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METAGEN

Your MetaGen Mutations Panel Report

Your Barcode:

Date:

Testing Laboratory:
HKG epiTherapeutics Limited

Your Information

Name:	
ID:	
Receive Date:	
Report Date:	
Results Reviewed By:	Sharon Luk, PhD, Technical Supervisor
Results Approved By:	Carlos Ruiz, MD, Laboratory Director

Your Results

Gene	SNP	Reference	Alternate	Your Genotype
AHCY	rs121918607	C	T	
AHCY	rs121918608	T	C	
COMT	rs4680	G	A	
MTHFR	rs1801133	G	A	
MTHFR	rs1801131	T	G	
MTR	rs1805087	A	G	
MTRR	rs1801394	A	G	
MTRR	rs1532268	C	T	

SNP (Single Nucleotide Polymorphism):

Refers to a common variation in a single nucleotide that occurs at a specific position in the genome, distinguishing one genome from another.

Reference Allele:

This is the common allele (version of a gene) found in the general population.

Alternate Allele:

This represents a different version of the gene that varies from the common type.

Your Genotype:

Indicates your specific alleles for each gene. '+/+' denotes both alleles are altered, '+/-' indicates one altered allele, and '-/-' means no mutations are found in either allele. This column reflects your unique genetic makeup as revealed by the tested SNPs.

qrcode

Have Some Questions?

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MetaGen Report V2.0
Bioinformatics Analysis: Metagen V .0

Introduction

Understanding DNA Methylation, SNPs, and Your Health

Welcome to the fascinating world of DNA methylation and Single Nucleotide Polymorphisms (SNPs), crucial components of our body's genetic regulation. Imagine DNA as a sophisticated computer in each cell, with DNA methylation acting like versatile software. This process adds chemical marks to our DNA, influencing whether a gene is active or silenced, ensuring the correct functioning of our body.

DNA methylation is vital as it enables the same gene to have different roles in various cells. In essence, it is like a binary code, programming our DNA to ensure our body functions correctly. Each cell type has a unique combination of active and silent genes, orchestrated by DNA methylation. This orchestration is crucial for our health and well-being, allowing cells to perform their specific functions while maintaining the overall harmony of our body's systems. This test provides genetic risk information based on the assessment of specific genetic variants but does not report on a user's entire genetic profile. This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease.

There are other companies offering a genetic risk test that may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company. Other factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.

Beyond its role in cellular function, DNA methylation is tightly associated with our biological age, which is suggested by several studies to be a more accurate measure of health than chronological age. Certain alterations in DNA methylation patterns can lead to disease development, as research has shown that these changes can transform healthy tissues into diseased ones, impacting conditions like cancer. Furthermore, environmental factors, including diet and maternal care, can influence DNA methylation, highlighting its dynamic nature and the interplay between genetics and lifestyle.

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Introduction

Understanding DNA Methylation, SNPs, and Your Health

Our report delves into what is known in the literature about the intricate relationship between DNA methylation, genetic mutations, and health, providing insights that are crucial for understanding your personal health journey. By exploring these genetic and epigenetic interactions, we offer a comprehensive view of how your unique genetic makeup and lifestyle choices impact your overall health and aging process.

In tandem with the intricate processes of DNA methylation, Single Nucleotide Polymorphisms, or SNPs (pronounced 'snips'), represent another crucial aspect of our genetic landscape. SNPs are the most common type of genetic variation among people, occurring at specific positions within the DNA sequence. These small yet significant variations contribute to the unique traits that make each individual distinct, such as variations in eye color. But their impact extends beyond physical attributes. SNPs can significantly influence our health, including our susceptibility to certain diseases and how we respond to various medications.

Introduction

Understanding Your Unique Genetic Makeup: The Role of SNPs

What are SNPs?

Think of your DNA as a long string of letters, each representing a building block of your genetic code. Sometimes, these letters can differ slightly between individuals. These tiny differences are what we refer to as Single Nucleotide Polymorphisms.

A Common Type of Genetic Variation

SNPs are the most frequent type of genetic variation in the human genome. To visualize this, imagine you and a friend have nearly identical recipes for a cake, but in one recipe, a single ingredient is slightly different. This minor difference can change the cake's flavor, just as SNPs create small variations in our genetic 'recipes.'

Why are SNPs Important?

- **Unique Traits:** SNPs are a key factor in determining our individual traits, such as eye color or hair texture.
- **Health Connections:** Certain SNPs can influence how our bodies react to medications or increase our risk of developing specific health conditions.
- **Ancestral Insights:** These genetic variations are also tools for scientists to trace human history and understand population migrations.

