**Welcome**

Dear Jane Doe,

NMC Genetics is pleased to provide your **Kidney Health** report based on your unique genomic profile. The report offers you a snap-shot of your genetic response pertaining to your kidney health. The interpretations and recommendations made in your report are based on data curated by our scientific experts from hundreds of clinical studies, clinical trials and Genome Wide Association Studies (GWAS) spanning decades of global research. Your DNA was extracted from your saliva/blood sample and processed in our labs equipped with next generation sequencing and microarray; utilizing globally validated procedures. The information received from your genetic code determines your kidney health. We continuously strive to update our proprietary genomic and clinical databases to improve our tests and recommendations. With insights from this report, your clinicians or Wellness consultant has a guidance map to device a personalized drug and accordingly lifestyle changes to help you achieve optimal health. By seeking professional advice and following the recommendations you can improve your health holistically.

Wishing you good health!

**About Us**

NMC Genetics is a clinical genomics company with a vision to innovate healthcare using genomics and data science.

Our services, delivered from a state-of-the-art genomics laboratory, empower clinicians and health care professionals with precise and actionable results to help improve patient care. NMC Genetics has strong focus and domain expertise in clinical genomics, preventive health, and personalized medicine. Led by a unique team of highly skilled molecular biologists, bioinformaticians and data scientists, NMC Genetics is poised for a big leap into the future of healthcare.

NMC Genetics is a subsidiary of NMC Healthcare LLC. a largest private healthcare company in the UAE and ranks amongst the leading fertility service providers in the world. Over the last forty-three years, NMC has earned the trust of millions, thanks to its personalized care, genuine concern and a sincere commitment to the overall well-being of the patient.

NMC was the first company from Abu Dhabi to list on the London Stock Exchange and is now part of the premium FTSE 100 Index, an elite club of top 100 blue-chip companies by market cap. NMC's strategic acquisitions coupled with its legacy institutions have allowed us to clear the service gap in our healthcare delivery system and offer a continuum of care to patients.

**Legal Disclaimer**

This report is based on your unique DNA results obtained by testing your buccal swabs/blood/saliva

samples in response to a selection of key genes that are associated with the individual health. NMC

Genetics provides genetic assessment services only for investigational purposes and the information

thus given should be interpreted and used exclusively only by qualified medical practitioners, certified physicians, dieticians, nutritionist, sports therapists and others in similar professions. The company does not provide any medical advise and this report does not constitute a medical diagnostic report.

Genetic results are unique but being associated with a futuristic technology, the same must be used only under proper advice. NMC Genetics does not guarantee or in any way confirm any future disease or ailment associated with the genetic data disclosed in this report. For any contraindications you are advised to get supportive tests conducted from appropriate hospitals/laboratories.

The company's role is limited to providing results of genetic tests and providing a broad set of general recommendations. More detailed instructions that may be specific to you are to be made by qualified professional practitioners only. General guidelines provided in our reports are for informational purposes only. They do not constitute professional or medical advice. While assessing your report and providing these recommendations we assume that you are in a general state of good health and do not take into account your past or existing health conditions and or any medication taken by you (either in the past or currently), even if you have provided us with such information. You should consult your medical practitioner before acting on it.

To the fullest extent permitted by law, neither NMC Genetics and nor its officers, employees or representatives will be liable for any claim, proceedings, loss or damage of any kind arising out of or in connection with acting, or not acting, on the assertions or recommendations in the report. This is a comprehensive exclusion of liability that applies to all damage and loss, including, compensatory, direct, indirect or consequential damages, loss of data, income or profit, loss of or damage to property and claims of third parties, howsoever arising, whether in tort (including negligence), contract or otherwise. Nothing in this statement is intended to limit any statutory rights you may have as a consumer or other statutory rights which may not be excluded, nor to exclude or limit our liability to you for death or personal injury resulting from NMC Genetics negligence or that of its officers, employees or other representatives. Nothing in this statement will operate to exclude or limit liability for fraud or fraudulent misrepresentation.

