GenepoweRx™ TOTAL

ASSESSMENT REPORT AND RECOMMENDATIONS

K&H Personalized Medicine

Ghost Y

PATIENT INFORMATION

Patient Nam e: Ghost Y **Age:** 38 **Gender:** Female

Sample Details:

Collection Date	21st December 2024
Type of sample	Blood
Genomic Specimen ID	KHANAICGPTTL3

Sequencing Details:

Sequencing Type	Germline
WGS/WES/Targeted Seq	Whole Exome Sequencing
Mean Sequencing Depth (x)	207.0
Encoding	Illumina 1.9
Sequence length	151bp
Overall Alignment Rate (%)	99.97
Q30 score (%)	92.59

INDICATION FOR TESTING

Ghost Y 38 year old Female presented with skin issues, Gastritis, frequent sinus infections, frequent urinary tract infections.

CONTENTS

WELCOME

Date: 8th January 2025

Dear Ghost Y

Welcome to Your Personalized Genomics Report!

We are excited to accompany you on your journey to better health through the power of personalized genomics. This report offers unique insights into your genetic profile, empowering you to understand how your genes influence various aspects of your health, from metabolism and nutrition to overall wellness.

This report combines advanced genomic science with actionable recommendations to help you make informed decisions about your lifestyle, diet, and healthcare, enabling you to take proactive steps toward a healthier, more fulfilling life.

Thank you for placing your trust in us to guide your health journey. Together, let us unlock the potential of your genes and pave the way to a brighter, healthier future.

With regards

Dr. Kalyan Uppaluri M.D FACP

Director

GenepoweRx.

This Report is Confidential and belongs to:
Ghost Y

K&H Personalized Medicine

KNOW WHY GENEPOWERX

GenepoweRx, established in Hyderabad in 2019, is a pioneering Genomic Data Analysis and Interpretation company founded by Dr. Kalyan Uppaluri and Dr. Hima Jyothi Challa, USA board-certified internal medicine physicians with advanced clinical genomics training from Stanford and Harvard institutions. With a mission to bring personalized medicine to every individual, GenepoweRx integrates cutting-edge science, technology, and healthcare to transform genomic data into actionable clinical insights.

What sets us apart? Over 60,000 hours of meticulous research have been spent creating a proprietary database and AI-powered tools that transform complex genomic data into actionable insights tailored for the Indian population. By adhering to global standards, including ACMG guidelines, GenepoweRx ensures unmatched accuracy and clinical relevance.

The founders are active members of the variant curation committees of globally recognized platforms such as ClinVar**,** ClinGen**,** and PharmVar, funded by the National Institutes of Health (NIH). Their contributions have enriched these databases with critical genomic interpretations specific to the Indian population, providing a valuable voice in global precision medicine efforts.

From predicting genetic predispositions to enabling targeted healthcare, such as personalized fitness and nutrition recommendations, our reports serve as a bridge between advanced genomics and personalized wellness. Whether assessing health risks, optimizing fitness plans, or tailoring nutritional strategies, we empower individuals and clinicians to make informed, proactive decisions.

At GenepoweRx, we are committed to advancing the frontiers of precision medicine, ensuring secure, innovative, and impactful solutions for a healthier tomorrow.

PERSONALIZED CLINICAL SUMMARY

Dear Ghost Y,

Below is a cumulative clinical summary based on your genomic analysis.

No.	Your Medical Concerns	Recommendation
1	Respiratory Allergies	• Avoid known allergens. • Occasional steaming. • Avoid cold foods. • Avoid fried foods, processed foods. • Prefer diet rich in complex carbohydrates
2	Migraines, Headaches	• Recommend inclusion of Riboflavin rich foods. • Avoid bright lights, sounds and other triggers. • Stress reduction with meditation and gentle yoga. • Maintain a regular sleep pattern. • Follow a healthy dietary compliance. • Regular physical activity is recommended which will improve your oxygen circulation.
3	Gut Health-Constipation	• Include foods which improve colon health like buttermilk, fermented foods (Idli, dosa, Bean sprouts), dark green vegetables. • Avoid any artificial, processed foods and refined sugars. • Maintain proper meal timing. • Avoid excessive stress. • Recommend good amount of hydration.
4	Gastritis	• Avoid excessive fatty foods like fried foods, fast foods, oily foods, cheese and refined sugars. • Maintain a low fat diet. • Recommend regular physical activity.

MEDICAL RECOMMENDATIONS

GenepoweRx®

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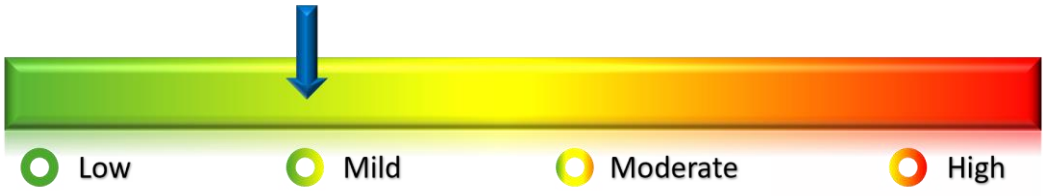
Ghost Y

YOUR MEDICAL CONCERNS ANSWERED

• Respiratory Allergies

You have **Mild** chances of developing **Respiratory Allergies** .

Scale based on Algorithm, compared to general population:



Recommendations:

- **Avoid known allergens.**
- **Occasional steaming.**
- **Avoid cold foods.**
- **Avoid fried foods, processed foods.**
- **Prefer diet rich in complex carbohydrates**

List of Top Publications:

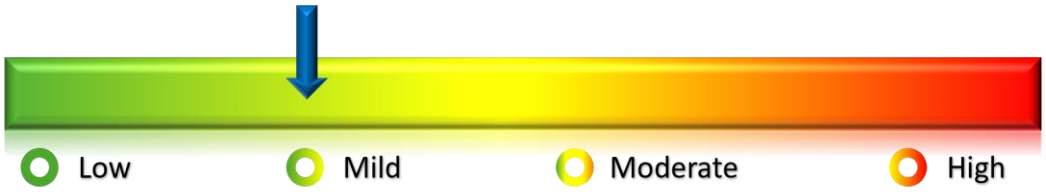
- Genetic studies of the etiology of asthma. - https://pubmed.ncbi.nlm.nih.gov/21543791/
- Introduction to genetics and genomics in asthma: genetics of asthma. - https://pubmed.ncbi.nlm.nih.gov/24162907/

YOUR MEDICAL CONCERNS ANSWERED

• Migraines, Headaches

You have **Mild** chances of developing **Migraines, Headaches** . Mutations seen nonspecific for headaches. Present symptoms could be mostly related to lifestyle factors.

Scale based on Algorithm, compared to general population:



Recommendations:

- **Recommend inclusion of Riboflavin rich foods.**
- **Avoid bright lights, sounds and other triggers.**
- **Stress reduction with meditation and gentle yoga.**
- **Maintain a regular sleep pattern.**
- **Follow a healthy dietary compliance.**
- **Regular physical activity is recommended which will improve your oxygen circulation.**

List of Top Publications:

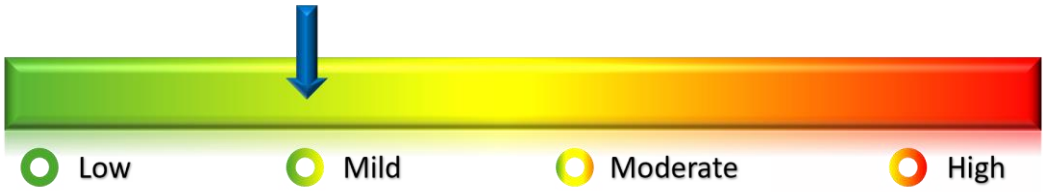
- Migraine Headache: Updates and Future Developments. - https://pubmed.ncbi.nlm.nih.gov/30392545/
- Migraine. - https://pubmed.ncbi.nlm.nih.gov/29523342/

YOUR MEDICAL CONCERNS ANSWERED

• Gut Health- Constipation

You have **Mild** chances of developing **Gut Issues**. Heterozygous moderate impact mutations seen in ***ECM1, NOD2*** genes, can cause symptoms of abdominal pain, constipation, bloating.

Scale based on Algorithm, compared to general population:



Recommendations:

- **Include foods which improve colon health like buttermilk, fermented foods (Idli, dosa, Bean sprouts), dark green vegetables.**
- **Avoid any artificial, processed foods and refined sugars.**
- **Maintain proper meal timing.**
- **Avoid excessive stress.**
- **Recommend good amount of hydration.**

List of Top Publications:

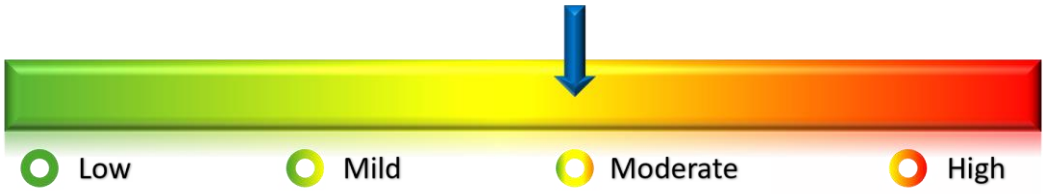
- The genetics of Crohn's disease and ulcerative colitis--status quo and beyond. - https://pubmed.ncbi.nlm.nih.gov/25523552/
- Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease. - https://pubmed.ncbi.nlm.nih.gov/23128233/

YOUR MEDICAL CONCERNS ANSWERED

• Gastritis

You have **Moderate** chances of developing **Gastritis** . Mutations seen in ***HNF1A, C1QTNF6, CEL*** genes

Scale based on Algorithm, compared to general population:



Recommendations:

- **Avoid excessive fatty foods like fried foods, fast foods, oily foods, cheese and refined sugars.**
- **Maintain a low fat diet.**
- **Recommend regular physical activity.**

List of Top Publications:

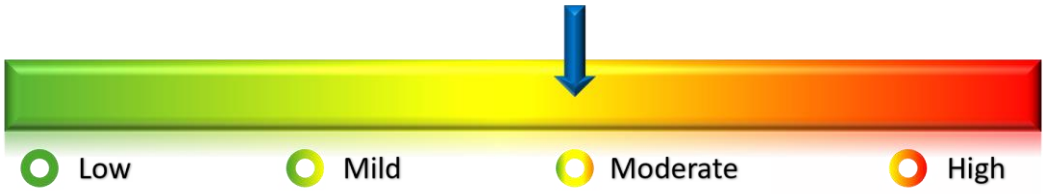
- A Comprehensive Review of Upper Gastrointestinal Symptom Management in Autoimmune Gastritis: Current Insights and Future Directions - https://pubmed.ncbi.nlm.nih.gov/37706145/
- From genes polymorphisms to mucosal expression of cytokines: evaluating IL-23/IL-17 axis in adult patients with gastritis - https://pmc.ncbi.nlm.nih.gov/articles/PMC7751554/

OTHER MEDICAL CONCERNS ANSWERED

• Diabetes

You have **Moderate** chances of developing **Diabetes** . Homozygous mutations seen in ***HNF1A*** gene, can cause disturbance in glucose metabolism.

Scale based on Algorithm, compared to general population:



Recommendations:

- **Please monitor your HbA1C every year.**
- **Yearly eye checkup and renal function tests.**
- **Strictly recommend dietary and lifestyle compliance.**
- **Avoid refined sugars, white rice, sugary drinks.**
- **Your carbohydrate sources should be complex carbs, whole grains (Brown Rice, Hand Pounded Rice, Millets, Whole wheat flour), Green leafy vegetables.**

List of Top Publications:

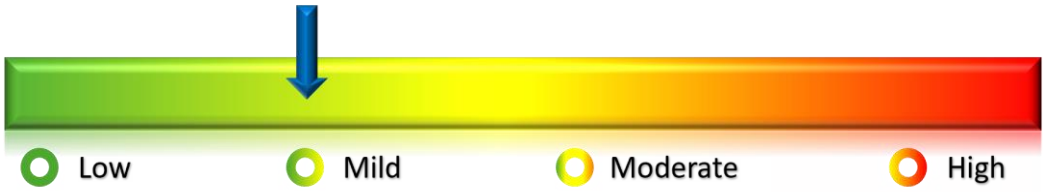
- The past, present, and future of genetic associations in type 1 diabetes - https://pubmed.ncbi.nlm.nih.gov/21792535/
- Genetics of diabetes mellitus and diabetes complications - https://pubmed.ncbi.nlm.nih.gov/32398868/

OTHER MEDICAL CONCERNS ANSWERED

• High Blood pressure

You have **Mild** chances of developing **High Blood pressure** . Few mutations seen for polygenic hypertension.

Scale based on Algorithm, compared to general population:



Recommendations:

- **Monitor BP occasionally.**
- **Maintain your BP about 140/90 mmHg.**
- **Include more vegetables in your diet- Recommended as per DASH (Dietary Approaches to Stop Hypertension) guidelines.**
- **Regular physical activity recommended.**

List of Top Publications:

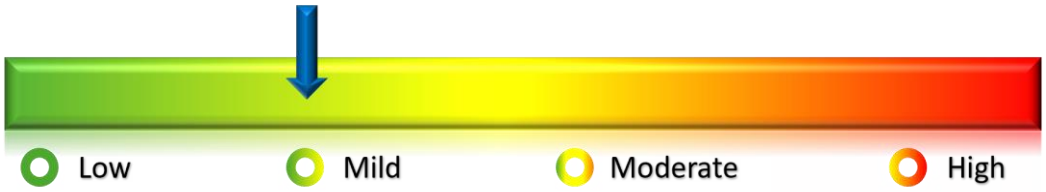
- The genetics of pulmonary arterial hypertension. - https://pubmed.ncbi.nlm.nih.gov/39209481/
- Heritable Pulmonary Arterial Hypertension Overview. http://www.ncbi.nlm.nih.gov/books/NBK1485/

OTHER MEDICAL CONCERNS ANSWERED

• Coronary Artery Disease

You have **Mild** chances of developing **Coronary Artery Disease** . Clinically significant mutations seen in ***APOB*** genes.