SNPs in Our Health Report

- **Focus on Health-Related Genes:** In this report, we specifically analyze SNPs in genes crucial for your body's methylation processes—chemical reactions vital for many bodily functions.
- **Personal Health Insights:** By examining these SNPs, we offer insights into your health risks, potential for certain conditions, and how your body might react to changes in diet or lifestyle.
- **A Personalized Guide to Your Health:** Understanding your SNPs is akin to having a personalized guide to your body. This knowledge, combined with what we learn from studying your DNA methylation, empowers you with detailed information for managing your health and wellness more effectively.

Together, the study of DNA methylation and SNPs provides a comprehensive picture of how our genetic and epigenetic makeup interacts with our environment, lifestyle, and health. This report aims to bring these insights to you in a clear and actionable way, helping to guide your personal health decisions.

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Introduction

The MetaGen Mutations Panel: A Comprehensive Genetic Analysis

Following our exploration of DNA methylation and SNPs, we now introduce the MetaGen Mutations Panel. This report presents a comprehensive analysis of your genetic methylation test results, focusing on key mutations in the AHCY, COMT, MTHFR, MTR, and MTRR genes.

Understanding Gene Functions and Implications

- **Mutation Analysis:** The report reviews the implications of mutations in these genes, drawing from the latest scientific literature.
- **Gene Functions:** We discuss the primary functions of each gene, shedding light on how they contribute to your body's methylation processes and overall health.

Recommendations and Consultations

- **Dietary Modifications:** Based on the literature, we provide a summary of what the current literature recommendations for dietary modifications that might be beneficial considering your genetic profile.
- **Consulting Healthcare Professionals:** While we offer insights and suggestions based on published literature, for personalized medical advice, it is essential to consult your physician or dietician. This report is intended to provide some background, not replace, professional medical consultation.

An Integrated Approach for Holistic Understanding

Our integrated approach in the MetaGen Mutations Panel report aims to provide some information on your genetic predispositions and what is currently known about their possible influence on your health and aging process. By combining genetic analysis with epigenetic assessment, we offer a view of factors that might be contributing to your well-being.

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Introduction

Kit Contents

The following materials are included in your saliva collection kit:
1. Saliva Collection Device: A container for collecting the saliva sample, marked to indicate the required 2ml volume.
2. Transportation Medium: A liquid medium that needs to be poured into the saliva collection device to preserve the sample.
3. Funnel: Attached to the collection device for easy transfer of saliva.
4. Cap for the Collection Device: Used to securely seal the collection tube after adding the transportation medium.
5. Biohazard Bag: For placing the sealed collection tube for safe transportation.
6. Instructions Card: Provides detailed steps for collecting the saliva sample, handling the transportation medium, and sealing the tube.

Methodology

Saliva Collection and Stabilization: The collected saliva sample is preserved using a stabilization buffer to ensure the integrity of the DNA during transportation to the laboratory.

DNA Extraction: Once the sample arrives at the laboratory, DNA is extracted from the saliva using an automated system. This process isolates the DNA from other cellular materials.

PCR Amplification: The extracted DNA undergoes Polymerase Chain Reaction (PCR) amplification, targeting specific regions associated with single nucleotide polymorphisms (SNPs) in the following five genes:
- AHCY
- COMT
- MTHFR
- MTR
- MTRR

Next-Generation Sequencing (NGS): The amplified DNA is then sequenced using the MiSeq Next Generation Sequencing platform. This technology allows for the high-throughput analysis of the SNPs, providing detailed genotype information for each variant.

Data Analysis: The sequencing data is processed through a bioinformatics pipeline on AWS (Amazon Web Services). This pipeline analyzes the genetic data, determining the genotype of the SNPs.

This comprehensive methodology ensures accurate and reliable genetic information, which is then compiled into a detailed report for the user.

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Mutation Analysis, Gene Functions, and Recommendations

1 rs121918607 and rs121918608

AHCY Gene Mutations

AHCY, SAM, and SAH in the Methylation Cycle:

AHCY (Adenosylhomocysteinase) is an enzyme that plays a crucial role in breaking down S-adenosylhomocysteine (SAH), a by-product of methylation processes, back into homocysteine. This functions is a key part of methionine metabolism (Baric et al., 2004), helping to maintain the balance of SAM and SAH in the body (Cooper et al., 2006; Motzek et al., 2016). Such balance is essential for the proper functioning of various biochemical processes, including DNA repair, neurotransmitter synthesis, and detoxification (Vizan, Di Croce, & Aranda, 2021).

Mutation Details and Health Implications:

rs121918607 (TRP112TER) Mutation:

Type: SNP where cytosine (C) is replaced by thymine (T).

Frequency: This genetic variation is rare, occurring in about 0.2% of the global population.

