MEDGENOME

258/A, 3rd Floor, Narayana Nethralaya Building, Narayana Health City,
Hosur Road, Bommasandra, Bangalore, Karnataka, India – 560 099

(Toll Free) 1800 103 3691 techsupport@medgenome.com
www.medgenome.com customersupport@medgenome.com

TEST REQUISITION FORM

Bill type ☐ MOU ☐ Retail ☐ Research

Each sample must be accompanied by this completed requisition.

*** Fields are mandatory**

Test Details

Test Name:* _____ Test Code:* _____

Sample type:

- ☐ Blood (in EDTA tube)
- ☐ Blood (in streck tube)
- ☐ DNA, Specify Source:_____
- ☐ Buccal swab
- ☐ Amniotic Fluid
- ☐ CVS
- ☐ Cultured CV
- ☐ Cultured amniocytes
- ☐ Fetal Blood (PUBS)
- ☐ Maternal blood for MCC (please send for prenatal studies)
- ☐ Products of Conception (POC), specifiy tissue:_____
- ☐ FFPE tissue Block (Block no. .....)
- ☐ DBS/FTA
- ☐ Fresh Frozen Tissue
- ☐ Saliva
- ☐ Other sample type (specify site) _____

Patient had a blood transfusion ☐Yes ☐No Date of last transfusion ___/___/___ (minimum 3 days of wait time is required for genetic testing)

Has he/she undergone allogenic bone marrow transplant: ☐Yes ☐No.

*Derived samples from non-accredited (CAP/NABL) laboratories will not be accepted for clinical processing.

Patient Details

Name:* (In Capital Letters) ..... D.O.B. DD MM YY Age:* _____ Gender:* M / F

Address: .....

Phone:..... E-mail I.D: .....

Clinician Details

Clinician's Name:* ..... Hospital Affiliation: .....

Address: ..... Phone : .....

..... Email id : .....

.....

Date of sample collection* DD MM YY

I understand that the current analysis is limited to molecular findings which co-relate with disease phenotype/symptoms/terms as mentioned in the clinical details provided by me. Incidental findings which may or may not be actionable are not routinely reported. They can however be provided on request after informed consent from the patient/guardian. As disease phenotype may evolve over time, the appearance of new symptoms/signs may alter test results or their significance: MedGenome laboratories cannot be held responsible for this. A re-analysis or a re-test may be required due to the former; this will be performed (if deemed necessary) at an additional cost. I am authorised to order the above tests as I am the treating physician/consulting physician in this case. I confirm that the patient/guardian (in case of minors) has been provided complete information regarding the test, including its limitations in a language of their understanding.

Medical Professional Signature* Date: Place:

Clinical notes/diagnosis:

Disease affection status Yes NO Parental consanguinity present Yes NO Age of manefestation:_____

Affected Sibllings Yes NO Details:_____

MEDGENOME

258/A, 3rd Floor, Narayana Nethralaya Building, Narayana Health City,
Hosur Road, Bommasandra, Bangalore, Karnataka, India – 560 099

(Toll Free) 1800 103 3691 techsupport@medgenome.com
www.medgenome.com customersupport@medgenome.com

Clinical Proforma

Auditory:
- ☐ SNHL-prelingual/postlingual
- ☐ Mondini defect/Enlarged Vestibular Aqueduct
- ☐ Microtia/Anotia/Large/Prominent ears
- ☐ Ear tags/creases/preauricular sinus

Cardiology:
- ☐ CHD: Septal defects/conotruncal/hypolasia
- ☐ Cardiomyopathy-Dilated/Hypertrophic/Non compaction/Arrhythmogenic dysplasia
- ☐ Arrhythmias.:LongQT/Brugada syndrome/ shortQT/others.....
- ☐ ECHO findings.....
- ☐ ECG findings.....

Disorders of sex development
- ☐ Karyotyping.....(specify)
- ☐ Ambiguous genitalia.....(specify)

Dermatology:
- ☐ Albinism: occular/OCA.....(type)
- ☐ Ectodermal dysplasia: Hidrotic/Hypohidrotic
- ☐ Epi. Bullosa: Simplex/Junctional/Dystrophica
- ☐ Icthyosis: Harlequin/Lamellar/Eythroderma
- ☐ Photosensitvity/Keloids/Lax,wrinkled skin
- ☐ Neurocutaneous markers.....