**Kidney Health**

Kidneys are fist-sized organs located at the bottom of the rib cage, on both sides of the spine. They perform several functions. Most importantly, they filter waste products, excess water, and other impurities from the blood. These waste products are stored in the bladder and later expelled through urine. In addition, kidneys regulate pH, salt, and potassium levels in the body. They also produce hormones that regulate blood pressure and control the production of red blood cells.

Thus, maintaining kidney health is important to overall health and general well-being. By keeping the kidneys healthy, the body can filter and expel waste properly and produce hormones to help the body function properly.

**Genetics & Kidney Health**

Diabetes and high blood pressure are the most common causes of kidney disease. Kidney diseases can be hereditary. Kidney disease also runs in families. You may be more likely to get kidney disease if you have a close relative with kidney disease.

**Introduction**

DNA is the building block of all living organisms. Human DNA consists of about 3 billion bases, and more than 99% of those bases are the same in all people. The order or sequence of these bases is the set of instructions for building and maintaining an organism, similar in a way by which letters of the alphabet appear in a certain order to form words and sentences.

**What is DNA?**

DNA (Deoxyribonucleic), is the hereditary material in humans and almost all other organisms.

Most DNA is located in the cell nucleus (where it is called nuclear DNA), but a small amount of DNA can also be found in the mitochondria (where it is called mitochondrial DNA or mtDNA). The information in DNA is stored as a code made up of four chemical bases: adenine (A), guanine (G), cytosine (C), and thymine (T).

A gene is the basic physical and functional unit of heredity. Genes are made up of DNA. Some genes act as instructions to make molecules called proteins. However, many genes do not code for proteins. In humans, genes vary in size from a few hundred DNA bases to more than 2 million bases. The Human Genome Project estimated that humans have between 20,000 and 25,000 genes. Every person has two copies of each gene, one inherited from each parent.

**What is a gene mutation and how do mutations occur?**

A gene mutation is a permanent alteration in the DNA sequence that makes up a gene, such that the sequence differs from what is found in most people.

Mutations range in size; they can affect anywhere from a single DNA building block (base pair) to a large segment of a chromosome that includes multiple genes.

Gene mutations can be classified in two major ways:

Hereditary (or germline) mutations are inherited from one or both parents and are present throughout a person’s life in virtually every cell in the body.

Acquired (or somatic) mutations occur at some time during a person’s life and are present only in certain cells, not in every cell in the body

**What is Genetic testing?**

Genetic testing is a type of medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person’s chance of developing or passing on a genetic disorder. Genetic tests can be performed using either saliva, blood, tissue, or other human cells.

**What do the results of Genetic tests mean?**

A positive test result means that the laboratory found a change in a particular gene, chromosome, or protein of interest. Depending on the purpose of the test, this result may confirm a diagnosis, indicate that a person is a carrier of a particular genetic mutation, identify an increased risk of developing a disease in the future, or suggest a need for further testing.

A negative test result means that the laboratory did not find a change in the gene, chromosome, or protein under consideration. This result can indicate that a person is not affected by a particular disorder, is not a carrier of a specific genetic mutation, or does not have an increased risk of developing a certain disease. However, there is still the possibility that any unknown genetic variation can still be a risk factor. A variant of unknown significance (VUS) can also be found in a genetic sequence for which the association with disease risk is unclear.

**How genetic testing impacts your wellbeing**?

Genetic defects can affect our health, although in many cases they don't manifest into a disease, but increases the risk of disease. External factors (such as the environment or lifestyle) influences the manifestation of the disease. For example, If a person is intolerant to lactose, due to a genetic defect, this person is perfectly healthy as long as she does not consume milk or milk products. In many cases health issues appear only in conjunction with certain environmental influences - in this case, consuming products that contain lactose.

Genetic variations called SNPs (pronounced “snips”) or “deletions” or “additions” can affect the way our bodies absorb, metabolize, and utilize nutrients, and determine how effectively we eliminate Xenobiotics (drugs, pollutants) and even potential carcinogens. By understanding the mechanisms by which these genes work and analysing data generated from genome-wide association studies (known as GWAS) and Mendelian randomization, scientists can now understand what impact SNPs may have on disease risk and relationship with certain gene-environmental contexts.