Scale based on Algorithm, compared to general population:



Recommendations:

- **Monitor lipid profile every 6 months.**
- **Follow a healthy dietary plan which includes vegetables and good quality fats such as nuts.**
- **Avoid fried, fatty, processed foods and refined sugars.**
- **Only consume cold pressed oils.**
- **Regular physical activity for 30 to 40 minutes a day.**

List of Top Publications:

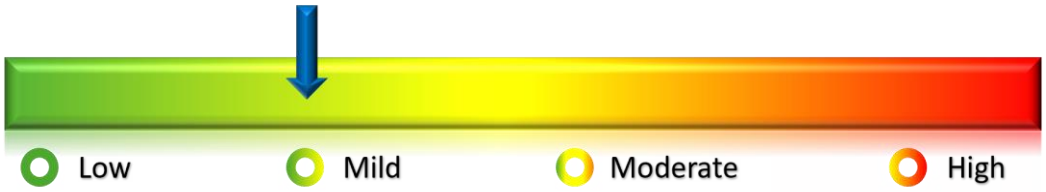
- Relationships Among Heart Rate, β-Blocker Dosage, and Prognosis in Patients With Coronary Artery Disease in a Real-World Database Using a Multimodal Data Acquisition System. - https://pubmed.ncbi.nlm.nih.gov/36216562/
- Combining European and U.S. risk prediction models with polygenic risk scores to refine cardiovascular prevention: the CoLaus|PsyCoLaus Study. - https://pubmed.ncbi.nlm.nih.gov/36652418/

OTHER MEDICAL CONCERNS ANSWERED

• Cholesterol disorders and Hypertriglyceridemia

You have **Mild** chances of developing **Cholesterol disorders and Hypertriglyceridemia** . Clinically significant heterozygous mutations seen in ***APOB, ABCA1, MTTP*** genes, can cause mild elevation in cholesterol levels.

Scale based on Algorithm, compared to general population:



Recommendations:

- **Monitor lipid profile every 6 months/yearly.**
- **Maintain strict dietary compliance which prevents any onset of elevated cholesterol levels.**
- **Recommend a diet low in saturated fats, preferably home cooked meals.**
- **Include more vegetables in your diet.**
- **Consume only cold pressed oils such as sesame, groundnut for Indian cooking and cold pressed olive oil only for stir fries and salads.**
- **Avoid fatty, fried, processed foods and refined sugars.**

List of Top Publications:

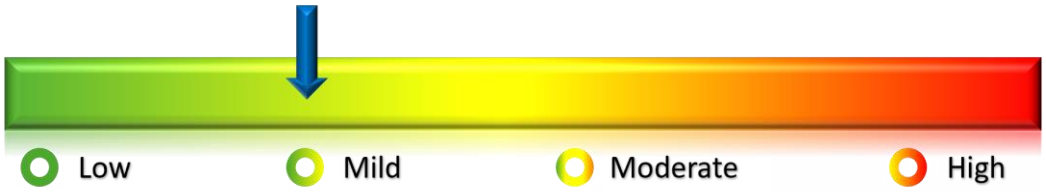
- Familial HDL deficiency characterized by hypercatabolism of mature apoA-I but not proapoA-I. - https://pubmed.ncbi.nlm.nih.gov/9555873/
- Mutations in the ABC1 gene in familial HDL deficiency with defective cholesterol efflux. - https://pubmed.ncbi.nlm.nih.gov/10533863/
- Familial Lipoprotein Lipase Deficiency. http://www.ncbi.nlm.nih.gov/books/NBK1308/
- Molecular analysis of chylomicronemia in a clinical laboratory setting: diagnosis of 13 cases of lipoprotein lipase deficiency. - https://pubmed.ncbi.nlm.nih.gov/24291057/

OTHER MEDICAL CONCERNS ANSWERED

• Alzheimer's Disease

You have **Mild** chances of developing **Alzheimer's Disease** . Clinically significant mutations seen in ***APOE*** genes.

Scale based on Algorithm, compared to general population:



Recommendations:

- **Recommend regular physical activity.**
- **Recommend keeping mind active like solving quizzes in free time.**
- **Recommend stress management.**
- **Recommend healthy diet. Your diet should be rich in complex carbs whole grains (Brown Rice, Hand Pounded Rice, Millets, Whole wheat flour), Green leafy vegetables.**
- **Avoid processed foods.**

List of Top Publications:

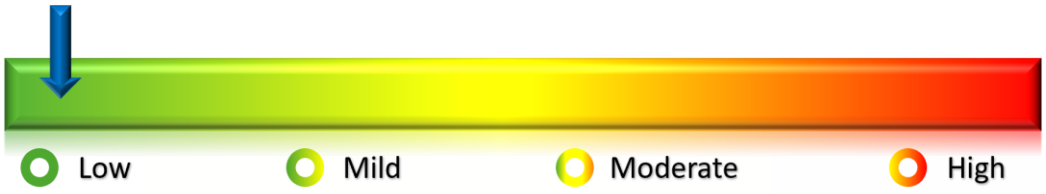
- Genetic aspects of Alzheimer disease. - https://pubmed.ncbi.nlm.nih.gov/18414205/
- Alzheimer's disease: clinical update on epidemiology, pathophysiology and diagnosis. - https://pubmed.ncbi.nlm.nih.gov/29614878/

OTHER MEDICAL CONCERNS ANSWERED

• Fatty Liver

You have **Low** chances of developing **Fatty Liver** .

Scale based on Algorithm, compared to general population:



Recommendations:

- **Monitor liver enzymes, CBC yearly.**
- **Maintain Low fat diet.**
- **Regular physical activity.**
- **Follow dietary compliance.**
- **Maintain healthy blood sugars and cholesterol levels.**
- **Avoid processed,fried foods and also processed sugars or fats.**

List of Top Publications:

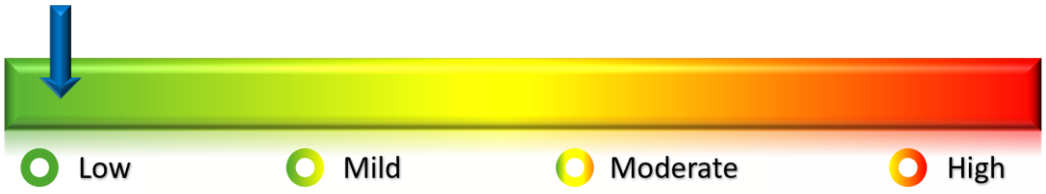
- Clinical outcomes of cryptogenic compared with non-cryptogenic cirrhosis: A retrospective cohort study. - https://pubmed.ncbi.nlm.nih.gov/25867030/
- Identification of 16 novel mutations in the argininosuccinate synthetase gene and genotype-phenotype correlation in 38 classical citrullinemia patients. - https://pubmed.ncbi.nlm.nih.gov/12815590/

OTHER MEDICAL CONCERNS ANSWERED

• Gall stones

You have **Low** chances of developing **Gall stones**.

Scale based on Algorithm, compared to general population:



Recommendations:

- **Recommend to avoid excessive fried, fatty and processed foods**
- **Recommend to maintain a low fat diet.**
- **Include Fiber rich food in your diet.**

List of Top Publications:

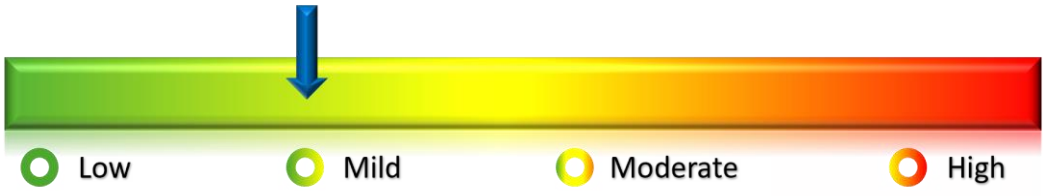
- Increased prevalence of primary sclerosing cholangitis among first-degree relatives. - https://pubmed.ncbi.nlm.nih.gov/15664252/
- A frequent PNPLA3 variant is a sex specific disease modifier in PSC patients with bile duct stenosis. - https://pubmed.ncbi.nlm.nih.gov/23505555/

OTHER MEDICAL CONCERNS ANSWERED

• Musculoskeletal Issues

You have **Mild** chances of developing **Musculoskeletal Issues**.

Scale based on Algorithm, compared to general population:



Recommendations:

- **Monitor for micronutrients and other nutritional deficiencies, specifically magnesium, calcium, and vitamin D, as they play a major role in musculoskeletal health. Replenish them as required.**
- **Include phosphate rich foods like sunflower seeds, pumpkin seeds, buttermilk, aged cheese, green leafy vegetables, beans.**
- **Include regular physical activity such as musculoskeletal strengthening exercises like yoga, brisk walking can help you with the pains and better mobility.**
- **Include calcium rich foods.**

List of Top Publications:

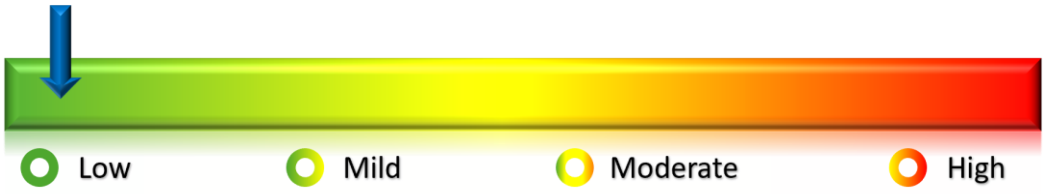
- Kalb S, Martirosyan NL, Kalani MY, Broc GG, Theodore N. Genetics of the degenerated intervertebral disc. World Neurosurg. 2012 Mar-Apr;77(3-4):491-501. doi: 10.1016/j.wneu.2011.07.014. Epub 2011 Nov 7. Citation on PubMed
- The role of polymorphisms of genes encoding collagen IX and XI in lumbar disc disease. - https://pubmed.ncbi.nlm.nih.gov/24636772/
- Molecular diagnosis of neurological disorders in India - https://pubmed.ncbi.nlm.nih.gov/10771899/
- Dystrophinopathies. http://www.ncbi.nlm.nih.gov/books/NBK1119/

OTHER MEDICAL CONCERNS ANSWERED

• Obesity

You have **Low** chances of developing **Obesity** .

Scale based on Algorithm, compared to general population:



Recommendations:

- **Avoid excessive fatty foods like fried foods, fast foods, oily foods, cheese and refined sugars.**
- **Maintain a low fat diet.**
- **Recommend regular physical activity.**

List of Top Publications:

- The Genetics of Obesity - https://pubmed.ncbi.nlm.nih.gov/39343500/
- Leptin and leptin receptor-related monogenic obesity. - https://pubmed.ncbi.nlm.nih.gov/22627381/

NO CLINICALLY SIGNIFICANT MUTATIONS FOUND FOR THESE CONDITIONS:

1. No elevated risk of Thyroid Disorders- Hypothyroidism, Hyperthyroidism compared to the general population.

2. No elevated risk of Anemia- Microcytic, Hemolytic compared to the general population.

3. No elevated risk of Predisposition to Blood clots- Thrombophilia compared to the general population.

4. No elevated risk of Bleeding Disorders compared to the general population.

5. No elevated risk of Parkinson's Disease compared to the general population.

6. No elevated risk of Pancreatic Disorders compared to the general population.

7. No elevated risk of Nephrotic Syndrome (Focal Segmental Glomerulosclerosis, Membranous nephropathy, Minimal Change Disease) compared to the general population.

8. No elevated risk of Interstitial Nephritis, Tubulo interstitial Disease compared to the general population.

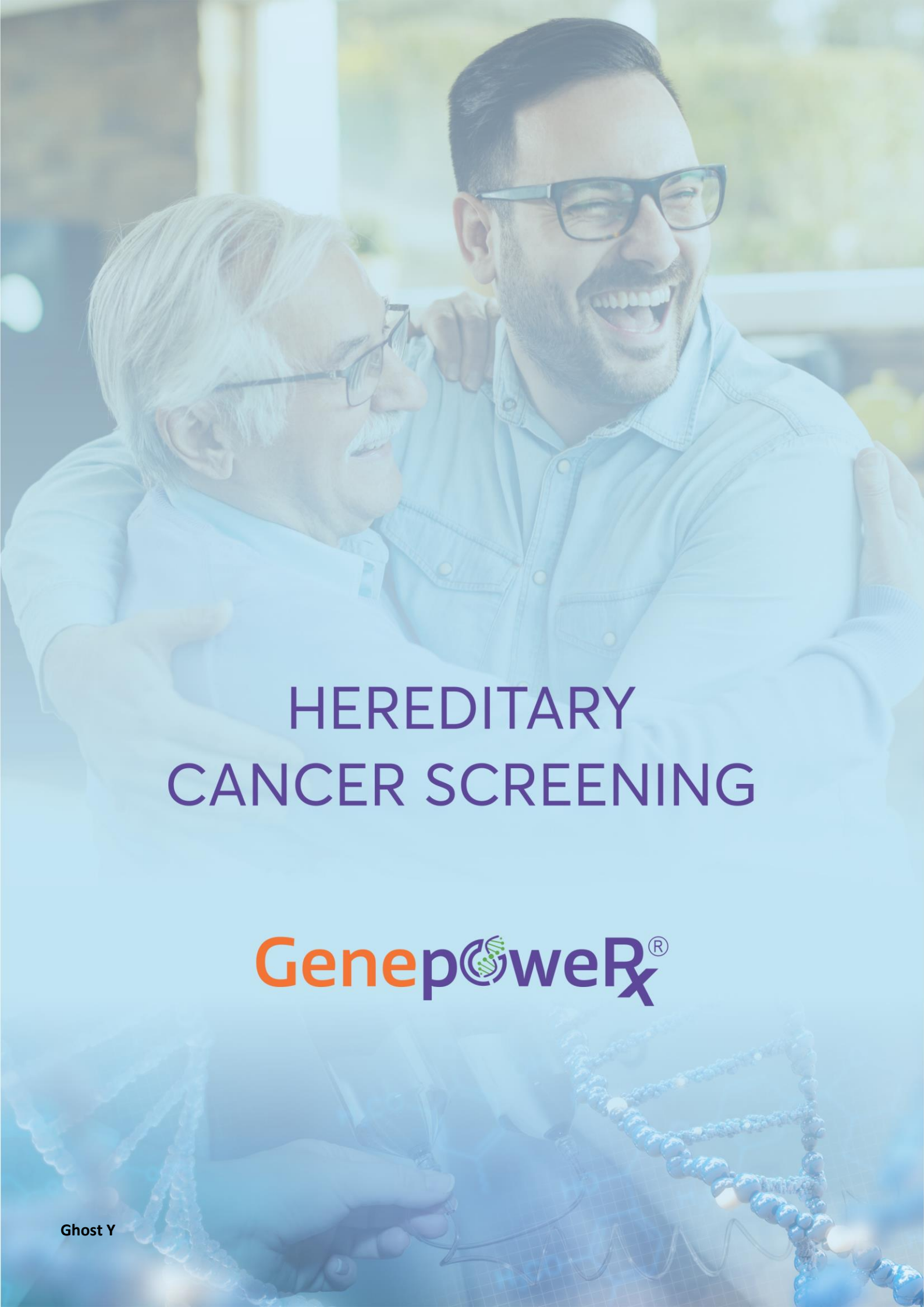
9. No elevated risk of Renal Stones- Calcium Oxalate stones, Cystine stones, Uric Acid Stones compared to the general population.