Endocrine:
- ☐ Diabetes Mellitus: Type 1/Type 2/MODY/Neonatal onset Hyperlipidemias.....(specify)
- ☐ Hypothyroidism/ Graves disease
- ☐ Hypoparathyroidism//Pseudohypoparathyroidism/ Hyperparathyroidism Pheochromocytoma/ paraganglioma/Adrenal insufficiency/CAH

Gastrointestinal and Liver:
- ☐ Hyperbilirubinemia: Unconjugated/Conjugated/
- ☐ Cholestasis/Neonatal Hepatitis/
- ☐ Liver failure/Chronic liver disease/Wilson disease
- ☐ Recurrent Pancreatitis/Chronic diarrhea
- ☐ Liver biopsy findings.....
- ☐ USG findings.....

Haematology/Immunology:
- ☐ Anemia.....(type)
- ☐ Bleeding disorders.....(specify)
- ☐ Recurrent infections.....(specify)
- ☐ Immunological workup.....
- ☐ Bone marrow examination.....

Movement disorders:
- ☐ Ataxia: episodic/progressive/telangiectasia
- ☐ Chorea/Athetosis/Dyskinesia
- ☐ Dystonia: .....(site, if focal)

Nephrology
- ☐ CAKUT.....(specify)
- ☐ Haematuria/Renal tubulopathy/Nephrotic syndrome
- ☐ Cystic kidneys: ARPKD/ADPKD/Other.....
- ☐ Renal biopsy findings.....

Neurological:
- ☐ Developmental delay: global/motor/speech
- ☐ Intellectual disability: mild/moderate/severe
- ☐ Autism/Hyperactivity/stereotypical movements
- ☐ Neuroregression: .....(age of onset)
- ☐ Seizures:.....(type)
- ☐ EEG:.....
- ☐ Recurrent headaches/migraine
- ☐ Suspected IEM.....(type,copy of report)

Neuromuscular and autonomic:
- ☐ Hypotonia: central/peripheral
- ☐ Weakness:proximal/distal/both/episodic
- ☐ Easy fatiguability/myalgia/cramps/myoglobinuria
- ☐ Weakness of: UL/LL/neck/face/bulbar muscles
- ☐ Calf hypertrophy/Scapular winging
- ☐ Contractures: Proximal/distal
- ☐ Joint Laxity: proximal /distal/dislocations
- ☐ Spasticity Autonomic involvement.....(specify)

Neuroimaging:
- ☐ Migration abnormalities: .....(specify)
- ☐ Calcifications.....(site)
- ☐ Atrophy: cerebral/ cerebellar/midbrain
- ☐ Hypoplasia: cerebellar/vermis/ pontocerebellar/pons/cerebellar cysts
- ☐ Hypomyelination/Demyelination:.....(specify)
- ☐ Basal ganglia abnormalities/Cerebral edema/Stroke/ Congenital malformations.:Holoprosencephaly/ Agenesis of corpus callosum/ Dandy Walker/ Hydrocephalus/Aqueductal Stenosis
- ☐ Intraventricular hemorrahge/Porencephaly/ Hydranencephaly

Opthalmology:
- ☐ Cataracts- congenital/unilateral/bilateral
- ☐ Cloudy cornea/Cherry red spot
- ☐ Coloboma.....(site)
- ☐ Glaucoma/Buphtahlmos
- ☐ Hyper/hypotelorism/K-F ring
- ☐ Microphthalmia/Anophthalmia
- ☐ Nystagmus/Ptosis/Ophthalmoplegia
- ☐ Optic atrophy/Retinitis Pigmentosa
- ☐ Retinoblastoma- unilateral/bilateral
- ☐ ERG/OCT findings.....
- ☐ Color vision test.....
- ☐ Fundus examination..... (attach photographs if available).