Another example is, if a regulatory gene for iron intake is defective, this can increase the risk of iron assimilation into the body. Your healthcare practitioner can adjust iron intake through natural foods and supplements to mitigate the risk of iron deficiency.

Once researchers understand how specific genotypes can affect how our genes function, this enables development of the most favourable nutritional and lifestyle strategies specific to a person’s genotype.

A healthy lifestyle is, of course, generally preferable, because it can neutralize many genetic predispositions even without knowing underlying risks. However, genetic testing provides you with appropriate information about underlying risk factors and help an individual to implement pro-active health plan with his/her healthcare practitioner to lead a healthy life.

**SOME FACTS:**

In human beings, 99.9% bases are same, remaining 0.1% makes a person unique in terms of:

* Different attributes / characteristics / traits
* How a person looks and what disease risks he or she may have
* Harmless (no change in our normal health)
* Harmful (can develop into diseases like diabetes, cancer, heart disease, Huntington's disease, and hemophilia)
* Latent (These variations found in genes but are not harmful on their own. The change in each gene function only becomes apparent under certain conditions e.g. increase in stress and susceptibility to heart attack)

**About Your Kidney Health Report**

This comprehensive genetic report consolidates up-to-date research on most of the common SNPs that research suggests may have actionable nutritional and lifestyle interventions based on scientific evidence. We use hundreds of studies to bring you the genetic information in the Genetic report.

The reporting format is very consistent and very lucid to understand. The report comprises of following sections in that order.

1. **Summarized results section** : This section comprise of master summary.
2. **Detailed report section**: This section gives the detailed overview of every condition. There is summarized results table, a group of relevant traits, corresponding genetic response and interpretations are listed. Each trait or phenotype has its response is marked as good, bad or average.

This information provides you insight into specific risks such as effect of the marker on kidney diseases. Summary of recommendations in terms do's and dont’s of lifestyle, nutrition, supplementation or exercise are included. This is how the result for a genetic marker associated to an individual trait is graded:

|  |  |  |  |
| --- | --- | --- | --- |
| **Response** | **Risk Level** | **Zone** | **Interpretation** |
| Good | Low/Normal risk |  | Your genetic predisposition to the disease is normal or low. |
| Average | Medium risk |  | Your genetic predisposition to the disease is average. Hence, act as per the recommendations. |
| Bad | High Risk |  | Your genetic predisposition to the disease is high. Hence, act as per the recommendations or consult your healthcare practitioner. |

**Your Summarized Kidney Health Report**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **conditions** | **markers tested** | **gene name** | **your genotype\*** | **your response** |
| **Risk of Hyperuricemia** | rs2725220 | ABCG2 | GG | **Bad** |
| **Autosomal Dominant Polycystic Kidney Disease (ADPKD):** | rs28937907 | PKHD1 | CC | **Moderate** |
| rs137852944 | CC |
| rs369925690 | CT |
| rs199531851 | AA |
| rs760222236 | AA |
| rs200511261 | AA |
| rs137852949 | TT |
| rs398124502 | del/del |
| **Hypomagnesemia (Low Magnesium)** | rs4072037 | MUC1 | CT | **Moderate** |
| rs7965584 | ATP2B1 | AG |
| rs3925584 | DCDC5 | CT |
| rs13146355 | SHROOM3 | AG |
| rs6746896 | CNNM4 | AG |
| **Chronic Kidney Disease** | rs10906850 | NMT2 | CT | Moderate |
| rs73885319 | APOL1 | AG |
| **Childhood Sensitive Nephrotic Syndrome** | |  |  | | --- | --- | | rs6478109 |  | |  |  | | TNFSF15 | CT | Moderate |
| rs1071630 | HLA-DQA1 | AG |
| **Idiopathic membranous Nephropathy** | rs2187668 | HLA-DQA1 | AG | Moderate |
| rs4664308 | PLA2R1 | AG |

**Your Detailed Report**

**Risk of Hyperuricemia:**

Hyperuricemia occurs when there’s too much uric acid in your blood. High uric acid levels can lead to several diseases, including a painful type of arthritis called gout. Elevated uric acid levels are also associated with health conditions such as heart disease, diabetes, and kidney disease.

