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

Disease Segment*

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.

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 variants 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

Dysmorphism (attach photographs if available)

Details of dysmorphism

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

Pedigree / Family History (Other Clinical Details)

Details of accompanying samples (if any)

Name	Age	Sex	Relationship with patient	Clinical features (if any)

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 that explains 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).

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

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 complicity 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.

Please refer most recent version of the MedGenome test menu for turnaround time of specific tests.

***Disclaimer:** While the laboratory takes utmost care to return results within the turnaround time, there may be unforeseen circumstances due to which the TAT is sometimes exceeded. In such situation, MedGenome will not be liable.

MedGenome prefers to send the genetic test reports to the referring clinician considering the complexity of the test, patients are advised to contact the referring clinician for the test report.

Patient/Guardian Authorization

By my signature below I attest to the following:

I have read and I understand the information provided on this form.

Patient Consent (sign here or on the consent document)

☐ I have read the Informed Consent document and I give permission to MedGenome to perform genetic testing as described. I also give permission for my specimen / genetic data to be used in (de-identified) studies at MedGenome to improve genetic testing for other patients.

By agreeing to this informed consent below I am confirming that I understand the benefits, risks and limitations associated with genetic testing. Furthermore, I am affirming that I recognize the seriousness of conditions for which {I/my child} am being tested, and that disease descriptions, prognoses, and treatment options have been made available to me by {my/my child's} health care provider. Finally, if I have the legal authorization to provide this informed consent on behalf of another person, I am attesting that the sample provided belongs to that person.

Patient/Guardian Name _____ _____ _____ _____

First Name | Middle Name | Last Name | Date of Birth: mm/dd/yyyy

_____ _____ _____

Patient/Guardian Signature* | Date: | Place:

Relationship with the proband _____

*Fields are mandatory