Clinical Significance:

This mutation is associated with Hypermethioninemia (Baric et al., 2004), a condition characterized by high methionine levels in the blood, leading to various health issues (Mudd, 2011).

rs121918608 TYR143CYS Mutation:

Type: This is a SNP where thymine (T) is replaced by cytosine (C).

Frequency: The variation is very rare, observed more frequently in European populations but remains uncommon overall.

Clinical Significance:

The mutation is considered pathogenic or likely pathogenic, being associated with conditions such as Hypermethioninemia—a metabolic disorder marked by elevated methionine levels, leading to potential neurological issues—and rhabdomyolysis, a serious condition caused by the breakdown of muscle tissue that can lead to kidney damage. This mutation results in reduced enzyme activity, significantly affecting methionine metabolism.

We suggest that you consult your health provider as to the health style changes that might be required.

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rs121918607 and rs121918608

AHCY Gene Mutations

Take Action:

This guidance is what the literature is suggesting for individuals with either of the AHCY gene mutations rs121918607 or rs121918608.

Enhance B Vitamin Intake: Emphasize the importance of B vitamins (B6, B12, and folate) due to their crucial role in the methylation process. This process is vital in managing the impacts of these mutations on methionine metabolism.

B Vitamin Dosage Guidelines:
For Vitamin B12, refer to the NIH guidelines here.
For Vitamin B6, consult the NIH guidelines here.
For Folate, guidelines can be found here.

Beneficial Foods:

Incorporate foods high in B vitamins into your diet, such as leafy greens, eggs, and whole grains. These foods can naturally support your body's methylation processes.

Interpretation:

This dietary enhancement with B vitamins is advised for individuals with the specified AHCY gene mutations. B vitamins are key in supporting methylation processes (Selhub, Bagley, Miller, & Rosenberg, 2000), essential for mitigating disruptions in methionine metabolism associated with these mutations.

Remember, this advice is based on current literature and is tailored to those with these specific genetic mutations. The effectiveness and need for such dietary changes may vary for individuals without these mutations.

Important Note:

Consult with healthcare professionals or a genetic counselor for personalized advice, especially before making significant changes to your diet or starting new supplements. This is crucial in the context of managing health with genetic mutations.

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2 COMT Gene Mutation

Rs4680 (Val158Met)

COMT Gene Role

Function: The COMT (Catechol-O-Methyltransferase) gene plays a critical role in the metabolism of catecholamines, including neurotransmitters like dopamine, epinephrine, and norepinephrine (Tunbridge, Bannerman, Sharp, & Harrison, 2004). This metabolic process is essential for maintaining balanced neurotransmitter levels in the brain, impacting various aspects of neurological and mental health.

Mutation Details and Health Implications:

Type: This is a SNP where guanine (G) is replaced by adenine (A).

Frequency: This genetic variation is relatively common, occurring in approximately 36.921% of the global population. The frequency varies by population:

- European: Approximately 50.9%
- African: Approximately 30.4%
- Asian: Approximately 26.5%
- Latin American: Approximately 41.6%

Metabolic Impact:

This variant can lead to differences in dopamine metabolism, which may affect neurological and psychiatric health (Sundermann et al., 2015).

Take Action:

Dietary Recommendations: The literature recommends a diet enriched with omega-3 fatty acids and antioxidants to support neurological health, as highlighted in Artemis P. Simopoulos's study (Simopoulos, 2011). These nutrients are vital for brain function and may help in balancing neurotransmitter activity.

- Sources of Omega-3s: To support brain function, the literature advises to include sources of Omega-3s such as fish, nuts, and seeds in your diet, as they are suggested by the literature to be beneficial for neurological health. For more detailed information on Omega-3 fatty acids and their sources, you can refer to the NIH's resource.
- Sources of Antioxidants: Incorporate berries and dark chocolate for their reported beneficial effects on brain health (Shukitt-Hale, Lau, & Joseph, 2008; Tokede, Gaziano, & Djousse, 2011).

Important Note:

It's important to discuss any health concerns or symptoms, particularly those related to neurological conditions or medication responses, with a healthcare provider.

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rs1801133 (C677T) and rs1801131 (A1298C)

MTHFR

MTHFR Gene Role:

The MTHFR gene, short for Methylenetetrahydrofolate Reductase, plays a pivotal role in your body's handling of folate or vitamin B9. This gene is involved in converting folate into a form your body can effectively use, impacting vital functions like DNA creation and cell repair. Understanding how this gene operates can offer insights into your overall health, particularly in how your body utilizes this essential vitamin.