**Symptoms:**

Only about one-third of people with hyperuricemia experience symptoms. This is known as asymptomatic hyperuricemia.

Although hyperuricemia isn’t a disease, if uric acid levels remain high, over time they can lead to several diseases.

* Gout
* Kidney stones

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **markers tested** | **gene name** | **your genotype\*** | **your response** | **interpretation** | **Recommendation** |
| rs2725220 | ABCG2 | GG | Bad | Your genotype is associated with increased risk of hyperuricemia. | Limit purine-rich foods like organ meat, fishes like mackeral, sardines, tuna etc.Avoid sugar and alcohol.Maintain ideal body weight.Balance insulin levels. Eat adequate fibre in the diet. Reduce stress.Check medications and supplements. Vitamin C can be beneficial. |

**Your Genetic Profile**

**\*some of the genotypes may be imputed**

**Gene Description:**

**ABCG2**: This gene encodes ATP binding cassette subfamily G member 2. The membrane-associated protein encoded by this gene is included in the superfamily of ATP-binding cassette (ABC) transporters. ABC proteins transport various molecules across extra- and intra-cellular membranes.

**Autosomal Dominant Polycystic Kidney Disease (ADPKD):**

Mutation only in single copy of gene is strong enough to overcome the effects of the other, normal gene. Hence, the name Autosomal Dominant Polycystic Kidney Disease. It is an inherited kidney disorder. It causes fluid-filled cysts to form in the kidneys. PKD may impair kidney function and eventually cause kidney failure. PKD is the fourth leading cause of kidney failure. People with PKD may also develop cysts in the liver and other complications.

**Symptoms:**

* pain or tenderness in the abdomen
* blood in the urine
* frequent urination
* pain in the sides
* urinary tract infection (UTI)

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| --- | --- | --- | --- | --- | --- |
| **markers tested** | **gene name** | **your genotype\*** | **your response** | **interpretation** | **Recommendation** |
| rs28937907 | PKHD1 | CC | Moderate | Your genotype is associated with moderate risk of polycystic kidney disease. | Keep a check on your Blood pressure and Kidney profile. Maintain heathy weight. Quit smoking and drinking alcohol |
| rs137852944 | CC |
| rs369925690 | CT |
| rs199531851 | AA |
| rs760222236 | AA |
| rs200511261 | AA |
| rs137852949 | TT |
| rs398124502 | del/del |

* kidney stones
* pain or heaviness in the back
* skin that bruises easily
* pale skin color
* fatigue
* joint pain
* nail abnormalities

**Your Genetic Profile**

**\*some of the genotypes may be imputed**

**Gene Description:**

**PKHD1**: This gene encodes PKHD1 ciliary IPT domain containing fibrocystin/polyductin. The protein encoded by this gene is predicted to have a single transmembrane (TM)-spanning domain and multiple copies of an immunoglobulin-like plexin-transcription-factor domain. Alternative splicing results in two transcript variants encoding different isoforms. Other alternatively spliced transcripts have been described, but the full length sequences have not been determined. Several of these transcripts are predicted to encode truncated products which lack the TM and may be secreted. Mutations in this gene cause autosomal recessive polycystic kidney disease, also known as polycystic kidney and hepatic disease-1.

**Hypomagnesemia (Low Magnesium)**

Magnesium is one of the most abundant essential minerals in your body. Low magnesium is typically due to decreased absorption of magnesium in the gut or increased excretion of magnesium in the urine. Magnesium levels are largely controlled by the kidneys. The kidneys increase or decrease excretion (waste) of magnesium based on what the body needs. Continually low dietary intake of magnesium, excessive loss of magnesium, or the presence of other chronic conditions can lead to hypomagnesemia.