10. No elevated risk of Mood Disorders- Anxiety, Schizophrenia, Depression compared to the general population.

List of Biomarkers Analyzed

Diabetes	CTLA4, INS, IL2RA, HNF1A, CEL, HLADQ, HLADR, PTPN22, IL2, ERBB3, BACH2, C1QTNF6, PRKQ, RGS1, IL18RAP, CCR5, TAGAP, SH2B3, STAT3, FOXP3, HNF1A, IRS1, SLC2A2, WFS1, GCK, PAX4, WRN, KCNJ11, ABCC8, PDX1, AKT2, HNF4A, KLF11, ENPP1, PPP1R3A, ACADS, LIPC, INSR, TCF7L2
Hypertension	GBA1, DARS2, AGT, PRKAG2, GNB3, INF2, PKD1, MYH9, CELA2A, PKD2, F12, MEN1, COL4A4, COL1A1, NOS3, CACNA1H, CLCN2, CUL3, CYP11B1, CYP11B2, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C2, PDE3A, SCNN1B, SCNN1G, WNK1, WNK4
Cardiac Disorders	LDLR, LDLR-AS1, NOS3, PRKAG2, LRP6, CETP, ABCA1, DYNC2H1, MYH7, LMNA, TNNT2, ACTN2, TTN, CAV3, MYL3, TNNC1, TTR, PLN, FLNC, PRKAG2, VCL, LDB3, ANKRD1, CSRP3, MYBPC3, CACNA1C, MYL2, MYH6, ACTC1, TPM1, TNNI3, MYLK2, JPH2, SGCD, MYPN, LAMA4, TTN, KCNE1, SCN5A, SCN10A, SLMAP, KCND3, CACNA2D1, KCNH2, CACNB2, KCNQ1, KCNE3, CACNA1C, KCNJ8, MYH6, FBN1, HCN4, TTR, SCN1B, TRPM4, ANK2, MYBPC3, KCNA5, PKP2, NPPA, GJA5, TTN, SCN5A, ANK2, KCNH2, BAG3, KCNQ1, SCN2B, SCN3B, KCNA5, ABCC9, HCN4, MYL4, KCNJ2, SCN1B, KCNE2, PITX2, LAMA4, MYBPC3, SNTA1, LAMA4, RYR2, CALM2, TTN, CAV3, TMEM43, SCN5A, ANK2, DSG2, TRPM4, MYLK2, KCNH2, VCL, LDB3, ANKRD1, RBM20, KCNQ1, ILK, MYBPC3, SCN4B, KCNJ5, CACNA1C, MYH6, CALM1, HCN4, KCNJ2, SCN1B, CALM3, SNTA1, JPH2, KCNE2, KCNE1, TECRL, PKP2, LMNA, ACTN2, CACNB2, MYPN, RYR2, CACNA2D1, KCNH2, CACNB2, KCNQ1, CACNA1C, KCNJ2, CASQ2, RYR2, ANK2, TRDN, MYBPC3, CACNA1C, CALM1, TECRL, LMNA, SCN5A, TPM1, DSG2, CACNB2, LDLR, PCSK9, APOB
Cholesterol disorders	PCSK9, LDLR, APOB, APOE, APOC3, MTTP, APOA1, ABCA1, EDN1, CETP, PANK2, LPL, FLAD1, ABCB4, PSAP, PNPLA2, SMPD1, GNPTAB, LIPC, ACADVL, ZMPSTE24, CPT2, CPT1A, HADH, ACADM, AGPAT2, LMNA, PIGC, HADHA, DYNC2LI1, CYP27A1, SLC25A20, OXCT1, SAR1B, NSD1, PEX7, LIPA, DHCR7, LCAT, MLYCD, MC4R, ERCC2, BCHE, LDLRAP1, APOA4, APOC2, GHR, APOA5, STAP1, APBB1, GNPTG, ACADL, BSCL2, ETFDH, ABCG5, PHYH, NPC1, UNKL, APOA2, ANGPTL3, GPIHBP1, SCARB1, LMNB2, ETFA, ABCG8, MCOLN1, PPARG, ETFB, PNPLA6
Thyroid	CTLA4, ALB, DUOX2, GNB1, TSHB, PAX8, STAT1, NKX2-5, TG, RET, DUOX2, ADNP, TUBB1, CHD7, TPO, SLC26A7, TSHR, DUOXA2, TRHR, IGSF1, FOXE1, THRB, IYD, SLA
Parkinson's Disease	MYO6, GBA1, PRKN, MRE11, LRRK2, TBC1D24, VPS35, PARK7, VPS13C, TNR, DCTN1, TNK2, MT-ND6, NR4A2, GNAO1, CACNA1A, ATP1A3, CSTB, KCNN2, GAB1, SLC6A17, POLG, PLA2G6, NR4A2, TRPM7, SNCA, PRKN, LRRK2, PINK1, PARK7
Dementia	APOE, APP, PSEN1, PSEN2, Lewy Body, SNCA, MAPT, GBA, CNTN1, APP
Migraine	CACNA1S, CACNA1A, MAT1A, SLC7A7, KCNK18, CSNK1D, ATP1A2, SCN1A, PRRT2
Respiratory Allergies	LRRC6, DNAH5, HYDIN, LRRC56, DNAH8, DNAI2, GAS8-AS1, CCDC151, CCDC65, CCDC39, TTC12, DNAAF5, C11orf70, LOC105371891, GAS8, DNAAF2, DNAH11, DNAAF1, DNAH9, HSDL1, MCIDAS, DRC1, ARMC4, FAM187A, DNAAF3, DNAJB13, CCDC40, CCDC103, DOCK8
Microcytic anemia	LARS1, SLC11A2, TMPRSS6, IREB2, TRNT1, SLC25A38, LARS2, DNM1L, YARS2, PUS1, HBB, TF, SLC19A2, DHFR, POLE, MTHFD1, AMN, CUBN, TERT, SPTA1, NT5C3A, GSR, AK1, HK1, VPS4A, SLC4A1, GPI, SPTB, SOCS1, GATA2, TTR, G6PD, CDIN1, CDAN, KLF1, SEC23B, RPL11, RPL5, RPS7, RPL35A, RPS10, RPS24, RPS26, RPS29, RPS28, RPS19, TP53, RPL15, BRCA2, POLG, BRCA1, VRK2, FANCL, FANCD2, FANCE, XRCC2, FANCG, FANCC, FANCF, FANCM, RAD51, FANCI, SLX4, ERCC4, PALB2, FANCA, RAD51C, BRIP1, TJP2, BAAT, PAH, TERC, TERT, NBN, PRF1, SBDS, VPS4A, MPIG6B
Fatty liver	PNPLA3, IFNL4, TM6SF2, MBOAT7, GCKR, HSD17B13
Gallstones	ABCG8, APOE, APOC3, MTTP, APOA1, ABCA1, LDL, CETP
Gut Health	IL17RA, LAMB1, TNFRSF13B, JAK2, NOD2, LRRK, IRGM, ATG16L1, STAT2, STAT3, IL24R, IL12B, TYK2, TNFSF15, OCTN2, ECM1, CDH1, HNF4A, GNA12, FCGR2A, FOXO3, XACT, IGFBP1
Gastritis	CTLA4, INS, IL2RA, HNF1A, CEL, HLADQ, HLADR, PTPN22, IL2, ERBB3, BACH2, C1QTNF6, PRKQ, RGS1, IL18RAP, CCR5, TAGAP, SH2B3, STAT3, FOXP3, HLADR4, HLADR7, HLADQ7, HLADR1, HLAB13, HLADQW3, HLACW6, HLAA30, AIS1, FOXD3, NALP1, CXCL10, HLAA2, PTPN22, LPP, IL2RA, UBASH3A, C1QTNF6

Glomerular Diseases	AQP2, AVPR2, AVP, PARK7, HNF1B, PRKAG2, NPHP4, NPHS2, CFH, NLRP3, NPHP1, SMARCAL1, COL4A4, COL4A3, LAMB2, COQ2, CFI, CFB, CD2AP, SLC17A5, ANLN, EYA1, CRB2, LMX1B, PLCE1, PAX2, WT1, TRPC6, NUP107, INF2, RPGRIP1L, C3, NPHS1, ACTN4, APOL1, MYH9, PDSS2, MME, KANK1, KANK2, KANK4, CFH, CFHR5, DGKE, BLK, CR2, CTLA4, DNASE1L3, ITGAM, TNF, TREX1
Tubular Interstitial disease, Interstitial Nephritis	FAN1, MUC1, UMOD, NPHP4
Renal stones	AGXT, GRHPR, HOGA1, SLC26A1, CYP24A1, SLC26A1, XDH, MOCOS, PREPL, SLC2A9, SLC34A1, SLC3A1, SLC22A12, AGXT2
Skin Health	ITGA3, ALOXE3, RTEL1-TNFRSF6B, ST14, LRP1, SULT2B1, COL17A1, DSG1, LIPN, ACD, NLRP1, CYP4F22, KRT74, NOP10, ABCA12, LOC105378642, KRT5, BIVM-ERCC5, FLG, ATP2A2, POLH, PLEC, TMC6, KRT10, TRPV3, ALOX12B, DSP, KRT6C, SERPINB8, ITGB4, LAMA3, LSM3, DYNC2H1, KRT9, DSG1-AS1, ERCC4, AAGAB, CTC1, XPA, KRT14, ERCC2, COL7A1, TGM5, KRT1, DDB2, XPC, KRT2, LOC107986063, EXPH5, LAMB3, LAMC2, MIR4260
Rheumatoid Arthritis	HLA DRB1, STAT4, PTPN22, TRAF1-C5, OLIG3, TNFAIP3, IL20RA, PADI4, CTLA4, FOXO3, IL10
Muscular Dystrophy	TSPAN1, POMGNT1, LMNA, TOR1AIP1, DYSF, LIMS2, TTN, DAG1, GMPPB, SGCB, TRAPPC11, SGCD, LAMA2, CRPPA, PLEC, FKTN, POMT1, ANO5, TUBA1A, SGCG, POMT2, CAPN3, TCAP, SGCA, FKRP, STRN4, POGLUT1, BVES, TTN-AS1, CRPPA-AS1, SACS, CAV3, RYR1, SNTA1, LMNA, SELENON, PLEC, LAMA2, MYH7, TTN, SIGMAR1, PLEKHG5, HNRNPDL, POMGNT2, TSPAN1, SMCHD1, TMEM43, COL6A3, BICD2, SMN1, MYOT, ESR1, PABPN1, COL12A1, IGHMBP2, SH3TC2, NEFL, PKD2L2-DT, SUN2, COL6A2, SETX, SYNE1-AS1, RXYLT1, ASCC1, GOSR2, TRPV4, TRIM32, POMK, DYNC1H1, CHKB, RYR1, RIF1, CHKB-CPT1B, TRIP4, SPG7, PMP22, VAPB, MORC2, TYMP, SMN2, SCO2, TNPO3, SYNE1, NEB, GARS1, DNAJB6, ASTN2, B3GALNT2, SUN1, BCL2L2-PABPN1, ASAH1, PYGM, SYNE2, OXTR, GTPBP1, COL6A1, KIF1B, SPTAN1, PMM2, FKBP14, VRK1, LARGE1, DPM2, B4GAT1, ITGA7, ACTA1, INPP5K, LOC123864065, RXYLT1-AS1
Mood Disorders	GAD, SLC6A4, COMT, DAOA/G30, DAO, DISC1, DTNBP1, GABRB2, LRG1, ZNF804A
Obesity	FTO, PCSK1, MC4R, LEPR, LEP, FABP2, ENPP1, PRMT7, KCNH2, POMC, KSR2, BDNF, SH2B1, MRAP2, ADCY3, CPE, ADRB3, AFF4, PPARG, GHRL, PHIP, PDSS1, TUB, SDC3, CEP19, ABCC8, SMARCA4, MYH9, RYR1, SIM1, BRCA2, SCN1A, NTRK2
Hypertriglyceridemia	LPL, APOC2, APOA5, LMF1, GPIHBP1, APOA1, TRIB1, MLXIPL, GCKR, FADS1, FADS2, FADS3, NCAN, APOB, PLTP, ANGPTL3, APOE
Osteoporosis & Degenerative Joint Disease, Cartilage degeneration	MATN3, SMAD6, SMAD3, ACAN, HPGD, COL9A2, COL11A1, COL5A2, SLC26A2, COL11A2, COL5A1, COL2A1, TRPV4, FBN1, COL1A1, COMP, LACC1, CIITA, NOD2, NLRP1, LRP5, ANO5, IFITM5, FGFR1, GNPTAB, BICD2, GNAS, SFRP4, TENT5A, LRP5, COL1A1, FKBP10
Pancreatic Disorders	BRCA2, PALB2, CDKN2A, ATM, TP53, STK11, MLH1, MSH2, MSH6, PMS2, EPCAM



HEREDITARY CANCER SCREENING

GenepoweRx®

Ghost Y

GENOMIC TEST REPORT FOR CANCER RISK PREDISPOSITION

NCCN clinical practice guidelines were employed for testing hereditary cancer risk. As part of these guidelines, 46 genes were screened to identify the risk of predisposition to 5 types of cancers.