Folate, a critical B-vitamin, is naturally present in a variety of foods. It's vital for DNA synthesis and cell division. According to the literature different life stages have varying recommended amounts of folate, measured in micrograms (mcg) of dietary folate equivalents (DFEs). For instance, adults typically need 400 mcg DFE, while pregnant women require 600 mcg DFE. This literature recommended intake can be met through a balanced diet including foods like liver, green vegetables, nuts, and beans, as well as fortified bread and cereals.

Additionally, folate is available in supplements, commonly as folic acid or methylfolate (5-MTHF). The latter might be more suitable for individuals with certain gene variants like MTHFR C677T, as their bodies can utilize this form more efficiently. However, those who could become pregnant are advised to consume folic acid, not 5-MTHF, even with an MTHFR C677T gene variant, due to its effectiveness in preventing neural tube defects in babies.

For more comprehensive information about folate, its recommended intake, sources, and supplements, you can refer to the U.S. Department of Health & Human Services, National Institutes of Health, Office of Dietary Supplements.

Mutation Details and Health Implications:

Impact on Folate Metabolism: Mutations in the MTHFR gene can lead to reduced efficiency in folate metabolism (Ozarda, Sucu, Hizli, & Aslan, 2009), which might affect various health aspects, particularly those related to cellular growth and repair.

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rs1801133 (C677T) and rs1801131 (A1298C)

MTHFR

rs1801133 (C677T) Mutation:

Type: This is a SNP where guanine (G) is replaced by either adenine (A) or cytosine (C).
Frequency: The frequency of the rs1801133 SNP for the G>A variation is common, with global frequencies around 30-35%. However, the frequency of the G>C variation is not well-established and is likely to be significantly lower, indicating it is a much rarer occurrence in the general population.

Clinical Significance:

Associated with methotrexate response and potentially impacts drug metabolism.
Population Frequency: Varied frequency across populations, generally higher in Europeans and lower in African and Asian groups.

Take Action:

For individuals with the MTHFR C677T mutation, the NIH Office of Dietary Supplements recommends:

- **Folate-rich Foods:** Include leafy greens like spinach, fruits like oranges, legumes such as peanuts and beans, and liver in your diet. These foods are naturally high in folate.
- **Fortified Foods:** Consume foods fortified with folic acid, such as enriched bread, cereals, flour, and pasta, to help meet your daily folate needs.
- **Supplement Consideration:** Methylfolate (5-MTHF) supplements might be more effective for those with the mutation, but women of childbearing age should focus on folic acid intake.
- **Professional Consultation:** Always seek advice from healthcare professionals, especially for dietary changes or supplementation related to the MTHFR mutation.

For more information, refer to the NIH Office of Dietary Supplements.

rs1801131 (A1298C) Mutation:

Type: This is a SNP where thymine (T) is replaced by guanine (G).
Frequency: Relatively common, with minor allele frequencies around 25-29% globally.

Clinical Significance:

It has conflicting interpretations; linked to Homocystinuria (Price, Wilcock, & Weekman, 2018), Neural tube defects (folate-sensitive) (Zhang et al., 2019), and more. While some studies have shown an association between the A1298C polymorphism and NTDs (Abbas, Sifi, Benembarek, & Abadi, 2021; Aranda-Sánchez et al., 2021; Fu, Wang, & Kong, 2017; Isotalo, Wells, & Donnelly, 2000; Put et al., 1998; Yaliwal & Desai, 2012), others have not found a significant link (Das, 2014).

A1298C maybe associated with increased homocysteine level (Kumar et al., 2005). The C677T variant in the *MTHFR* gene is known to affect the enzyme's function, often leading to higher levels of homocysteine and reduced folate levels. However, the implications of the A1298C variant are less clear-cut. This variant tends to have a milder impact on the enzyme's activity. Typically, individuals with the A1298C variant maintain normal levels of homocysteine and plasma folate (Yamada, Chen, Rozen, & Matthews, 2001).

Take Action:

Conflicting results and controversies regarding A1298C mutation significance indicate the need for further research to elucidate its precise role and implications for dietary recommendations (Botto & Yang, 2000; Ghaznavi, Zahra, Shahram, & Soltanpour, 2015; Jiang, Liu, Townsend, & Wang, 2023; Kim et al., 2015; B. J. Wang et al., 2015; Zhu & Li, 2016)

Important Note:

For a comprehensive understanding of how these mutations might affect individual health, it's important to consult healthcare professionals. They can provide tailored advice and recommendations based on one's genetic makeup.

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rs1805087 (A2756G)

MTR

MTR Gene Role:

Function: The MTR (Methionine Synthase) gene plays a pivotal role in the conversion of homocysteine to methionine, a critical step in the methylation cycle (Ding, Zhou, Jiang, & Lu, 2013). Methionine synthesis is essential for DNA synthesis and the overall functioning of cellular processes (Maddocks, Labuschagne, Adams, & Vousden, 2016).