**Symptoms**

* nausea
* vomiting
* weakness
* decreased appetite

As magnesium deficiency worsens, symptoms may include:

* numbness
* tingling
* muscle cramps
* seizures
* muscle spasticity
* personality changes
* abnormal heart rhythms

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| --- | --- | --- | --- | --- | --- |
| **markers tested** | **gene name** | **your genotype\*** | **your response** | **interpretation** | **Recommendation** |
| rs4072037 | MUC1 | CT | Moderate | Your genotype is associated with moderate risk of lower serum magnesium levels. |  |
| rs7965584 | ATP2B1 | AG | Moderate |  |
| rs3925584 | DCDC5 | CT | Moderate |  |
| rs13146355 | SHROOM3 | AG | Moderate |  |
| rs6746896 | CNNM4 | AG | Moderate |  |

**Your Genetic Profile**

**\*some of the genotypes may be imputed**

**Gene Description**

**MUC1**: This gene encodes mucin 1, cell surface associated. Plays an essential role in forming protective mucous barriers on epithelial surfaces. Overexpression, aberrant intracellular localization, and changes in glycosylation of this protein have been associated with carcinomas.

**ATP2B1**: This gene encodes ATPase plasma membrane Ca2+ transporting 1. These enzymes remove bivalent calcium ions from eukaryotic cells against very large concentration gradients and play a critical role in intracellular calcium homeostasis.

**DCDC5**: This gene encodes doublecortin domain containing 5. The protein encoded by this gene is a hydrophilic, intracellular protein. It contains a single doublecortin domain and is unable to bind microtubules and to regulate microtubule polymerization.

**SHROOM3**: This gene encodes shroom family member 3. This protein may be involved in regulating cell shape in certain tissues.

**CNNM4**: This gene encodes cyclin and CBS domain divalent metal cation transport mediator 4. The encoded protein may play a role in metal ion transport.

**Chronic Kidney Disease (CKD)**

Chronic kidney disease, also called chronic kidney failure, describes the gradual loss of kidney function. Your kidneys filter wastes and excess fluids from your blood, which are then excreted in your urine. When chronic kidney disease reaches an advanced stage, dangerous levels of fluid, electrolytes and wastes can build up in your body.

**Symptoms:**

* Nausea
* Vomiting
* Loss of appetite
* Fatigue and weakness
* Sleep problems
* Changes in how much you urinate
* Decreased mental sharpness
* Muscle twitches and cramps
* Swelling of feet and ankles
* Persistent itching
* Chest pain, if fluid builds up around the lining of the heart
* Shortness of breath, if fluid builds up in the lungs
* High blood pressure (hypertension) that's difficult to control

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| --- | --- | --- | --- | --- | --- |
| **markers tested** | **gene name** | **your genotype\*** | **your response** | **interpretation** | **Recommendation** |
| rs10906850 | NMT2 | CT | Moderate | Your genotype is associated with moderate risk of chronic kidney disease (CKD). |  |
| rs73885319 | APOL1 | AG | Moderate |  |

**Your Genetic Profile**

**\*some of the genotypes may be imputed**

**Gene Description**

**NMT2**: This gene encodes N-myristoyltransferase 2. N-terminal myristoylation is a lipid modification that is involved in regulating the function and localization of signaling proteins.

**APOL1**: This gene encodes apolipoprotein L1. This apolipoprotein L family member may play a role in lipid exchange and transport throughout the body, as well as in reverse cholesterol transport from peripheral cells to the liver. Several different transcript variants encoding different isoforms have been found for this gene.

**Childhood Steroid Sensitive Nephrotic Syndrome**

Nephrotic syndrome (NS) is the most common glomerular disease seen in the pediatric age group. It is the second most common kidney disease seen in pediatric nephrology clinic.

Patients with steroid-resistant nephrotic syndrome (SRNS) represent a challenging subset of patients with nephrotic syndrome who often fail standard immunosuppression and have a higher likelihood of progressing to end-stage renal disease.