- Breast
- Ovarian
- Pancreatic
- Prostate
- Colorectal cancers.

Test Result

Negative for Pathogenic/Likely pathogenic variants associated with Hereditary Cancer risk.

Recommendations:

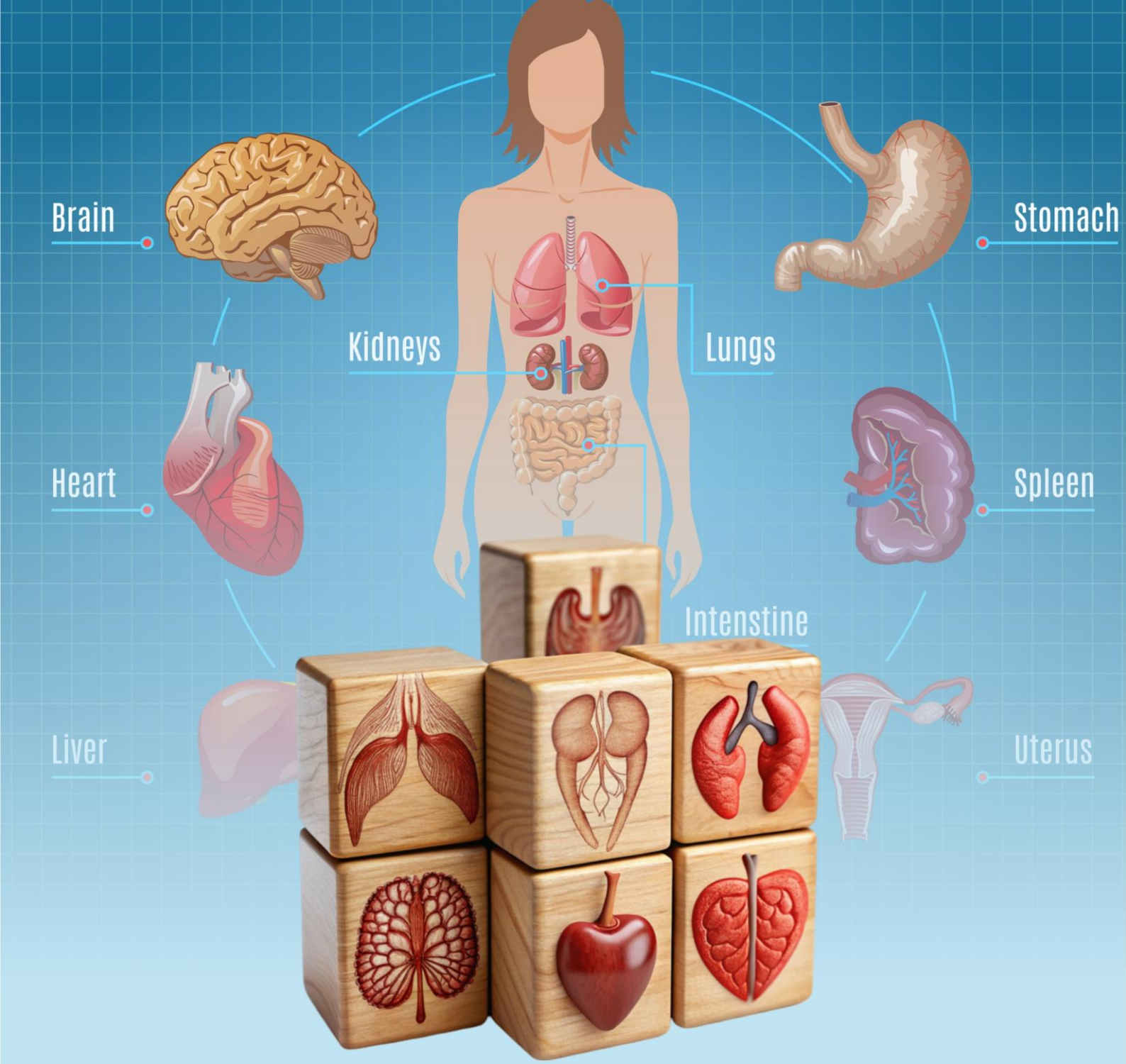
- **After the age of 50 years, PAP smear, Mammogram and a gynecologist follow up every year is recommended.**
- **Follow a healthy diet (Anti Inflammatory diet principles).**
- **Regular physical activity and healthy lifestyle.**

Clinically Significant Variants

- No Pathogenic/Likely Pathogenic Single Nucleotide Variants(SNV's), Copy Number variants (CNV's) associated with condition indicated for testing found.

Biomarkers Evaluated

APC, ATM, AXIN2, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, EPCAM, FANCC, GALNT12, GREM1, HOXB13, MBD4, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD50, RAD51C, RAD51D, RECQL, RINT1, RNF43, RPS20, STK11, SLX4, SMAD4, SMARCA4, TP53, XRCC2



CONDITION SPECIFIC REPORT

GenepowerRx®

Ghost Y

ROSACEA/ AUTOIMMUNE CONDITION

A total of 315 genes associated with Rosacea/ autoimmune condition have been primarily screened from the whole exome panel, which consists of ~22,000 genes.

Test Result

Negative for Pathogenic/Likely pathogenic variants associated with Rosacea/ autoimmune condition.

Recommendations:

- **Genetically, you are at a LOW risk.**
- **Clinical correlation is recommended.**
- **Avoid excessive sun exposure. Consider sunscreen usage.**
- **Use medications as recommended by the dermatologist.**
- **Avoid fried, processed foods, and refined sugars as they can increase skin inflammation.**

Clinically Significant Variants

- No Pathogenic/Likely Pathogenic Single Nucleotide Variants(SNV's), Copy Number variants (CNV's) found.
- However, one Variant of Uncertain Significance (VUS) associated with indication of testing identified.

Gene	Location	Variant	Zygosity	Classification
HLA-DRB1	Exon 2	p.Tyr107Ter	Heterozygous	Uncertain significance

Variant Details

p.Tyr107Ter in Exon 2 of HLA-DRB1

Criteria : BS1, PVS1

Classification : Uncertain significance

Variant Interpretation

The stop gained NM_002124.4(*HLA-DRB1*):c.321C>A (p.Tyr107Ter) has not been reported previously as a pathogenic variant nor as a benign variant, to our knowledge. The p.Tyr107Ter variant is observed in 115/30,436 (0.3778%) alleles from individuals of gnomAD v4 EastAsian background in gnomAD v4 All, which is greater than expected for the disorder. This variant is predicted to cause loss of normal protein function through protein truncation. This variant is a stop gained variant which occurs in an exon of *HLA-DRB1* upstream of where nonsense mediated decay is predicted to occur. There is another pathogenic loss of function variant downstream of this variant, indicating that the region is critical to protein function. For these reasons, this variant has been classified as Uncertain Significance.

Technical Data

Genomic Change	NC_000006.12:g.32584158G>T
Transcript and Coding Change	NM_002124.4:c.321C>A (p.Tyr107Ter)
Location	6:32584158
dbSNP Identifier	rs11554463
ClinVar Variant ID	
gnomAD v4 EuropeanNonFinnish Allele Frequency	115/30436 (0.38%)
NGS Reads Supporting Change	34.33% (23 of 67)
****Insilico parameters**	SIFT: — Polyphen2: --

**Insilico parameters- predict the effect of the variant on the protein (SIFT, PolyPhen-2)

Gene Summary

HLA-DRB1 belongs to the HLA class II beta chain paralogs. The class II molecule is a heterodimer consisting of an alpha (DRA) and a beta chain (DRB), both anchored in the membrane. It plays a central role in the immune system by presenting peptides derived from extracellular proteins. Class II molecules are expressed in antigen presenting cells. The beta chain is approximately 26-28 kDa. It is encoded by 6 exons. Exon one encodes the leader peptide; exons 2 and 3 encode the two extracellular domains; exon 4 encodes the transmembrane domain; and exon 5 encodes the cytoplasmic tail. Within the DR molecule the beta chain contains all the polymorphisms specifying the peptide binding specificities. Hundreds of DRB1 alleles have been described and some alleles have increased frequencies associated with certain diseases or conditions.

Biomarkers Evaluated

TBX2, IL18BP, PLCG2, POLG, FOXD3, C1S, ADA2, SEC23B, CDKN1B, CLCNKB, SLC12A3, STAT1, TOM1, NBN, LRBA, STK4, NLRP1, NFKB1, PIK3CG, CD3G, CBLB, LCP2, DEF6, STIM1, ARPC5, TLR8, PSMG2, FCGR3B, SASH3, NFKB2, CD81, TLR7, CTNNBL1, SMPD1, MAGT1, SOCS1, LAT, GALC, TNFSF12, MS4A1, IRF2BP2,+300 genes

References

PMID	CITATION
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26582918, 29165669	Landrum MJ, Lee JM, Benson M, Brown G, Chao C, Chitipiralla S, Gu B, Hart J, Hoffman D, Hoover J, Jang W, Katz K, Ovetsky M, Riley G, Sethi A, Tully R, Villamarin-Salomon R, Rubinstein W, Maglott DR. ClinVar: public archive of interpretations of clinically relevant variants. Nucleic Acids Res. 2016 Jan 4;44(D1):D862-8. doi: 10.1093/nar/gkv1222. Epub 2015 Nov 17
10447503	Sherry ST, Ward M, Sirotkin K. dbSNP-database for single nucleotide polymorphisms and other classes of minor genetic variation. Genome Res. 1999 Aug;9(8):677-9.
19344873	Firth HV, Richards SM, Bevan AP, Clayton S, Corpas M, Rajan D, Van Vooren S, Moreau Y, Pettett RM, Carter NP. DECIPHER: Database of Chromosomal Imbalance and Phenotype in Humans Using Ensembl Resources. Am J Hum Genet. 2009 Apr;84(4):524-33. doi: 10.1016/j.ajhg.2009.03.010. Epub 2009 Apr 2.

FITNESS AND NUTRI GENOMICS REPORT

GenepowerRx®

YOUR FITNESS AND NUTRITION GENES

Carbohydrate , Lipid , Protein, Musculoskeletal Metabolism

MUTATIONS SEEN IN GENES	RECOMMENDATIONS
ACSF3, CD320, CUBN, MCEE, MTRR, MVK, TCN2	• Include dark green leafy vegetables, Vitamin B12 rich foods (Eggs, Fish, seafood). • Recommend Vitamin B12 and folate supplementation.
NLRP12	• Avoid excessive cold exposure, ingestion of cold foods. Avoid excessive stress.
GATM	• Avoid heavy workouts. Please ensure to include foods like Pumpkin seeds, Walnuts, Peanuts, Almonds, Eggs, Buttermilk.
CDH23	• Recommend yearly eye exam and hearing tests after age 50
ARSA	• Include foods with complex carbohydrates. Fats to constitute only 20% of the total meal.
CRACR2B	• Avoid high fat diets and prolonged periods of fasting. Include riboflavin supplementation or riboflavin rich foods like eggs, dairy, green vegetables, fish.
ACOX1, PEX16, PEX2	• Can cause neuropathy, tingling, numbness, shooting pains. Include Fish Oil supplements.
PCK1	• Can cause hair loss and skin aging. Include biotin rich foods like soy bean, eggs.
ASNS	• Avoid high protein diets.Include diet rich in complex carbohydrates.
FLVCR1	• Include foods which improve iron stores like dates, legumes, dark green vegetables. Poor oral absorption of iron seen with this mutation. Recommend monitoring CBC and Iron profile yearly.

MUTATIONS SEEN IN GENES	RECOMMENDATIONS
PDHX	• Include thiamine rich foods like brown rice, legumes, whole grains
ANK2, CACNA1C-AS1, CACNA1F, KCNJ5, RBM20	• Prefer exercises like brisk walking, strength training with lesser weights. Avoid Fasting
ALG12, COG4, COG6, DPM2, MPDU1, SLC16A1	• After work out, include a meal rich in complex carbohydrates and proteins.
COL11A1, IFITM5, VDR	• Include phosphate rich foods like sunflower seeds, pumpkin seeds, buttermilk, aged cheese, green leafy vegetables, beans
COL6A1, COL6A3, NEB	• To avoid muscle cramps after strenuous workouts, include foods like bone broth, nuts, beans, fish, broccoli, whole grains.
TRPM6	• Recommend daily magnesium supplementation
CDH23, LCT	• Milk, fresh cheeses, fresh milk cream can cause symptoms of bloating, malabsorption. Avoid fresh dairy. Can include buttermilk, aged cheeses. Increase consumption of fruits.
FBP1	• Strictly avoid KETO diets. Avoid prolonged periods of fasting. Fructose overload with processed drinks and artificially sweetened foods can cause malabsorption and change of the gut bacterial flora. This can lead to decreased immunity, allergies.

