Tuberculosis, Efavirenz, Egfr Inhibitors, Enalapril, Enflurane, Ephedrine, Epirubicin, Erlotinib, Escitalopram, Esomeprazole, Etanercept, Ethambutol, Ethanol, Etoposide, Faldaprevir, Febuxostat, Fenofibrate, Fentanyl, Flucloxacillin, Fludarabine, Fluindione, Fluorouracil, Fluoxetine, Flupirtine, Fluvastatin, Folfiri, Folfirinox, Folic Acid, Furosemide, Gefitinib, Geldanamycin, Gemcitabine, Gemtuzumab Ozogamicin, Gentamicin, Gimeracil , Glucarpidase, Glyburide, Halothane, Hmg Coa Reductase Inhibitors, Hydralazine, Hydrochlorothiazide, Hydrocodone, Ibuprofen, Idarubicin, Iloperidone, Imatinib, Imipramine, Indinavir, Indomethacin, Infliximab, Interferon Alfa-2A Recombinant, Interferon Alfa-2B Recombinant, Interferon Beta-1A, Interferons, Irbesartan, Irinotecan, Isepamicin, Isoflurane, Isoniazid, Ivacaftor, Kanamycin, Ketoprofen, Ketorolac, L-methylfolate, Lamotrigine, Lansoprazole, Lapatinib, Ledipasvir, Leucovorin, Levetiracetam, Lisinopril, Losartan, Lovastatin, Lumacaftor, Lumacaftor, Lumefantrine, Maprotiline, Maraviroc, Mephenytoin, Mercaptopurine, Methacholine, Methadone, Methazolamide, Methimazole, Methotrexate, Methoxyflurane, Methylene Blue, Methylphenidate, Metoprolol, Micronomicin, Minocycline, Mirtazapine, Mitoxantrone, Mycophenolate Mofetil, Naltrexone, Neomycin, Nevirapine, Nicotine, Nilotinib, Nitrofurantoin, Nitroprusside, Nitrous Oxide, Nortriptyline, Olanzapine, Omeprazole, Ondansetron, Opioid Anesthetics, Opioids, Opipramol, Opium Alkaloids And Derivatives, Oteracil, Other General Anesthetics, Oxaliplatin, Oxcarbazepine, Oxycodone, Paclitaxel, Panitumumab, Pantoprazole, Pazopanib, Pegaspargase, Peginterferon Alfa-2A, Peginterferon Alfa-2B, Pegloticase, Pemetrexed, Penicillin G, Penicillin V, Perindopril, Phenazepam, Phenazopyridine, Phenobarbital, Phenprocoumon, Phenylephrine, Phenytoin, Platinum Compounds, Prasugrel, Pravastatin, Primaquine, Propafenone, Propofol, Propranolol, Propylthiouracil, Purine Analogues, Pyrazinamide, Pyrimethamine, Quinapril, Rabeprazole, Radiotherapy, Raloxifene, Ramipril, Rasburicase, Remifentanil, Repaglinide, Ribavirin, Rifampin, Risperidone, Ritonavir, Ritonavir, Rituximab, Rocuronium, Rosiglitazone, Rosuvastatin, Salbutamol, Salmeterol, Selective Serotonin Reuptake Inhibitors, Sertraline, Sevoflurane, Sildenafil, Simeprevir, Simvastatin, Sirolimus, Sn-38, Sofosbuvir, Sorafenib, Spironolactone, Stavudine, Streptomycin, Succinylcholine, Sufentanil, Sulfadoxine, Sulfamethoxazole , Sulfasalazine, Sulfonamides Urea Derivatives, Sunitinib, Tacrolimus, Tamoxifen, Tegafur, Telaprevir, Terbutaline, Tezacaftor, Tezacaftor, Thalidomide, Thiazides, Thioguanine, Ticlopidine, Tiotropium, Tobramycin, Tocilizumab, Topoisomerase I Inhibitors, Torasemide, Tramadol, Tranilast, Trastuzumab, Trichloroethylene, Trimethoprim, Tropisetron, Tumor Necrosis Factor Alpha (Tnf-alpha) Inhibitors, Ustekinumab, Valproic Acid, Vancomycin, Varenicline, Velpatasvir, Venlafaxine, Vitamin B-complex Incl. Combinations, Vitamin B-complex, Volatile Anesthetics, Voxilaprevir, Warfarin, Zonisamide