Mutation Details and Health Implications:

Type: This is a SNP where adenine (A) is replaced by guanine (G).

Frequency: About 21.8% of people worldwide have a specific variation of a gene.

Population-Specific Frequencies:

· European: ~18.8%
· African: ~26.5%
· Asian: ~11.9%
· Latin American: ~19.8%

Clinical Significance according to the literature:

- Homocysteine and Methionine Levels: Individuals with the MTR A2756G mutation may have altered homocysteine and methionine levels, is believed to be risk factor for various health conditions, including cardiovascular diseases (Biselli et al., 2009; Leclerc et al., 1996; Paradkar, Padate, Vora, & Ashavaid, 2019).
- Vitamin B12 Metabolism: The MTR gene's function is also closely linked with vitamin B12 metabolism. Mutations in this gene, including A2756G, can potentially affect the efficiency of vitamin B12 use in the body(Raghubeer & Matsha, 2021).
- Variable Expression: The effects of the MTR A2756G mutation can vary widely among individuals. Diet, lifestyle, and other genetic factors can influence how this mutation manifests in each person (Wen-Xing Li, Dai, Zheng, Liu, & Huang, 2015).
- Folate Usage: Although the exact impact of the A2756G mutation on how the body uses folate isn't fully clear, it's involved in processes that are linked with folate. This means it might have some indirect effects on folate in the body, especially in how it relates to homocysteine, a substance that folate helps process (W. X. Li et al., 2017).

While there are various interpretations, the predominant view suggests a benign nature of this mutation (Y. Wang, 2013).

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rs1805087 (A2756G)

MTR

Take Action:

B Vitamin Intake: Given the role of the MTR gene in homocysteine and methionine metabolism, and its connection to vitamin B12, maintaining adequate levels of B vitamins, particularly vitamin B12, may be beneficial. This aligns with general recommendations from the NIH Vitamin B12 Health Professional Fact Sheet, which emphasizes the importance of vitamin B12 for health, including its role in methionine synthesis and homocysteine metabolism.

Dietary Sources: Include foods rich in B12 such as meat, poultry, fish, eggs, and dairy products, along with fortified grains and cereals, in your diet.

Monitoring and Consultation: Regular monitoring of homocysteine and methionine levels, along with consultation with healthcare professionals, is advisable for personalized management based on individual health needs and genetic profiles.

For more detailed information, refer to the NIH Vitamin B12 Health Professional Fact Sheet.

Important Note:

It's essential to discuss these insights with healthcare professionals for personalized recommendations, ensuring that your health decisions are well-informed and tailored to your genetic profile.

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5 rs1801394 (A66G) and rs1532268 (C524T)

MTRR

MTRR Gene Role:

The MTRR gene, known as Methionine Synthase Reductase, is integral to the metabolic pathway of homocysteine and methionine. It's pivotal in producing S-adenosyl methionine, the primary methyl donor in cellular processes (Greenop et al., 2015), and plays a key role in the folate and vitamin B12-dependent conversion of homocysteine to methionine (Shi et al., 2019). MTRR also has significant functions in embryonic stem cell development and folate metabolism. Crucially, it maintains the active form of vitamin B12, which is essential for DNA synthesis and methylation, underscoring its importance in cellular health and genetic stability.

Mutation Details and Health Implications:

MTRR rs1801394 (A66G)

Type: This SNP involves the change of adenine (A) to guanine (G)

Frequency: Approximately 36.4% of people globally have this genetic variant.

Population-Specific Frequencies:

European: Approximately 54.4%
African: Around 28.7%
Asian: About 26.9%

Clinical Significance:

The MTRR A66G polymorphism has been the subject of extensive research due to its potential impact on DNA methylation. This polymorphism in the MTRR gene is believed to influence the efficiency of methionine synthase reductase, an enzyme vital for maintaining adequate levels of methionine and S-adenosylmethionine (SAM), a key methyl donor in DNA methylation processes (X. Y. Li et al., 2015). Variations in the MTRR gene could potentially influence DNA methylation patterns, thereby affecting gene expression and susceptibility to various health conditions (Crescenti et al., 2013).

However, it is important to note that while these studies provide valuable insights into the potential implications of the MTRR A66G polymorphism on DNA methylation and health outcomes, more research is needed to fully understand its broader health effects (X. Y. Li et al., 2015).

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rs1801394 (A66G) and rs1532268 (C524T)

MTRR

MTRR rs1532268 (C524T)

Type: This SNP involving the change of cytosine (C) to thymine (T).
Frequency: Relatively common, approximately 27% of people globally have this genetic variant.