List of Top Publications in Fitness:

- https://pubmed.ncbi.nlm.nih.gov/31381138?_ga=2.194843053.1854089511.1683708633-990785819.1682571628
- https://febs.onlinelibrary.wiley.com/doi/full/10.1111/febs.14028
- https://pubmed.ncbi.nlm.nih.gov/21943391?_ga=2.266818958.1854089511.1683708633-990785819.1682571628

List of Top Publications in Lipids:

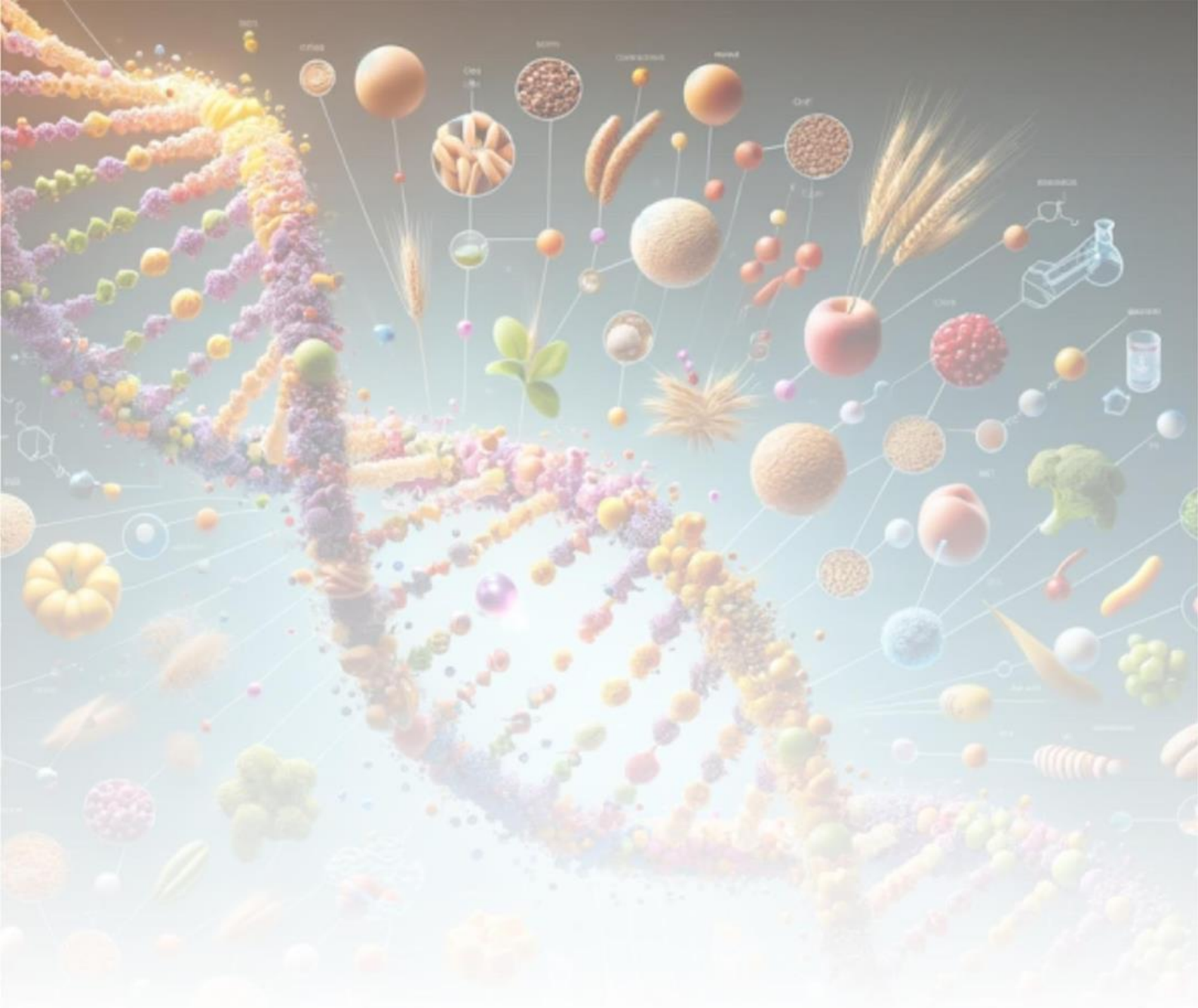
- https://pubmed.ncbi.nlm.nih.gov/17186413/
- https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8750194/
- https://pubmed.ncbi.nlm.nih.gov/11929856/

List of Top Publications in Carbs:

- https://pubmed.ncbi.nlm.nih.gov/26964834/
- https://pubmed.ncbi.nlm.nih.gov/24270786/
- https://pubmed.ncbi.nlm.nih.gov/27707936/

List of Top Publications in Proteins:

- https://pubmed.ncbi.nlm.nih.gov/35568239/
- https://pubmed.ncbi.nlm.nih.gov/19301095/
- https://pubmed.ncbi.nlm.nih.gov/36639369/



VITAMINS AND MINERALS REPORT

GenepoweRx®

YOUR FITNESS AND NUTRITION GENES

Vitamins and Minerals

ANALYSIS FOR	MUTATIONS SEEN IN GENES	RECOMMENDATIONS
MAGNESIUM	TRPM6	• Recommend daily magnesium supplementation to avoid leg cramps and fatigue
VIT-K	No clinically significant mutations identified.	No supplementation required for this nutrient
VIT-E	No clinically significant mutations identified.	No supplementation required for this nutrient
PHOSPHATE	ALPL, COL11A1, IFITM5, SLC34A3, TNFRSF11A, VDR	• Include phosphate rich foods like sunflower seeds, pumpkin seeds, buttermilk, aged cheese, green leafy vegetables, beans
VIT-B12 and Iron	ACSF3, CD320, CUBN, MCEE, MTRR, MVK, TCN2, CP, FLVCR1, TMPRSS6, CD320, MTRR, MVK, ABCD4	• Include dark green leafy vegetables, Vitamin B12 rich foods (Eggs, Fish, seafood) • Include foods which improve iron stores like dates, legumes • Recommend supplementation along with B complex
MOLYBDENUM	No clinically significant mutations identified.	No supplementation required for this nutrient
BIOTIN, holocarboxylase synthetase	MCEE, PCK1	• Can cause skin aging and hair loss • Need to supplement biotin • Required for smooth skin and prevents hair fall • Include biotin rich foods like soy bean, eggs
VITAMIN C	No clinically significant mutations identified.	No supplementation required for this nutrient

ANALYSIS FOR	MUTATIONS SEEN IN GENES	RECOMMENDATIONS
Calcium	CACNA1S, COL11A1, SLC26A1, SLC34A3, TNFRSF11A	• Include calcium rich foods like buttermilk, yogurt, aged cheeses, nuts, dark green leafy vegetables • Recommend regular physical activity
Vitamin-D	CD27-AS1	• Monitor Vitamin D levels regularly • Use Vitamin D supplements as needed (if serum levels are low)
Pantothenic acid	No clinically significant mutations identified.	No supplementation required for this nutrient
NIACIN (VITAMIN B 3)	No clinically significant mutations identified.	No supplementation required for this nutrient
RIBOFLAVIN (VITAMIN B2)	No clinically significant mutations identified.	No supplementation required for this nutrient

List of Top Publications:

- Surendran, S., Adaikalakoteswari, A., Saravanan, P. et al. An update on vitamin B12-related gene polymorphisms and B12 status. Genes Nutr 13, 2 (2018). https://doi.org/10.1186/s12263-018-0591-9
- https://rarediseases.org/rare-diseases/biotinidase-deficiency/
- Ortigoza-Escobar JD, Alfadhel M, Molero-Luis M, Darin N, Spiegel R, de Coo IF, Gerards M, Taylor RW, Artuch R, Nashabat M, Rodríguez-Pombo P, Tabarki B, Pérez-Dueñas B; Thiamine Deficiency Study Group. Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Ann Neurol. 2017 Sep;82(3):317-330. doi: 10.1002/ana.24998. Epub 2017 Aug 30. PMID: 28856750.

DIET PLAN RECOMMENDATIONS

GenepoweRx®

Your Diet Chart

Day-1

Morning Detox Water 5:00 AM	**Lukewarm Water with Honey** **Ingredients:** 1 cup lukewarm water 1-2 teaspoons of honey (use raw, unprocessed honey for best results) Optional: a squeeze of fresh lemon juice.	
Breakfast 9:00 AM	**Oats Idly with Chutney**	
Lunch 1:00 PM	**Beetroot Rice** **Ingredients:** 1 cup basmati rice - washed and soaked for 30 minutes then drained, 1 medium-sized beetroot - peeled and grated, 1 small onion finely chopped, 2 cloves of garlic minced, 1-inch piece of ginger minced, 1 green chili finely chopped (adjust to your spice preference), 1/2 teaspoon cumin seeds, 1/2 teaspoon mustard seeds, 1/2 teaspoon turmeric powder, 1/2 teaspoon coriander powder, 1/2 teaspoon cumin powder, 1/2 teaspoon red chili powder (adjust to your spice preference), 1 tablespoon olive oil or ghee (for a healthier option, use olive oil), Salt to taste, Fresh coriander leaves for garnish (optional). **Instructions**: 1. Cook rice in boiling water, then drain and set aside. 2. Sauté aromatics until onion is translucent. 3. Add spices and grated beetroot, sauté until beetroot softens. 4. Combine cooked rice, mix for vibrant color. 5. Season and cook until beetroot is done. 6. Garnish and serve the healthy Beetroot Rice as a colorful side dish or a light meal.	
Workout Snack 5:00 PM	**Peanut and Oat Energy Bars:** **Ingredients:** Crushed peanuts, rolled oats, dates, and a touch of cocoa powder. Method: Blend ingredients together, shape into bars, and refrigerate for a nutritious and satisfying energy bar.	
Dinner 9:00 PM	**2 Jowar Roti With Chicken Curry / Mushroom Curry** **+ 1 Glass Buttermilk**	

Day-2

Morning Detox Water 5:00 AM	**Jeera Water** **Ingredients:** 1-2 teaspoons of whole cumin seeds (jeera), 1-2 cups of water. **Instructions:** 1. Boil/heat water in a kettle for 10 minutes and add 1-2 teaspoons of cumin into it. 2. Let the water cool at room temperature for infusion. 3. Strain and serve in glass.	
Breakfast 9:00 AM	**Millet Pongal / Pongal**	
Lunch 1:00 PM	**Gajar-Matar Sabzi (Carrot-Peas Curry), Pair with Roti or Brown Rice** **Ingredients**: 2 cups carrots peeled and diced, 1 cup green peas (fresh or frozen), 1 large onion finely chopped, 2 tomatoes finely chopped, 1 tablespoon ginger-garlic paste, 1 teaspoon cumin seeds, 1 teaspoon turmeric powder, 1 teaspoon ground cumin, 1 teaspoon ground coriander, ½ teaspoon red chili powder (adjust to taste), ½ teaspoon garam masala, 2 tablespoons cooking oil (olive oil or any vegetable oil of your choice), Salt to taste, Fresh cilantro chopped (for garnish). **Instructions**: 1. Heat oil in a pan over medium heat. Add cumin seeds and let them splutter. 2. Add chopped onions and sauté until they become golden brown. 3. Add ginger-garlic paste and sauté for a minute until the raw smell disappears. 4. Add chopped tomatoes, turmeric powder, ground cumin, ground coriander, red chili powder, and salt. Cook until the tomatoes are soft and the oil starts to separate from the masala.	

Day-2

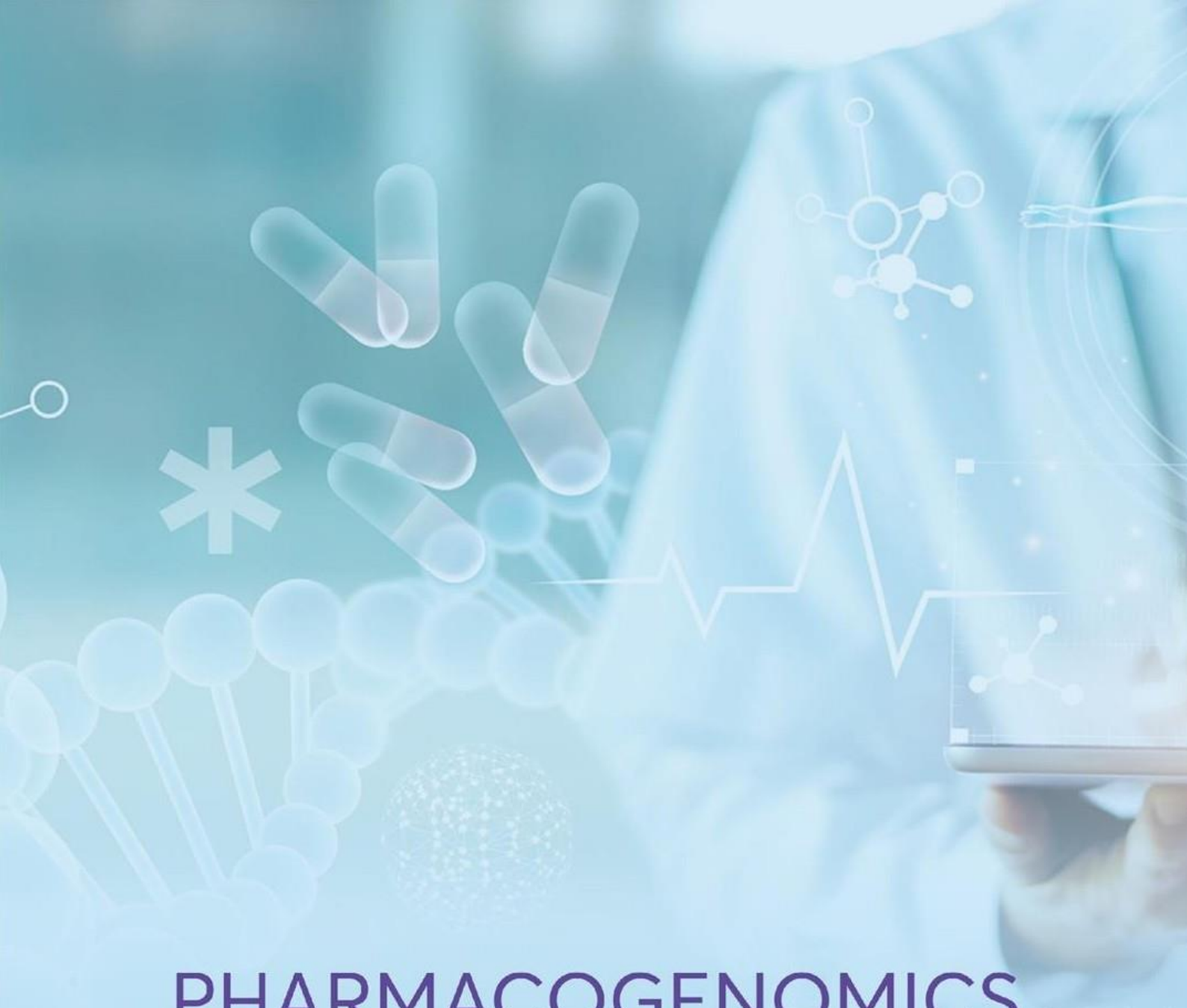
Workout Snack 5:00 PM	**Pumpkin Seeds (Pepitas)**  **Ingredients:** Fresh pumpkin seeds (pepitas) Olive oil (or your preferred cooking oil) Salt Optional seasonings (choose one or more): Paprika Garlic powder Cayenne pepper Cinnamon and sugar (for sweet pepitas) Your favorite spice blend (e.g., chili powder, curry powder) **Instructions:** 1. Prepare the Pumpkin Seeds: Remove the pumpkin seeds from the pumpkin, if you're starting with a whole pumpkin. Rinse the seeds thoroughly under cold water to remove any pulp and pat them dry with paper towels. 2. Preheat the Oven: Preheat your oven to 300°F (150°C). 3. Toss with Oil and Seasonings: In a mixing bowl, toss the pumpkin seeds with a drizzle of olive oil, just enough to lightly coat them. Sprinkle salt and your choice of seasonings over the seeds. Use your hands or a spatula to evenly distribute the oil and seasonings. 4. Spread on a Baking Sheet: Spread the seasoned pumpkin seeds out on a baking sheet in a single layer. Make sure they are not crowded to allow for even roasting. 5. Roast in the Oven: Place the baking sheet in the preheated oven and roast the pumpkin seeds for about 20-30 minutes, or until they are golden brown. Be sure to stir or shake the pan every 10 minutes for even roasting. 6. Cool and Enjoy: Remove the roasted pumpkin seeds from the oven and let them cool on the baking sheet. They will continue to crisp up as they cool. Once completely cooled, transfer the pumpkin seeds to an airtight container for storage.
Dinner 9:00 PM	**1 Bowl Cracked Wheat Upma With Onions, Carrots, Green Peas.** **+ 1 Glass Buttermilk** 