Perinatal History:
- ☐ Prematurity/Birth asyphyxia
- ☐ Teratogen .....(specify)
- ☐ Maternal illness.....(specify)
- ☐ Oligoamnios/Polyamnios
- ☐ Growth retardation-symmetric/asymmetric
- ☐ Abnormal USG.....(specify)

Skull and Hair:
- ☐ Microcephaly-Primary/Secondary
- ☐ Macrocephaly
- ☐ Craniosynostosis:.....(suture)
- ☐ Abnormal skull shape.....(specify)
- ☐ Encephalocele-frontal/occiptial
- ☐ Hair: Hypopigmented/silvery
- ☐ Sparse/absent/cutis aplasia
- ☐ Tricchorehxis nodosa

Skeletal/Limb:
- ☐ Polydactyly-preaxial/postaxial: Hands/Feet
- ☐ Syndactyly/Ectrodactyly/Absent thumbs-UL/BL
- ☐ Limb hypoplasia/aplasia/hypertrophy
- ☐ Micromelia/Rhizomelia/Mesomelia/Acromelia
- ☐ Metaphyseal/Diaphyseal/Epiphyseal abnormality
- ☐ Osteopenia/Fractures/Osteopetrosis
- ☐ Spinal involvement

Pedigree / Family History (Other Clinical Details)

MEDGENOME

258/A, 3rd Floor, Narayana Nethralaya Building, Narayana Health City,
Hosur Road, Bommasandra, Bangalore, Karnataka, India – 560 099

(Toll Free) 1800 103 3691 techsupport@medgenome.com
www.medgenome.com customersupport@medgenome.com

Informed Consent and Authorization Form

General Information About Genetic Testing

What is genetic testing?

Genetic disorders are caused by changes in a person's DNA. DNA is the material that provides instructions for our body's growth and development. For example, DNA determines such things as eye color and how our lungs work. DNA is compacted into 46 chromosomes, which are found in almost every cell of the body. A gene is a stretch of DNA on a chromosome that has the instructions for making a protein.

Genetic testing is a type of medical test that identifies changes in chromosomes and the DNA of a gene. The purpose of this test is to see if I, or my child, have a genetic variant or chromosome rearrangement causing a genetic disorder or to determine the chance I, or my child, will develop or pass on a genetic disorder in the future. For the purposes of this Consent, 'my child' can also mean my unborn child.

Additional information about the specific test being ordered is available from my health care provider or I can go to the Medgenome website, www.medgenome.com. This information includes the specific types of genetic disorders that can be identified by the genetic test, the likelihood of a positive result, and the limitations of genetic testing.

What could I learn from this genetic test?

If {I/my child} have a family history of one of the conditions that is being tested, I should inform the laboratory of the specific gene variant(s) or chromosome rearrangement present in the family if it is known. The genetic test may identify the cause of the genetic disease that {I/my child} have or a normal genetic result may significantly reduce, but cannot eliminate, the likelihood that the condition in {me/my child} is genetic or that {I/my child} will develop the genetic disorder in the future. The following describes the possible results from the test:

1) Positive: A positive result indicates that a gene or chromosome variation has been identified thexplains the cause of {my/my child's} genetic disorder or that {I/my child} am at increased risk to develop the disorder in the future. It is possible to test positive for more than one genetic variant.

2) Negative: A negative result indicates that no disease-causing genetic variant was identified for the test performed. It does not guarantee that {I/my child} will be healthy or free from other genetic disorders or medical conditions. If {I/my child} test negative for a variant known to be present in other members of {my/my child's family}, this result rules out a diagnosis of the same genetic disorder in {me/my child}.

3) Inconclusive/Variant of Uncertain Significance (VUS): A finding of a variant of uncertain significance indicates that a change in a gene was detected, but it is currently unknown whether that change is associated with a genetic disorder. A variant of uncertain significance is not the same as a positive result and does not clarify whether {I/my child} am at increased risk to develop a genetic disorder. The change could be a normal genetic variant or it could be disease-causing. Further analysis may be recommended, including testing both parents and other family members. Detailed medical records or information from other family members also may be needed to help clarify results.

Result interpretation is based on currently available information in the medical literature, research and scientific databases. Because the literature, medical and scientific knowledge are constantly changing, new information that becomes available in the future may replace or add to the information MedGenome used to interpret {my/my child's} results. MedGenome does not routinely re-analyze test results or issue new test reports, and has no obligation to do so. I, or {my/my child's} health care providers may monitor publicly available resources used by the medical community, such as ClinVar (www.clinvar.com), to find current information about the clinical interpretation of my/my child's variant(s).