Clinical Significance:

Classification: Most often classified as Benign.

Take Action:

Dietary Recommendations: A diet rich in vitamin B12 and folate is advisable by the literature. These nutrients are essential for proper methylation processes and DNA synthesis.

Food Sources: Include foods like meat, dairy, fortified grains, leafy greens, and legumes to ensure sufficient intake of these vitamins.

Important Note:

Your dietary choices and any supplements should be in line with your unique genetic profile, and it's important to discuss these with healthcare professionals for personalized guidance.

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Conclusion

Conclusion

MetaGen Mutations Panel offers a comprehensive and holistic view of your health. This comprehensive assessment provides insights into your genetic and epigenetic makeup.

For personalized interpretation and advice, consult with healthcare professionals. Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other health care professional prior to the collection of a sample for testing. This test is not a substitute for visits to a doctor or other health care professional. Users should consult with their doctor or other health care professional if they have any questions or concerns about the results of their test or their current state of health.

Disclaimer

Disclaimer

Intended Use: This report is for informational purposes for a better understanding of how specific genetic variations might impact your health and should not be considered medical advice.

Consult Healthcare Professionals: Always seek professional advice for health-related decisions. It's important to discuss these findings with a healthcare provider for a comprehensive assessment and tailored recommendations. This report is not a substitute for medical consultation. For more information or to discuss your results, please contact a healthcare professional. We also recommend speaking with a healthcare professional, genetic counselor, or equivalent professional before getting the results of the test.

Regulatory Status: The tests and analyses here are not FDA or Health Canada approved and are meant for personal health insights only.

Limitations: Genetic data should be interpreted as part of a broader health assessment, including lifestyle and environmental factors. This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.

Recommendations: The suggestions in this report are for discussion with your healthcare provider and are not definitive medical directives.

Data Source Acknowledgment: The information in this report is derived from the dbSNP NCBI database and clinical genetic studies, ensuring a scientifically sound foundation for the provided insights and reflecting current scientific understanding in genetics. This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other health care professional.

HKG epiTherapeutics Limited is a US accredited lab and this test was validated for CAP. HKG epiTherapeutics Limited is committed to maintain the accuracy, security and confidentiality of your personal information.

Sample Processing Issues: The laboratory may not be able to process a sample if there is insufficient DNA. In this case, the sample is considered to have failed. Our laboratory will contact you and provide a new saliva collection kit.

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Summary of Validation

Summary of Validation for MetaGen Test

To ensure the accuracy and reliability of the MetaGen test, comprehensive validation processes were conducted. These validations are crucial for confirming that the test can consistently provide precise genetic information under various conditions and sample types. Here is a summary of the key validation performed.

1) Limit of Detection (LOD)

HeLa Genomic DNA: The LOD for the MetaGen test was determined using HeLa genomic DNA. This involved assessing the minimal DNA input at which all 8 SNPs of the five MetaGen genes (AHCY, COMT, MTHFR, MTR, MTRR) could be detected reliably.

MetaGen Synthetic Oligo Pool: An additional LOD was established using a synthetic oligo pool designed to mimic the target SNPs. This helped confirm the test's analytical precision in making the correct SNP calls at different amount of DNA input.

2) Repeatability and Reproducibility

Repeatability: The test was performed twice under the same conditions to ensure consistent results. This included performing the complete MetaGen test by the same technician in different days. The repeatability was validated by obtaining consistent SNP call results in different runs.

Reproducibility: The test's reproducibility was validated by conducting the assay by different technicians, on different days, and using different equipments, which ensured that the test results were reproducible across various conditions.

3) Reagent Stability

The stability of the reagents used in the MetaGen test was assessed by storing them for different durations to ensure that they remain effective.

4) Testing on Clinical Saliva Samples

DNA Extraction in HKG Laboratory: The test was performed on DNA extracted from clinical saliva samples in the HKG laboratory to validate the process within the in-house environment.

External Commercial Lab: To further validate the robustness of the test, it was also conducted on DNA extracted from clinical saliva samples in an external commercial laboratory. This helped ensure that the test would produce reliable results regardless of the DNA extraction site.

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Summary of Validation

5) Interference Testing

Food, Alcohol, Smoking: The test was evaluated for potential interference from common substances that might be present in saliva, including food particles, alcohol, and tobacco smoke residues.

Amylase, IgA, Albumin, Hemoglobin: The presence of these proteins, commonly found in saliva, was tested to ensure they do not interfere with the accuracy of the DNA analysis.

Microbial DNA: Given the diverse microbial content in saliva, the test was also validated to ensure that microbial DNA does not affect the detection of the human genomic SNPs being analyzed.