Day-3

Morning Detox Water 5:00 AM	**Lukewarm water with fenugreek seed powder** **Ingredients:** **1 teaspoon of fenugreek seed powder, 1 cup (8 ounces) of lukewarm water** **Instructions:** 1. Prepare Fenugreek Seed Powder: To make fenugreek seed powder, measure out the desired amount of seeds. 2. Grind the fenugreek seeds into a fine powder. 3. Heat a cup (8 ounces) of water until it's lukewarm. It should be comfortably warm to the touch but not too hot. 4. Once the water is lukewarm, add 1 teaspoon of fenugreek seed powder to it. 5. Use a spoon to stir the fenugreek seed powder into the water until it's fully dissolved, and when the water become slightly cloudy its ready to drink.	
Breakfast 9:00 AM	**Uttapam**	
Lunch 1:00 PM	**Aloo Gobi (Potato and Cauliflower Curry) with Rice** **Ingredients:** 1. Potatoes, cauliflower florets, rice, onion, tomato, ginger-garlic paste, cumin seeds, turmeric, red chili powder, garam masala, oil, salt, water. **Instructions:** 1. Heat oil, add cumin seeds, onion; sauté till golden. 2. Add ginger-garlic paste, tomatoes; cook till soft. 3. Add turmeric, red chili powder, salt; sauté briefly. 4. Add potatoes, cauliflower; mix well. 5. Cover, cook till veggies are tender. 6. Sprinkle garam masala, stir. 7. Serve hot with rice.	
Workout Snack 5:00 PM	**Walnut and Fruit Salad:** **Ingredients:** Chopped walnuts, diced apples, pomegranate seeds, and a squeeze of lemon juice. **Method**: Toss together walnuts and fruits for a refreshing and crunchy salad.	

Day-3

Dinner 9:00 PM	**2 Whole Wheat Parathas With Palak Saag** **+ Tadka Dal** **+ 1 Glass Buttermilk**	



PHARMACOGENOMICS REPORT

GenepoweRx®

PHARMACOGENOMICS

FDA, CPIC & PharmGkb Approved Biomarker

G6PD RISK ASSESSMENT REPORT

G6PD (Glucose-6-phosphate dehydrogenase) deficiency is a prevalent X-linked genetic characteristic with a linked enzyme phenotype. Males are either G6PD deficient or normal, whereas females show a wider range of G6PD deficits, ranging from severe deficiency to normal. Inherited deficiencies of glucose-6- phosphate Dehydrogenase can result in acute hemolytic anemia during times of increased reactive oxygen species (ROS) or free radicals production.

In particular, anti-malarial agents have a strong association with inducing hemolytic anemia in patients with Glucose-6-phosphate dehydrogenase deficiency. Children administered with anti-malarial drugs should specifically be tested for their genotype. Various factors can affect G6PD activity. These are

- G6PD variants – mutations variable stability,
- Age of RBCs – older RBC more vulnerable,
- Anemia (malaria/Fe deficiency),
- Hemoglobinopathies reducing RBC survival,
- Reticulocytes resistance to oxidative stress.

The WHO recommends testing of drugs to predict for risk of hemolysis in G6PD deficient individuals if the drugs are to be prescribed in areas of high prevalence of G6PD deficiency. As a consequence of adverse reactions in individuals with G6PD deficiency, the FDA has introduced warnings or precautions on the drug labeling of primaquine, chloroquine, dapsone, rasburicase, avandaryl tablets (glimepiride + rosiglitazone maleate) and glucovance tablets (metformin + glibenclamide).

G6PD Deficiency Assessment

WHO Classification	No CLASS I Variants Found
PREDICTED PHENOTYPE	
GENOTYPE	
VARIANTS FOUND	Class II, III, IV Variants are not reported.
IMPLICATIONS	No high risk of G6PD deficiency.
CLASSIFICATION OF RECOMMENDATIONS	
CONSIDERATIONS	

Drugs to be taken with caution:

No risk of G6PD deficiency. No reason to avoid drugs based on G6PD status.

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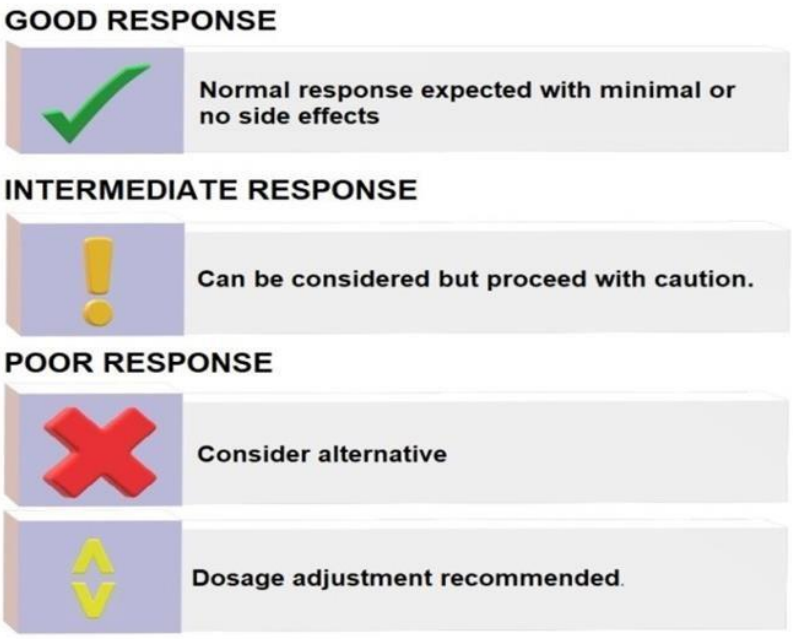
Ghost Y



PHARMACOGENOMIC ANALYSIS AND DRUG RESPONSE STATUS

(FDA, CPIC & PharmGkb Approved Biomarker evaluation)

Result Summary for Clopidogrel & Statins as per CIPIC guidelines (based on CYP2C19, SLCO1B1)

Clopidogrel Phenotype: Intermediate metabolizer

Statins Phenotype (Simvastatin, Lovastatin, Rosuvastatin, Pravastatin, Pitavastatin, Fluvastatin, Atorvastatin): Normal function

1. RESPONSE TO ANTIPLATELET AGENTS

No of variants analyzed: 64
No of gene markers evaluated: 40
Data validated on: 333935 individuals
No of studies evaluated /Supportive evidences (Publications): 358

Good Response	Intermediate Response	Poor Response
Aspirin+Clopidogrel	Aspirin	Clopidogrel, Prasugrel

CPIC Guidelines for Antiplatelet therapy recommendations

(When considering clopidogrel for cardiovascular and neurovascular indications based on CYP2C19 status)

Phenotype	Intermediate metabolizer
Genotype	An individual carrying one normal function allele and one no function allele or one increased function allele and one no function allele
Diplotype	***1/*2**
Implications	Reduced clopidogrel active metabolite formation; increased on-treatment platelet reactivity; increased risk for adverse cardiac and cerebrovascular events
Therapeutic recommendations	Consider an alternative P2Y12 inhibitor at standard dose if clinically indicated and no contraindication.
Classification of Recommendations	Moderate
Other recommendations	Alternative P2Y12 inhibitors not impacted by CYP2C19 genetic variants include ticagrelor and ticlopidine. Prasugrel is contraindicated in patients with a history of stroke or TIA.

To note: *2, *3, *17 are considered due to strong scientific evidence. *4 to *8 alleles of CYP2C19 are not weighed.

2. RESPONSE TO ANTICOAGULANTS

No of variants analyzed: 77
No of gene markers evaluated: 34
Data validated on: 107057 individuals
No of studies evaluated /Supportive evidences (Publications): 223

Good Response	Intermediate Response	Poor Response
Apixaban, Dabigatran, Rivaroxaban	Warfarin	Phenprocoumon

3. RESPONSE TO ANTI - ARRHYTHMIC DRUGS

No of variants analyzed: 14
No of gene markers evaluated: 6
Data validated on: 668 individuals
No of studies evaluated /Supportive evidences (Publications): 15

Good Response	Intermediate Response	Poor Response
--	--	Flecainide, Propafenone

4. RESPONSE TO ANTI - HYPERTENSIVE DRUGS

No of variants analyzed: 151
No of gene markers evaluated: 96
Data validated on: 256237 individuals
No of studies evaluated /Supportive evidences (Publications): 243

Molecule Class	Good Response	Intermediate Response	Poor Response	Evidence not found
Calcium Channel Blockers	Amlodipine	Nifedipine	Verapamil	Felodipine, Nitrendipine, Diltiazem
ARBs	Losartan, Telmisartan	--	Irbesartan, Candesartan	Olmesartan
Vasodilator Drugs	Regadenoson	--	--	Bepridil
Loop Diuretics	--	Torasemide, Furosemide, Bumetanide	Furosemide+ Spironolactone	--
Thiazide Diuretics	--	Hydrochlorothiazide	Chlorthalidone	Indapamide
Potassium Sparing Diuretics	Amiloride, Spironolactone	--	--	Triamterene, Eplerenone
ACE Inhibitors	Enalapril	Benazepril , Imidapril	Quinapril	Fosinopril, Ramipril
Beta Blockers	Bucindolol	Propranolol, Carvedilol, Atenolol, Metoprolol	Timolol	Esmolol, Pindolol, Nadolol

5. RESPONSE TO DIABETIC DRUGS

No of variants analyzed: 60
No of gene markers evaluated: 37
Data validated on: 39284 individuals
No of studies evaluated /Supportive evidences (Publications): 89

Molecule Class	Good Response	Intermediate Response	Poor Response	Evidence not found
Biguanides	Metformin	--	--	--
Sulfonylureas	--	--	Gliclazides, Glibenclamide, Glimepiride, Glipizide, Gliquidone	Glyburide Chlorpropamide Tolazamide Tolbutamide
GLP-1 receptor agonists	--	Liraglutide	--	Exenatide Liraglunatide Lixisenatide Dulagutide Albiglutide Semaglutide
Thiazolidinedione	Rosiglitazone	--	Pioglitazone	--
Glinides	--	Repaglinide	--	Nateglinide
SGLT-2 inhibitors	--	--	--	Canagliflozin, Dapagliflozin, Empagliflozin, Ertugliflozin
DPP4 Inhibitors	--	--	--	Linagliptin, Saxagliptin
Alpha - glucosidase inhibitors	--	--	--	Voglibose, Acarbose, Miglitol

6. RESPONSE TO HMG CoA REDUCTASE INHIBITORS (STATINS)

No of variants analyzed: 77.
No of gene markers evaluated: 48
Data validated on: 334903 individuals.
No of studies evaluated /Supportive evidence (Publications): 227

Good Response	Intermediate Response	Poor Response
Lovastatin, Rosuvastatin , Simvastatin	Atorvastatin	Pravastatin, Pitavastatin , Fluvastatin

CPIC Guideline HMG CoA REDUCTASE INHIBITORS (STATINS)
(Recommended dosing of Statins based on SLCO1B1 phenotype)

Phenotype	**Normal function**
Genotype	An individual carrying two normal function alleles or one normal plus one increased function allele
Diplotype	***1/*1**
Implications	Typical myopathy risk and statin exposure
Dosing recommendations	Prescribe desired starting dose and adjust doses based on disease-specific guidelines
Classification of Recommendations	Strong

7. RESPONSE TO ANTI - CHOLESTEROL MEDs OTHER THAN HMG CoA REDUCTASE INHIBITORS

No of variants analyzed: 16
No of gene markers evaluated: 10
Data validated on: 8509 individuals
No of studies evaluated /Supportive evidences (Publications): 17

Good Response	Intermediate Response	Poor Response
--	Fenofibrate	--

8. RESPONSE TO GASTRO INTESTINAL DRUGS (PROTON PUMP INHIBITORS)

No of variants analyzed: 5
No of gene markers evaluated: 3
Data validated on: 3232 individuals
No of studies evaluated /Supportive evidences (Publications): 10