What are the risks and limitations of this genetic test?

Genetic testing is an important part of the diagnostic process. However, genetic tests may not always give a definitive answer.

In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology.

Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in {my/my child's} family may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results.

Test results are interpreted in the context of clinical findings, family history and other laboratory data. Only variations in genes potentially related to the proband's medical condition are reported.

Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to: mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation, blood transfusion, or the presence of change(s) in such a small percentage of cells that may not be detectable by the test (mosaicism).

This test does not have the ability to detect all of the long-term medical risks that {I/my child} might experience. The result of this test does not guarantee my health or the health of my child/fetus. Other diagnostic tests may still need to be done, especially when only a genetic screening test has been performed previously.

Occasionally, an additional sample may be needed if the initial specimen is not adequate.

Allele drop out, which is a rare phenomenon, can affect the Sanger testing results. This is due to minor changes in the sequence where the primers bind resulting in non-amplification of these DNA strands. Less than 1% of cases are susceptible to this phenomenon leading to misdiagnosis.

Please note, Sanger sequencing is a customized test and the turnaround time (TAT) may vary depending on the complexity of the test

Disclaimer

In prenatal testing, Maternal cell contamination (MCC) of fetal sample will be tested using the MedGenome DNA Genotyping Panel. Even in cases of autosomal dominant disorders in which the father has the causative variant, blood or DNA from the mother is strongly encouraged to be sent for the MCC test. However, in cases where mother's sample is not available, it is noted that maternal cell contamination can affect the result.

The report shall be generated within Turnaround time (TAT), however, such TAT may vary depending upon the complexity of test(s) requested. MedGenome under no circumstances will be liable for any delay beyond the afore mentioned TAT.

Due to inherent technology limitations of the assay, not all bases of the exome/NGS panel can be covered by this test. 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 analysed, but have not been detected. The variants not detected by the assay that was performed may impact the phenotype. Coverage of the exome/NGS panel genes will be provided upon request.

Genes with pseudogenes, paralog genes and genes with low complexity may have decreased sensitivity and specificity of variant detection and interpretation due to inability of the data and analysis tools to unambiguously determine the origin of the sequence data in such regions.

Pathogenic variants may be present in a portion of the gene not covered by this test and therefore are not reported. The absence of reportable secondary findings for any particular gene does not mean there are no pathogenic variants in that gene.

Pathogenic variants that may be present in a relative, but are not present in the proband, will not be identified, or reported.

Interpretation of variants in this report is performed to the best knowledge of the laboratory based on the information available at the time of reporting. Re-analysis of variants in previously issued reports in light of new evidence is not routinely performed, but may be available upon request.

It is hereby clarified that the Report(s) generated from the Test(s) do not provide any diagnosis or opinion or recommends any cure in any manner. MedGenome hereby recommends the Patient and/or the guardians of the Patients, as the case may be, to take assistance of the Clinician or a certified physician or doctor, to interpret the Report(s) thus generated.

MedGenome hereby disclaims all liability arising in connection with the Report(s).

Only changes at the sequence level will be reported in the secondary findings report. Larger deletions/duplications, abnormal methylation, triplet repeat or other expansion variants, or other variants not routinely identified by whole exome sequencing will not be reported.

MedGenome recommends genetic counseling before and after having this genetic test. Further testing or additional consultations with a health care provider may be necessary.

MedGenome takes utmost care to maintain the intergrity of the sample. However there could be a loss or damage of sample during shipment for which MedGenome is not liable.

General information about genetic study based on clinical diagnostic data

About The Program:

You are being invited to take part in a study. This consent information is provided to help you decide if you want to contribute. You should not sign this document until you understand all of the information presented and until all of your questions about the research have been answered to your satisfaction.

The purpose of the study is to analyze and elucidate the importance of local genetics profile as one of the major contributing factors to human traits and diseases. In this context, most of the genetic studies have been done in European populations, ignoring a vast amount of genetic variation that exists in other populations and unfortunately reducing the potential for genetics-based learnings to improve health outcomes for Indians. MedGenome aims to democratize genomics technologies to benefit India's underserved population as well as other global populations. It is expected that the resources and results from MedGenome studies will contribute to the knowledge of genetics in India and global populations.