These validation steps ensure that the MetaGen test is reliable, sensitive, and robust, providing accurate genetic information under a variety of conditions and sample types.

For more information, please visit our website: Understand your gene mutations to prevent disease and increase personal health. (epimedtech.com)

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FAQ

FAQ: Understanding Your MetaGen Test Results

1. What health conditions or diseases are tested by the MetaGen panel?
The MetaGen panel tests for genetic variants associated with conditions like cardiovascular disease, cancer susceptibility, and metabolic disorders. Each variant tested has been selected based on well-established clinical implications and supported by peer-reviewed scientific studies.

2. What is the purpose of the MetaGen test?
The purpose of the MetaGen test is to provide you with genetic information that may influence your risk of developing certain health conditions. This test can inform lifestyle decisions and facilitate discussions with your healthcare provider about personalized health strategies.

3. What information will the MetaGen test provide?
The MetaGen test will provide information on specific genetic variants known to be associated with certain health conditions. It will indicate whether you carry a variant that may increase your risk for these conditions. The report includes a detailed explanation of each variant and its potential impact on your health.

4. What information will the MetaGen test not provide?
The MetaGen test does not provide a comprehensive genetic profile. It does not diagnose any disease, nor does it offer information on your current state of health. The absence of a variant in the test does not rule out the presence of other genetic factors that may influence disease risk.

5. How do race and ethnicity affect the test results?
Genetic variants can have different frequencies in different populations. The relevance and interpretation of your results may vary based on your ethnic background. The MetaGen report takes these differences into account to provide contextually accurate information.

6. Which populations are the variants in the test most applicable to?
The variants included in the MetaGen test are most applicable to individuals of European, Asian, and African descent, as these populations have been the primary focus of the studies referenced. The applicability to other populations may vary, and additional context may be necessary for accurate interpretation.

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FAQ: Understanding Your MetaGen Test Results

7. What do the test results mean?
The test results indicate whether you carry specific genetic variants that may increase your risk for certain health conditions. Each variant's description includes its known impact, the strength of the association with disease, and any relevant lifestyle or medical recommendations.

8. What are other risk factors that contribute to disease?
Beyond genetic factors, lifestyle choices such as diet, exercise, smoking, and alcohol consumption play significant roles in disease risk. Environmental exposures and family history also contribute to your overall risk profile.

9. What are the appropriate follow-up procedures?
If you receive a result indicating an increased genetic risk, it is recommended that you discuss these findings with your healthcare provider. They can provide personalized advice and may suggest additional screening or preventive measures based on your results and overall health.

10. How do the test results affect my family?
Genetic variants can be inherited, so your results may have implications for your family members. It is important to discuss your results with a healthcare provider to understand the potential impact on your relatives and consider genetic counseling if necessary.

11. Where can I find more information?
For more detailed information, please refer to the resources linked below:
- National Institutes of Health (NIH)
- Genetics Home Reference
- Centers for Disease Control and Prevention (CDC)
- Genetic and Rare Diseases Information Center (GARD)