52 GENES

ABCG2, ACE, ADD1, ADRB2, ALDH2, APOE, ATIC, CACNA1S, CES1, CFTR, CHRNA5, CYP2A6, CYP2B6, CYP2C19, CYP2C8, CYP2C9, CYP2D6, CYP3A4, CYP3A5, CYP4F2, DPYD, EGFR, F5, FCGR3A, G6PD, HLA-A, HLA-B, HLA-C, HLA-DPB1, HLA-DRB1, IFNL3, IFNL4, ITPA, KIF6, MT-ND1, MT-RNR1, MTHFR, NAT2, NUDT15, RARG, RYR1, SCN1A, SLC19A1, SLC28A3, SLCO1B1, TNF, TPMT, UGT1A1, UGT1A6, VKORC1, XPNPEP2, XRCC1

GENE

A gene is the basic physical and functional unit of heredity. Genes act as instructions to make proteins and other biological molecules.

MUTATION

A mutation is a permanent change in the DNA sequence of a gene. The following are the different types of mutations.

- **Missense:** Results in different amino acids being encoded at a particular position.
- **Stop-gain:** Prematurely terminates the protein coded by the gene.
- **Frameshift:** Shifts the way the DNA sequence is read.
- **Frameshift -truncation:** Shifts the way the DNA sequence is read and prematurely truncates the protein.
- **Splicesite mutation:** Occurs at the boundary of an exon (protein coding region) and an intron (non-coding region) of a gene.

ZYGOSITY

Zygosity refers to the degree of genetic similarity between the two copies of a gene. There are two types as follows:

- **Homozygous:** Having two identical forms (alleles) of a particular gene.
- **Heterozygous:** Having different forms (alleles) for a particular gene.

INHERITANCE

Inheritance refers to the passing down of traits, characteristics or diseases from one generation to another.

- **Autosomal Dominant:** One copy of a mutated gene from one parent is sufficient to cause the genetic condition.
- **Autosomal Recessive:** Two copies of a mutated gene, one from each parent, are required to cause the genetic condition.
- **X-linked Dominant:** One copy of a mutated gene on the X chromosome from one parent is enough to cause the genetic condition, similar to autosomal dominant in many cases.
- **X-linked Recessive:** Two copies of a mutated gene on the X chromosome, one from each parent, can cause the genetic condition in females, while in males, the mutation typically originates from the mother.

CLASSIFICATIONS

The mutation is classified based on the recommendations of American College of Medical Genetics and Genomics, and the Association for Molecular Pathology (ACMG-AMP), as described below:

- **Pathogenic:** This mutation may directly contribute to the development of disease.
- **Likely Pathogenic:** There is a high likelihood that this mutation is disease-causing. Additional evidence is expected to confirm this assertion of pathogenicity.
- **Variant of Uncertain Significance (VUS):** There is not enough scientific evidence at this time to support a more definitive classification of this mutation.

DIPLOTYPE

The two alleles (versions) inherited by an individual for a particular gene. Includes one maternal and one paternal allele that are both important to evaluate to predict phenotype.

PHENOTYPE

Clinical presentation or observable characteristics of an individual with a particular genotype

REFERENCES

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2. David T. Miller et. al. Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2021 update: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genet-ics in Medicine 2021; 23:1391–1398.

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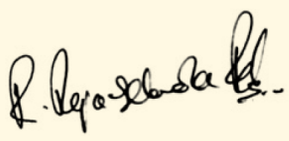

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