MEDGENOME

258/A, 3rd Floor, Narayana Nethralaya Building, Narayana Health City,
Hosur Road, Bommasandra, Bangalore, Karnataka, India – 560 099

(Toll Free) 1800 103 3691 techsupport@medgenome.com
www.medgenome.com customersupport@medgenome.com

Informed Consent

I have had the opportunity to discuss the Program described in this form with my physician. By signing this consent form, I am acknowledging that I understand the goals of the study and what my participating in the Program will involve.

I understand that participating in this Program is completely voluntary. I understand that the retention, storage, and further use of my/my child's health/medical history, biological samples, survey responses, and genetic data (the "Data") may help health care providers, scientists, and researchers understand the role of genes in human health. The Data could help enable discoveries including understanding the genetic basis of diseases, improving risk prediction for certain diseases, and/or the development of new or improved treatments or approaches for the prevention, diagnosis, treatment, or cure of diseases. I am aware that, although I/my child may not personally benefit from these discoveries, sharing data can help health care professionals offer better care for other patients in the future. I also understand that MedGenome does not intend to provide me or my child with ownership or commercial benefits that it may possibly derive from this study.

I, therefore, agree that MedGenome is allowed to store the Data for a period of at least 20 years and use it for the purposes outlined in this document and for related studies or commercial matters. I understand that any Data, including biological samples, shared with or transferred to health care providers, scientists, researchers and genetic databases will be anonymized or pseudonymized. This means information that may be used to identify me/my child, will be removed from the Data, and the Data will be bundled with data from other people so that it is very unlikely that I could be personally identified later. I am aware that my anonymous or pseudonymous data may also appear in scientific publications. Breach of privacy is the primary risk in being part of this study. To minimize any risks of a security breach that could leak my Data, MedGenome has security procedures in place.

I understand that the Data may be useful for further advising or testing me or my child and/or my or my child's family members and that the involvement of patients in medical studies can accelerate the efforts of scientists and drug developers to find better therapies, faster. I agree that I may be contacted regarding additional studies or testing and recognize that if I or my physician are contacted to inquire whether I, or my family members, would like to be involved in studies and I understand that I am not obligated in any way to participate in any future studies. This consent does not imply my consent to avail any specific treatment or participate in studies of any kind other than as described here. Furthermore, I also understand that prenatal sex determination is not allowed under the laws of India, and my consent to participate in the study does not imply solicitation of such information, in any form, from MedGenome, now or at any-time in the future, or from any of its affiliates, partners, contractors or sub-contractors whether MedGenome or its affiliates, partners, contractors or sub-contractors are subject to Indian laws or not.

If I change my mind and would like to opt-out of the Program, or would like to update any information, I will contact MedGenome's Customer Service at +91 (0) 80 67154990 / 991 to inform them. MedGenome will update their records to reflect the most current consent choice and/or any other updates to my information. After my request to opt-out, it may take up to 30 days to withdraw my information ("Withdrawal date"). I understand and agree that any analysis on the Data that has been performed or published prior to the Withdrawal Date will not be reversed, undone, or withdrawn. I understand that choosing not to give consent, or withdrawing from participation, will not affect access to my genetic reports or other information provided by my physician. Furthermore, if I have any questions, concerns, or inquiries around subject's rights I will email the Customer Service team at subjectprotection@MedGenome.com or call +91 (0) 80 67154990 / 991

I have understood the above and/or the research staff has explained to me/my child. Consent selections are as follows:

Primary Consent & Bio-banking Consent

For self/On behalf of this person (as I am legally authorized) - I give consent to participate in this Program and I agree to allow biological sample to be stored for future use for any biomedical studies.

(If consenting for child between ages 7-18 years)

I am this child's parent/legal guardian therefore I give consent to participate in this Program; and this child gives assent to participate.

(If consenting for child under the age of 7)

I am this child's parent/legal guardian therefore I give consent to participate in this Program.

☐ **YES** ☐ **NO**

Signature* _____

Date _____

*Fields are mandatory