About 80% children with idiopathic nephrotic syndrome show remission of proteinuria (increased levels of protein in the urine) following treatment with corticosteroids, and are classified as ‘steroid sensitive’.

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| --- | --- | --- | --- | --- | --- |
| **markers tested** | **gene name** | **your genotype\*** | **your response** | **interpretation** | **Recommendation** |
| |  |  | | --- | --- | | rs6478109 |  | |  |  | | TNFSF15 | CT | Moderate | Your genotype is associated with moderate risk of childhood onset steroid-sensitive nephrotic  syndrome. |  |
| rs1071630 | HLA-DQA1 | AG |

**Your Genetic Profile**

**\*some of the genotypes may be imputed**

**Gene Description**

**TNFSF15**: This gene encodes TNF superfamily member 15. This cytokine is found to inhibit endothelial cell proliferation, and thus may function as an angiogenesis inhibitor.

**HLA-DQA1:** This gene encodes major histocompatibility complex, class II, DQ alpha 1. It plays a central role in the immune system by presenting peptides derived from extracellular proteins.

**Idiopathic membranous Nephropathy**

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| --- | --- | --- | --- | --- | --- |
| **markers tested** | **gene name** | **your genotype\*** | **your response** | **interpretation** | **Recommendation** |
| rs2187668 | HLA-DQA1 | AG | Moderate | Your genotype is associated with moderate risk of idiopathic membranous nephropathy. |  |
| rs4664308 | PLA2R1 | AG |

**Your Genetic Profile**

**\*some of the genotypes may be imputed**

**Gene Description**

**HLA-DQA1:** This gene encodes major histocompatibility complex, class II, DQ alpha 1. It plays a central role in the immune system by presenting peptides derived from extracellular proteins.

**PLA2R1:** This gene encodes phospholipase A2 receptor 1. The encoded protein likely exists as both a transmembrane form and a soluble form. The transmembrane receptor may play a role in clearance of phospholipase A2, thereby inhibiting its action. Polymorphisms at this locus have been associated with susceptibility to idiopathic membranous nephropathy.

**Chronic glomerulonephritis**

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| --- | --- | --- | --- | --- | --- |
| **markers tested** | **gene name** | **your genotype\*** | **your response** | **interpretation** | **Recommendation** |
|  |  |  |  |  |  |

**Your Genetic Profile**

**\*some of the genotypes may be imputed**

**Gene Description**

8. 9.

10. Lupus nephritis

11. Renal calculi

**Science behind the test**

**Test Methodology**

Genomic DNA is extracted from individual’s Saliva/Tissue/Blood by commercial DNA extraction kits. The genotyping and variant detection is carried out based on illumina Infinium® array protocol. The DNA is then, amplified, fragmented and hybridized to known DNA fragments immobilized in arrays on a Bead- Chip. Millions of such known DNA fragments (50mer probes) containing the target genetic variants are immobilized on the chip. The hybridized chip is then washed to remove non-hybridized DNA fragments. Single-base extension of the oligos on the BeadChip, using the captured DNA as a template, incorporates detectable labels on the BeadChip and determines the genotype call for the sample. The Illumina iScan® or BeadArray Reader scans the BeadChip, using a laser to excite the fluorophore of the single-base extension product on the beads. The scanner records high-resolution images of the light emitted from the fluorophores.

**Analytical Performance**

The genotyping was performed using a custom genotyping array platform (Illumina Inc). This test is a laboratory developed test with high reproducibility > 99% and high call rates > 98% to detect the variants and its performance has been validated in-house. Note that some of the genotypes may be imputed.

**Analysis**

Illumina GenomeStudio® Software is used for efficient genotyping data normalization, genotype calling, clustering, data intensity analysis. Genotypes are called for each sample by their signal intensity (norm R) and Allele Frequency (Norm Theta) relative to canonical cluster positions for a given SNP marker. The reporting software tool is developed in-house by NMC Genetics India Pvt. Ltd. The report is manually reviewed by experts before release.

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