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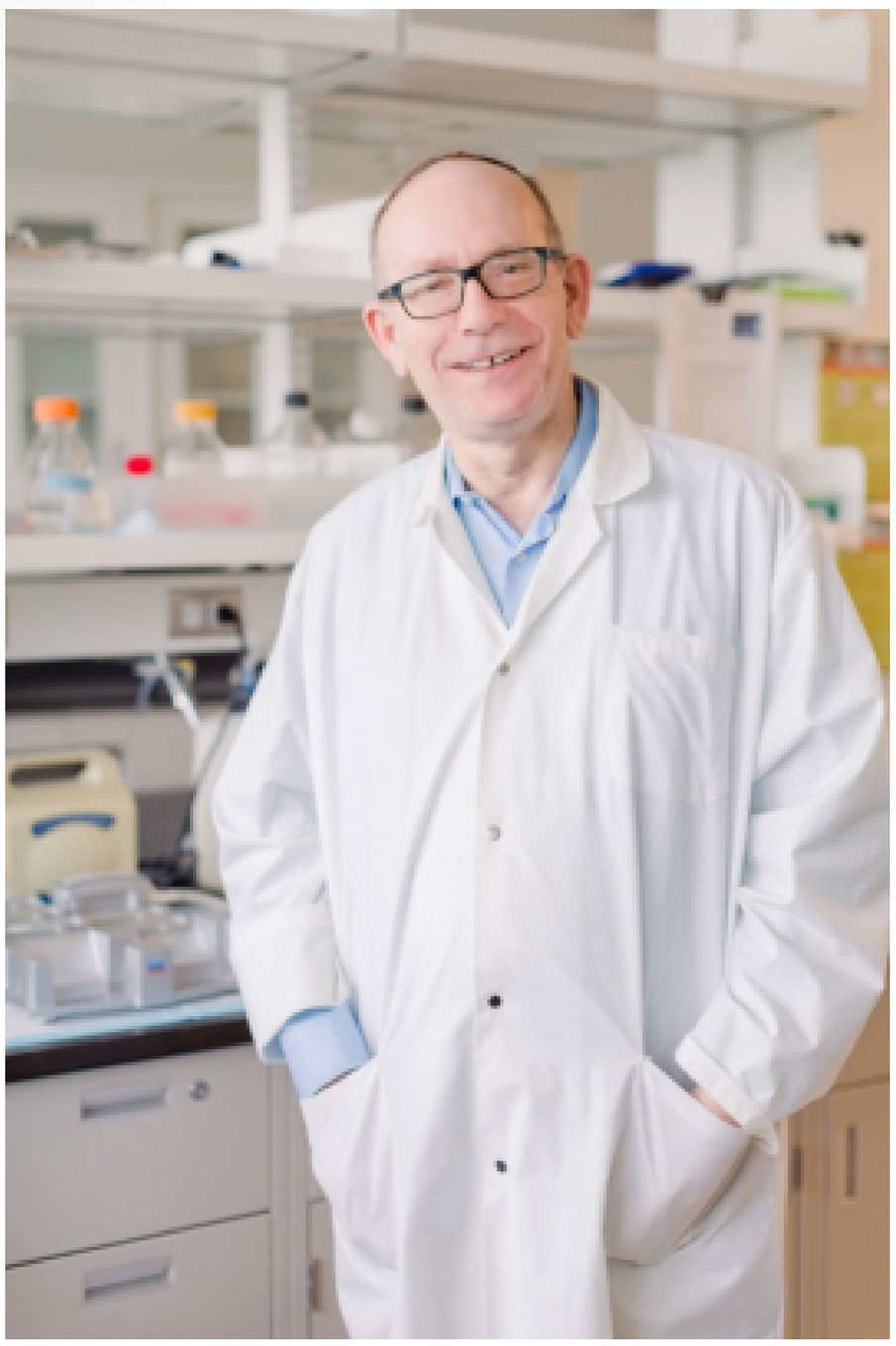
HKG epiTherapeutics
Pioneering Advances in Genetic Testing

HKG epiTherapeutics: Accredited Excellence

HKG epiTherapeutics, established in 2016, is renowned for its cutting-edge approach to healthcare, particularly in epigenetic research and testing. Our laboratory, situated within the prestigious HKG Science Park, has earned accreditations from the United States' College of American Pathologists (CAP) and the Clinical Laboratory Improvement Amendments (CLIA), signifying our commitment to the highest standards in laboratory testing and research integrity.



Dr. Moshe Szyf: Visionary Founder

The founding of HKG epiTherapeutics was inspired and led by Dr. Moshe Szyf, a luminary in the field of epigenetics and a former professor from McGill University's Department of Pharmacology and Therapeutics. Dr. Szyf's pioneering work includes:

- Being an inventor of the first broad patents in the field of epigenetics and the first patents linking DNA methylation and cancer.
- Founding the field of behavioral and psychiatry epigenetics.
- Publishing over 320 papers, significantly contributing to the advancement of epigenetics.
- Serving as a Fellow of the Royal Society of Canada and the Canadian Academy of Health Sciences.

Dr. Szyf's illustrious career extends beyond research and innovation. He has authored more than 320 peer-reviewed articles, solidifying his role as a key figure in epigenetics. He founded and served as the inaugural chief editor of a leading journal in the field, is an associate editor for Clinical Epigenetics, and holds a position on the Editorial Board of Environmental Epigenetics. His contributions have been instrumental in shaping the understanding and application of epigenetic science in medicine.

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The Company

HKG epiTherapeutics: Bringing Innovation to the world

HKG epiTherapeutics specialize in providing advanced genetic and epigenetic testing, ensuring that our customers benefit from the latest developments in epigenetics and genetic research.

Our Collective Mission and Expansion

HKG epiTherapeutics are dedicated to harnessing the power of epigenetics combined with state-of-the-art sequencing technologies. We are committed to early disease detection and prevention, with our expertise spanning across various aspects of personalized medicine. Our expansion plans aim to replicate the success of our Hong Kong laboratory in North America and beyond.

Conclusion

Customers of HKG epiTherapeutics can rest assured that they are receiving services backed by pioneering research, accredited facilities, and a team of experts led by one of the foremost authorities in epigenetics, Dr. Moshe Szyf. Our mission is to bring innovative health solutions to the forefront of medical science, improving healthcare outcomes worldwide.