Good Response	Intermediate Response	Poor Response
Omeprazole, Rabeprazole, Pantoprazole, Esomeprazole, Lansoprazole	--	Corticosteroids (In crohn's disease)

9. RESPONSE TO ANTIEMETICS (NAUSEA AND VOMITING DRUGS)

No of variants analyzed: 6
No of gene markers evaluated: 5
Data validated on: 576 individuals
No of studies evaluated /Supportive evidences (Publications): 10

Good Response	Intermediate Response	Poor Response
Granisetron, Palonosetron	--	--

10.RESPONSE TO PAINKILLERS (ANALGESIC, NSAIDs)

No of variants analyzed: 61
No of gene markers evaluated: 33
Data validated on: 10676 individuals
No of studies evaluated /Supportive evidences (Publications): 77

Good Response	Intermediate Response	Poor Response
Acetaminophen, Desmethyl Naproxen	Fentanyl	Sulindac

11.RESPONSE TO ASTHMA MEDICATIONS

No of variants analyzed: 28
No of gene markers evaluated: 26
Data validated on: 792 individuals
No of studies evaluated /Supportive evidences (Publications): 3

Good Response	Intermediate Response	Poor Response
Methacholine, Glucocorticoids, Aspirin (In Asthmatics)	--	Tiotropium, Flunisolide

12.RESPONSE TO ANTI - INFLAMMATORY DRUGS

No of variants analyzed: 137
No of gene markers evaluated: 90
Data validated on: 76247 individuals
No of studies evaluated /Supportive evidences (Publications): 47

Molecule Class	Good Response	Intermediate Response	Poor Response
Arthritis, Rheumatoid	Leflunomide, Etanercept (Arthritis), Tocilizumab	Adalimumab, Methotrexate, Infliximab	Glucocorticoids , Sulfasalazine, Rituximab, Certolizumab Pegol
Gout	Allopurinol	--	--
Inflammatory Bowel Diseases	Azathioprine, Tacrolimus	--	Cyclosporine, Adalimumab, Desloratadine+ Mizolastine, Thioguanine
Psoriasis	Dimethyl Fumarate	--	Ustekinumab, Etanercept (Psoriasis), Adalimumab

13.RESPONSE TO OPIOIDS

No of variants analyzed: 80
No of gene markers evaluated: 37
Data validated on: 149286 individuals
No of studies evaluated /Supportive evidences (Publications): 156

Good Response	Intermediate Response	Poor Response
Sufentanil, Butorphanol, O-desmethyltramadol, Dexmedetomidine, Tramadol, Tapentadol	Methadone, Alfentanil	Buprenorphine, Oxycodone

14.RESPONSE TO ALZHEIMER'S DRUGS

No of variants analyzed: 6
No of gene markers evaluated: 5
Data validated on: 2907 individuals
No of studies evaluated /Supportive evidences (Publications): 19

Good Response	Intermediate Response	Poor Response
Olanzapine, Risperidone	--	Rivastigmine, Donepezil

15.RESPONSE TO ANTIPSYCHOTIC DRUGS

No of variants analyzed: 80
No of gene markers evaluated: 37
Data validated on: 149286 individuals
No of studies evaluated /Supportive evidences (Publications): 196

Good Response	Intermediate Response	Poor Response
Clozapine, Olanzapine	Risperidone, Lithium	Quetiapine, Aripiprazole

16.RESPONSE TO ANTIDEPRESSANTS

No of variants analyzed: 51
No of gene markers evaluated: 38
Data validated on: 232462 individuals
No of studies evaluated /Supportive evidences (Publications): 569

Good Response	Intermediate Response	Poor Response
Citalopram+ Fluoxetine, Nicotine, Drugs used in Nicotine dependence, Imipramine, Bupropion, Selective serotonin reuptake inhibitors, Mianserin, Varenicline, Amitriptyline, Citalopram, Venlafaxine	Duloxetine, Morphine, Fluvoxamine, Escitalopram, Sertraline, Paroxetine	Agomelatine, Desipramine, Fluoxetine, Mirtazapine, Nortriptyline, Milnacipran, Morphine+ Nortriptyline, Clomipramine

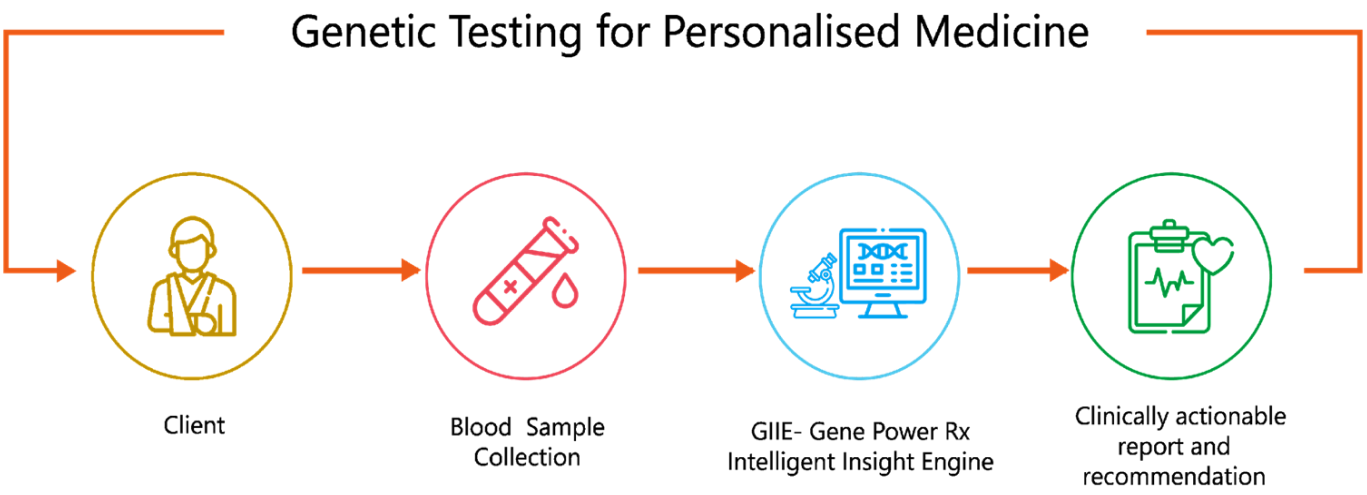
APPENDIX

GLOSSARY OF GENETIC TERMS IN THE REPORT

Variant	A variant refers to genetic alteration or differences in the DNA sequence at a specific position in an individual's genome.
Pathogenic	A variant which is more likely to cause a disease. All the variants which can cause loss of protein function or predictable significant damage to the gene product or can alter protein/protein interactions fall under this category.
Likely Pathogenic	A variant which is strongly suspected to contribute to disease development, but conclusive scientific evidence is currently lacking. Further research is anticipated to substantiate its potential pathogenicity.
Variant of Uncertain significance (VUS)	The variant's classification as pathogenic or benign is uncertain due to limited evidence. Additional patient or family testing, as advised by the clinician, may be needed, and the assessment may change as scientific knowledge evolves.
Risk variant	A variant which can slightly increase the risk of the disease predisposition.
Benign	A genetic change that could be common in the general population. These are not known to cause the disease directly but can have a cumulative effect when multiple benign variants are present in important regions of the genome.
Homozygous	Every person has two copies of same chromosome or DNA. Homozygous mutant refers to having mutated or altered DNA in both the copies of the gene.
Heterozygous	Indicates 2 non identical or mismatched copies of a gene. i.e One normal copy and one mutated copy. The mutations can be Inherited from the parents or acquired (due to various exposures) during one's lifetime.

DESCRIPTION OF TEST

Once the blood sample is collected, the first step is to culture harvest primary progenitor cells and extract genetic material (DNA) from them employing established protocol. The extracted genetic material is then prepped and subjected to sequence analysis. The raw data from the omics is then run through a pipeline of customized in-house tools, processes including our proprietary database. The resultant output data is then cataloged based on the cumulative effect of the different gene variants for each condition. Finally, the report is generated based on medical history, food habits and lifestyle.



RISK ASSESSMENT

GenepoweRx has organized all the variants into a disease specific database and a scoring system is developed, based on the clinical impact of the gene and the type of mutation itself.

To get incisive clinician-patient friendly reports, GenepoweRx has taken an in-house approach for translating genomic information into a measurable score to assess the possibility of predisposition to any specific health condition more accurately. This is accomplished by aggregating the effects of all the genomic variations (SNVs) into a single measure called the “genomic risk assessment score”. Various parameters considered to assess the effect are Zygosity, consequence of the variant, effect on protein structure and function, statistical strength of the variant and so on. Thresholds are set based on Indian data sets that were analyzed and variants are categorized as Mild, Moderate and High Predisposition.

Additional clinical non static measures are also considered. Combined investigation of genomic, clinical and demographic information is thus very scientific method built to predict the probable disease risk and personalize lifestyle and medicine for individuals.

INCLUSION - EXCLUSION CRITERIA

Variants which are reported to have considerable clinical significance such as Pathogenic, Likely pathogenic, risk factor variants, benign variants are screened for each case. Of these, variants with high scientific evidence such as reviewed by expert panel, practice guideline variants and submitted by multiple research groups are alone considered for reporting.

Variants with insufficient evidence or conflicting interpretations, novel variants are not considered for genomic evaluation. Gene variants with no defined criteria, poor evidence and uncertain clinical significance are also not considered for screening. However, in cases with specific clinical condition and existing familial conditions, the inclusion/exclusion criterion is relaxed and all variants are screened for reporting.

VARIANT IDENTIFICATION AND INTERPRETATION

These are critical steps in making genetic diagnosis and personalized medicine a reality. The in-house method of variant classification at GenePoweRx complies with the American College of Medical Genetics (ACMG) standards. We devised standard internal guidelines to assess the robustness of publicly available information, gene-disease relationship, the clinical impact of nucleotide variations, the availability of treatments, and preventive measures. Internal criteria are designed to refine ACMG/AMP guidelines based on the latest data available for assessing the strength of the variant and the most recent information specific to genes & gene-phenotype association. The term variant can be used to describe an alteration that may be benign, pathogenic, or of unknown significance. Though benign variants do not cause the disease directly, they are known to be associated with specific condition. These variants in combination with other potential genetic variants, environmental, and lifestyle factors can trigger the condition.

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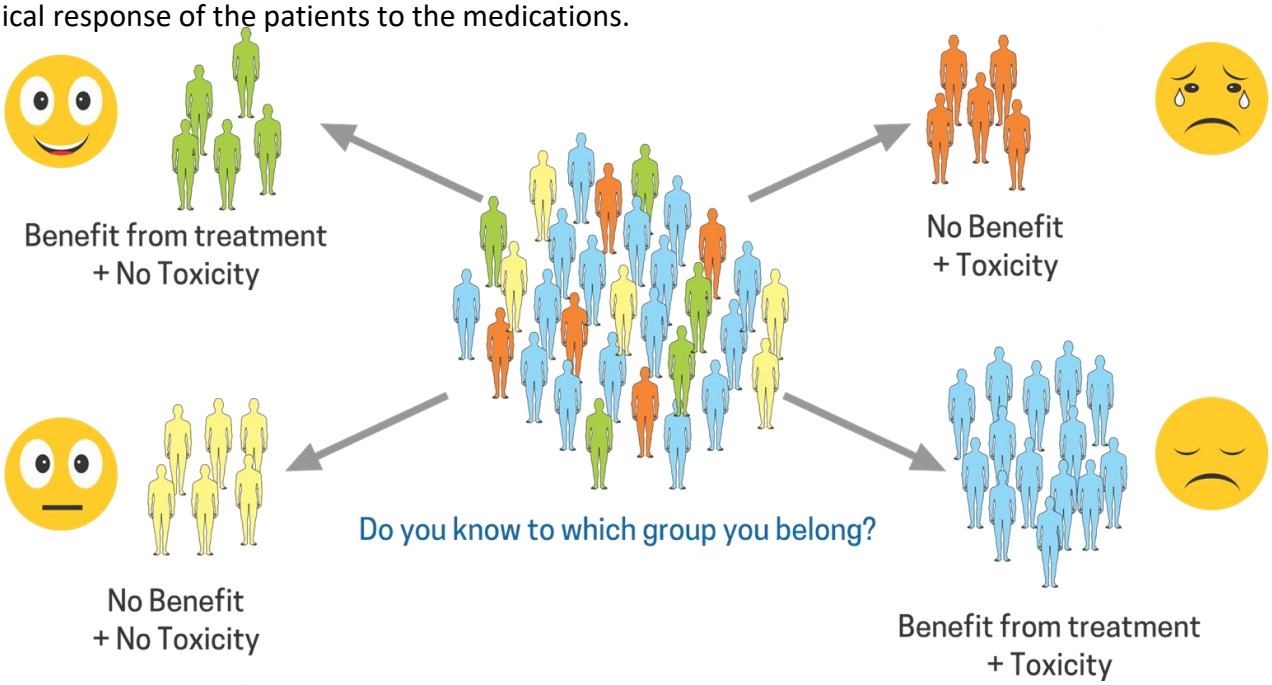
PHARMACOGENOMICS

This test also includes Pharmacogenomics (PGx), which looks for the patient's response to medications. PGx test is intended for use as a directional test for drug decision making explicitly for people with various metabolic disorders, cardiac conditions and can be extended to the general population for precision medicine. This test will aid in understanding the patients' probable reaction to specific medicines with the approved therapeutic product labeling and helps in choosing the right drug to prevent treatment failure and side effects.

PGx at GenepoweRx is done with next generation sequencing (NGS) based qualitative diagnostic test that utilizes innovative bead-based chemistry followed by exome capture technology and sequenced at a higher depth.

At GenepoweRx, collective information of altered & healthy DNA is considered to evaluate response to particular drugs. The genomic data is processed through standard bioinformatics tools, evaluated by various in-house pipelines and curated database information. Genomic information backed by scientific evidence from clinically and statistically significant studies is further considered for explicit personalized pharmacogenomics report preparation.

CPIC & PharmGkb approved pharmacogenetic biomarkers with significant genotype-phenotype relationship (from 1294 SNVs of 494 genes) are evaluated for FDA approved drugs, for assessing the clinical response of the patients to the medications.



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INTERPRETATION METHODOLOGY OF GENEPOWERX® COMPREHENSIVE GENOMICS TEST

1. Value allocation to disease specific variants includes the assignment of values based on 10 different attributes of the variant, to derive a single score for each variant.
2. Normalization of the variant scores based on the disease conditions
3. The statistical significance is calculated and the ambiguous SNPs which do not attain the statistical significance are excluded. [statistics used: (t tests and subsequent Z scores (for determining the importance of each component when predicting this set))]
4. Analyze data statistically to derive FDR values and eliminate the false positives according to the FDR threshold
5. Identify cumulative frequency for final risk estimation
6. Repeat the steps for Molecular scoring and Physician clinical score.

Test description & Methodology

Variants associated with the reported clinical condition are analyzed by Whole Exome Sequencing (WES) using the Next Generation Sequence (NGS) analyzer – Illumina's NovaSeq 6000. The DNA libraries were prepared using Roche KAPA hyper Exome panel and sequenced to a mean depth of >80-100x coverage. The panel works on a principle of probe hybridization using DNA probes of around 120mer length providing high and specific interactions with target DNA molecules along with - dual molecular barcodes with dual sample indexing for library preparation. The usage of these dual molecular barcodes reduces or eliminates the index hopping and false positives thus making it more reliable. The NGS test is performed with >95% coverage at a higher depth. Recommended read length of 2 x 150 bps is considered for WES panel sequencing and paired end reads are considered for analysis. Huge quantities of NGS based omics data is concised for better clinical guidance. The raw data with Q30≥80% is considered for analysis, checked for quality by fastQC and aligned to the human reference genome GRCh38.

Variant identification and interpretation are critical steps in making genetic diagnosis and personalized medicine a reality. GATK guidelines are followed for variant identification. The variants were annotated and filtered using the in-house and commercial analysis workflows. Interpretation of sequence variants strictly adheres to the latest ACMG guidelines. In the assessment of the variant classification, GenepoweRx considers information and evidence that includes, but is not limited to, the following 5 significant parameters. The functional impact of the gene in causing the disease phenotype, functional impact of variation in the gene product based on in silico, in vitro, and in vivo studies, variant-disease association, prevalence and significance. This includes comparison against the gnomAD population catalog of variants in 123,136 exomes, the 1000 Genomes Project Consortium's publication of 2,500 genomes, the NCBI ClinVar database of clinical assertions on variant's pathogenicity and multiple lines of computational evidence on conservation and functional impact. The test results are then carefully reviewed and manually curated by our team of highly trained and experienced genome analysts.

Variant Assessment Process

The following databases and algorithms are used to annotate and evaluate the impact of the variant in the context of human disease: 1000 genomes, gnomAD, ClinVar, OMIM, dbSNP, NCIB RefSeq Genes, ExAC Gene Constraints, VS-SIFT, VS-PolyPhen2, PhyloP, GERP++, GeneSplicer, MaxEntScan, NNSplice, PWM Splice Predictor. Analysis was reported using HGVS nomenclature (www.hgvs.org/mutnomen) as implemented by the VarSeq transcript annotation algorithm. The reported transcript matches that used most frequently by the clinical labs submitting to ClinVar.

Variant Impact (From Ensembl)

Indicates the strength of the variant based on a variant's probability to alter protein structure or function and its effect on the phenotype. The variant Impact is obtained from Ensembl, which is calculated by a rule based approach to predict the effects that each allele of the variant may have on each transcript. The set of consequence terms, defined by the Sequence Ontology (SO), that can be currently assigned to each combination of an allele and a transcript is shown in the table below. Note that each allele of each variant may have a different effect in different transcripts. Variant Impact is classified into 4 groups

High:
This variant is assumed to have (disruptive) impact in the protein, probably causing protein truncation, loss of function or triggering nonsense mediated decay. Variant types included under this category: Transcript ablation-complete deletion of functional region of the gene; Splice acceptor variant; Splice donor variant; Stop gained; Frameshift variant; Stop lost; Start lost; Transcript amplification.

Moderate:
A non-disruptive variant which can change protein effectiveness. Variant types included under this category: Inframe insertion; Inframe deletion; Missense variant; Protein altering variant.

Low:
Assumed to be mostly harmless and unlikely to change protein behavior. Variant types included under this category: Splice region variant; Incomplete terminal codon variant; Start/Stop retained variant; Synonymous variant.

Modifier:
Usually non-coding variants where predictions are difficult or there is no sufficient evidence of impact. Variant types included under this category: Mature miRNA variant; UTR region Variants, TF binding site variant; Regulatory region variant, Intergenic variant, and variants of many other location where's there's no sufficient evidence.
Ref: https://asia.ensembl.org/info/genome/variation/prediction/predicted_data.html

ACMG variant classification guidelines (PMID: 25741868)

	Benign		Pathogenic			
	Strong	Supporting	Supporting	Moderate	Strong	Very Strong
Population Data	MAF is too high for disorder *BA1/BS1* **OR** observation in controls inconsistent with disease penetrance *BS2*			Absent in population databases *PM2*	Prevalence in affecteds statistically increased over controls *PS4*	
Computational And Predictive Data		Multiple lines of computational evidence suggest no impact on gene /gene product *BP4* Missense in gene where only truncating cause disease *BP1* Silent variant with non predicted splice impact *BP7*	Multiple lines of computational evidence support a deleterious effect on the gene /gene product *PP3*	Novel missense change at an amino acid residue where a different pathogenic missense change has been seen before *PM5* Protein length changing variant *PM4*	Same amino acid change as an established pathogenic variant *PS1*	Predicted null variant in a gene where LOF is a known mechanism of disease *PVS1*
Functional Data	Well-established functional studies show no deleterious effect *BS3*		Missense in gene with low rate of benign missense variants and path. missenses common *PP2*	Mutational hot spot or well-studied functional domain without benign variation *PM1*	Well-established functional studies show a deleterious effect *PS3*	
Segregation Data	Non-segregation with disease *BS4*		Co-segregation with disease in multiple affected family members *PP1*	Increased segregation data →		
De novo Data				*De novo* (without paternity & maternity confirmed) *PM6*	*De novo* (paternity & maternity confirmed) *PS2*	
Allelic Data		Observed in *trans* with a dominant variant *BP2* Observed in *cis* with a pathogenic variant *BP2*		For recessive disorders, detected in *trans* with a pathogenic variant *PM3*		
Other Database		Reputable source w/out shared data = benign *BP6*	Reputable source = pathogenic *PP5*			
Other Data		Found in case with an alternate cause *BP5*	Patient's phenotype or FH highly specific for gene *PP4*			

Criteria to Classify Sequence Variants

Pathogenic **1 Very Strong (PVS1) AND** **≥1 Strong (PS1–PS4) OR** **≥2 Moderate (PM1–PM6) OR** **1 Moderate (PM1–PM6) and 1 Supporting (PP1–PP5) OR** **≥2 Supporting (PP1–PP5)** **≥2 Strong (PS1–PS4) OR** **1 Strong (PS1–PS4) AND** **≥3 Moderate (PM1–PM6) OR** **2 Moderate (PM1–PM6) AND ≥2 Supporting (PP1–PP5) OR** **1 Moderate (PM1–PM6) AND ≥4 Supporting (PP1–PP5)**	**Likely Pathogenic** **1 Very Strong (PVS1) AND 1 Moderate (PM1–PM6) OR** **1 Strong (PS1–PS4) AND 1–2 Moderate (PM1–PM6) OR** **1 Strong (PS1–PS4) AND ≥2 Supporting (PP1–PP5) OR** **≥3 Moderate (PM1–PM6) OR** **2 Moderate (PM1–PM6) AND ≥2 Supporting (PP1–PP5) OR** **1 Moderate (PM1–PM6) AND ≥4 Supporting (PP1–PP5)**
Benign **1 Stand-Alone (BA1) OR ≥2 Strong (BS1–BS4)**	**Likely Benign** **1 Strong (BS1–BS4) and 1 Supporting (BP1–BP7) OR ≥2 Supporting (BP1–BP7)**

*Variants are classified as Uncertain Significance if other criteria are unmet or the criteria for benign and pathogenic are contradictory.

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DISCLAIMER

1. Genetic testing using the methods applied at K&H is expected to be highly accurate.
2. Normal findings do not rule out the diagnosis of a genetic disorder since some genetic abnormalities may be undetectable with this test.
3. This test cannot reliably detect mosaicism. Some genes have inherent sequence properties (for example: repeat, homology, or pseudogene regions, high GC content, rare polymorphisms) that may result in suboptimal data, and variants in those regions may not be reliably identified.
4. This report and the usefulness of the information provided in the report shall not warranty or hold any liability or responsibility for any direct, indirect, incidental, consequential indemnities arising out of the use of, or inability to use the information. This doesn't hold any claims for medico-legal reasons also.
5. This report makes no promises or guarantees that the reported condition/s would develop anytime. Other genetic, environmental, and clinical factors might influence the patients' phenotypic response to the condition. Normal findings do not rule out the diagnosis of a genetic disorder since some genetic abnormalities may be undetectable with this test.
6. False negative results may also occur in the setting suboptimal DNA quality. The chance of a false positive or false negative result due to laboratory errors incurred during any phase of testing cannot be completely excluded. The mutations are not validated/reflex tested.
7. Interpretations are made with the assumption that any information provided on family relationships is accurate. Consultation with a genetics professional is recommended for better interpretation of results. Interpretation of this result should be done in context of a patient's medical record, family history and biochemical profile. Please note that interpretation and classification of the variants reported here may change overtime.
8. Collection, processing, use, storage and retention of the anonymized data, the sample collected and related anonymized reports from the tests conducted for ongoing test developments, educational, scientific research and/or other related activities.
9. Reflex testing by MLPA or multiplex PCR is recommended to confirm the presence of reported CNVs.
10. Please note that only a few key biomarkers are mentioned here, although the analysis was conducted on all biomarkers relevant to the condition of interest. The complete list is available upon request.

This test was developed, and its performance characteristics were determined by GenepoweRx. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. These interpretations are based on ACMG/AMP GATK, CPIC Guidelines. This test can be used for clinical screening and research purposes only. The test is registered in CDSCO, India, as Uppalu-Hyder-TE/M/IVD/008407, Class C of Medical Devices, 2022.

Report content approved by:

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Disclaimer of Liability

This report is provided as an information source for clinicians and is not intended it to be considered a substitute for professional medical advice. If you have or suspect you have a medical problem, contact your physician for personalized treatment or therapy. The translation of genomics knowledge and data into the current report requires generalization and continuous expansion of genomic insights from literature. Hence, minor errors are anticipated. This genomic report and the usefulness of the information provided in the report shall not warranty or hold any liability or responsibility for any direct, indirect, incidental, or consequential indemnities arising out of the use of or inability to use the information. This report makes no promises or guarantees that the reported condition/s will develop at any time. Other genetic, environmental, and clinical factors might influence the patients' phenotypic response to the condition. Proper understanding of the risks aids in better management of metabolic conditions with traditional therapies or other treatment options and helps in the prevention/delay of the disease or makes it less harmful.

Genetic testing plays a key role in the diagnosis of the root cause of a disease or condition. It provides excellent guidance in deciding the right medical regimen. But sometimes, a few non-treatable variants are also identified. Not all genetic changes affect health. It is difficult to know whether identified variants are involved in the condition of interest. Sometimes, an identified variant is associated with a different genetic disorder that has not yet been diagnosed (these are called incidental or secondary findings). A finding of biomarker alteration does not necessarily indicate the pharmacologic effectiveness (or lack thereof) of any drug or treatment regimen; a finding of no biomarker alteration does not necessarily indicate the lack of pharmacologic effectiveness (or effectiveness) of any drug or treatment. No Guarantee of Clinical Benefit: This Report makes no promises or guarantees that a drug will be effective in the treatment of disease in any patient. This Report also makes no promises or guarantees that a drug with a potential lack of clinical benefit will, in fact, provide no clinical benefit. It is possible that a pathogenic variant is present in a gene that was not selected for analysis and /or interpretation in cases where insufficient phenotypic information is available. Due to inherent technology limitations and constant upgradation of research and literature, not all bases of the exome can be covered. Accordingly, variants in regions of insufficient coverage may not be identified and/or interpreted. Therefore, it is possible that pathogenic variants are present in one or more of the genes analyzed but have not been detected. The variants not detected by the assay that was performed may impact the phenotype. For in vitro research use only. This test must be ordered by a qualified medical professional in accordance with required medical regulations.

Patient care treatment decisions must be based on the self-determining medical judgment of the respective physician. Do consider complete patient information such as patient preferences, medical history and family history, physical examination profiles, and other lab results per the standard of care medical practice. The reported results are for the information of the referring doctor only. It should be noted that this test is restricted to a limited number of genes and does not include all intronic and non-coding regions. This report only includes variants that meet a level of evidence threshold for cause or contribute to disease. More evidence for disease association of genes and causal pathogenic variants is discovered every year, and it is recommended that genetic variants are re-interpreted with updated software and annotations periodically.

ISO: TE/R-009

Meet the doctors

Dr. Kalyan Uppaluri is the cofounder and the owner of K&H Personalized Medicine Clinic and Research Institute. He did his medical training at the prestigious Gandhi Medical College. He then moved to the United States, where he specialized in Internal Medicine at the McLaren Hospital, Michigan. He also got a degree in Medical Genomics from Ivy league institute, Stanford University and pursued cancer research at Wayne State University, Michigan.

Dr. Hima Challa graduated from Gandhi Medical College and was among the top few in her batch. She specialized in Internal Medicine at St. Joseph Mercy Oakland, Michigan in the United States. She graduated in Medical Genomics from the Ivy league institution of Harvard Medical School. She also holds a master's in nutrition science from the Texas Women University and in integrative medicine from the Arizona University.

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