ORION
Intelligent Genomics

Neuberg DIAGNOSTICS | CENTER FOR GENOMIC MEDICINE

Test Requisition Form - Inherited Disorders

PATIENT DETAILS (In BLOCK letters)

Full Name - ☐☐

DOB - ☐☐ / ☐☐ / ☐☐☐☐ (D D / M M / Y Y Y Y)   Gender - ☐ M ☐ F   Ethnicity - ☐☐☐☐☐☐☐☐☐☐

E mail - ☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐ Contact No.- ☐☐☐☐☐☐☐☐☐☐☐☐☐☐

REFERRING CLINICIAN (In BLOCK letters)

Clinician Name - ☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐

Hospital - ☐☐

E mail - ☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐ Contact No.- ☐☐☐☐☐☐☐☐☐☐☐☐☐☐

SAMPLE DETAILS

Sample Type- ☐ Blood ☐ Amniotic Fluid ☐ CVS ☐ DNA
☐ Others - ☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐

☐ Please indicate here if this sample is prenatal (maternal cell contamination recommended for molecular tests)

Gestational age - ☐☐ wks (for fetal sample)

☐ Please indicate here if this sample needs a stat/urgent report (Rush charge may apply)

TEST REQUESTED

NGS based analysis

☐ Single gene by NGS - ____________________________
(Please specify gene of interest)

☐ Mitochondrial genome

☐ ORION - ___________________________________
(Phenotype based Whole Exome +CNV Analysis) (Please specify Phenotype)

☐ Mitochondrial genome(Analysis performed with ORION when specified)

☐ ORION Focus - _________________
(Pre designed disease specific genes)

☐ Scale up to ORION

☐ Whole Genome Sequencing

Non NGS Molecular based testings

☐ MLPA ☐ Digital PCR ☐ Sanger Analysis Sequencing

☐ Others - ☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐☐

Neuberg Center for Genomic Medicine
"KEDAR" Opposite Krupa Petrol Pump, Near Parimal Garden, Ahmedabad - 380 006 I Phone: +91- 79 - 4040 8181 / 6161 8181 / 2640 8181
Email: ncgm@neubergdiagnostics.com I CIN : U85195GJ2013PTC077365,2013-14 I Web: www.supratechlabs.com/orion

Clinical details/Pedigree (relevant documents can be emailed to ncgm@neubergdiagnostics.com)

Diagnosis: _____

(Please provide detailed clinical information including Age of symptom, disease progression , current status, response to treatment, presence of consanguinity, family history and investigations performed. Additional copies of this page as required for additional clinical information.)

Full Name	DOB	Relationship	Affected	If tested in Neuberg Supratech
1)				
2)				
3)				
4)				

Signature:

Name:

Relationship to patient:

Clinician Signature:

Clinician Name :

Date, time and place:

Neuberg Center for Genomic Medicine
"KEDAR" Opposite Krupa Petrol Pump, Near Parimal Garden, Ahmedabad - 380 006 I Phone: +91- 79 - 4040 8181 / 6161 8181 / 2640 8181
Email: ncgm@neubergdiagnostics.com I CIN : U85195GJ2013PTC077365,2013-14 I Web: www.supratechlabs.com/orion

CONSENT/ASSENT FORM FOR GENETIC TESTING

Patient Name: _____ Guardian Name: _____
(In case of minor)

Information on Genetic Testing:

Variations in human genes and chromosomes often lead to genetic disorders. Genetic tests are recommended by your referring clinician with an aim to identify these disease causing variations either in genes or chromosomes with respect to the patient symptoms and/or family history.

Next Generation Sequencing (NGS) based testing allows simultaneous assessment of multiple genes.

The various tests included in this category are:

1) **Single gene via NGS**: Analysis is limited to protein coding regions of the gene of interest only.

2) **ORION Focus*:** Testing of a pre-designed set of disease specific genes. Disorders associated with maximum of 20 genes only are included in analysis.

3) **ORION:** A customized phenotype based analysis on a whole exome backbone.
Only protein coding regions of genes which are well associated with a particular phenotype/genes requested by your referring clinician are analyzed in this test. Copy Number Variations will be analyzed, however may have to be validated by another technology.
Mitochondrial Genome sequencing is included in the analysis when requested.

4) **Whole Genome:** Testing of coding as well as non coding portions of all genes irrespective of co-relation with human disease.

5) **Mitochondrial Genome**: Mitochondrial disorders originate from variants in nuclear DNA or mitochondrial DNA (mtDNA) and result in a spectrum of pathological conditions. Mitochondrial genome testing involves testing of point mutations within mitochondrial genome only.

6) **Trio testing**: involves simultaneous genetic analysis by any of the above tests in three individuals (usually index case/ proband + parents). Though multiple samples are analyzed, a single comprehensive report will be issued for better understanding of familial contribution.

Variant interpretation and test results:

Variants are interpreted and scored according to a proprietary algorithm- ORIONSeek which incorporates the criteria defined by the American College of Medical Genetics.

Only variants related to patient phenotype are reported. Benign and likely benign variants are not reported. Since the criteria emphasizes on variant classification as opposed to disease

*For any queries, please contact lab.

Neuberg Center for Genomic Medicine
"KEDAR" Opposite Krupa Petrol Pump, Near Parimal Garden, Ahmedabad - 380 006 I Phone: +91- 79 - 4040 8181 / 6161 8181 / 2640 8181
Email: ncgm@neubergdiagnostics.com I CIN : U85195GJ2013PTC077365,2013-14 I Web: www.supratechlabs.com/orion

confirmation, detection of a variant may not always translate to confirmation of the diagnosis. For example: detection of a pathogenic heterozygous (single) variation in a phenotypically matched autosomal recessive disorder does not confirm the diagnosis of the same in affected individual.

Since the ACMG criteria are not purely objective, inter-laboratory variation in classification is known to occur. Similarly, variant classification may change over time, subject to accumulation of scientific information. Hence it is requested to re-contact the laboratory for any new updates periodically, especially before contemplating prenatal testing or screening of "at risk" relatives.

Data for variants unrelated to the phenotype can be provided to your health care provider if desired (additional charges may apply for the same).

Incidental findings: indicates the presence of variants in a designated set of genes as per the ACMG Secondary findings committee. These genes have been selected based on the benefit of early intervention. Variants in these genes are usually unrelated to patient phenotype. The gene content is updated periodically by ACMG and may vary across reports analyzed at different time periods. Currently the laboratory reports only pathogenic/likely pathogenic variants in these genes if desired. Analysis of incidental genes is performed only when requested.

Expected test results:

A) **Positive**: detection of a disease causing pathogenic/likely pathogenic variation. While this confirms the presence of a disease causing variation, it might NOT always translate into diagnosis as mentioned above.

B) **Negative**: No variants related to patient phenotype were detected (refer to test limitations)

C) **Variants of unknown significance**: implies detection of a variant whose significance is not known. The variant may or may not cause disease. Re-classification may be possible after segregation studies, ancillary testing, phenotype evolution and accumulation of further variant specific/related data in medical literature. It is recommended to contact the laboratory for periodic review of variant classification especially before considering prenatal testing/ carrier screening.

D) **Copy number variations:** Though the test analyzes phenotypically significant copy number variations, they may be reported as variants of unknown significance until confirmed by an alternative test methodology.

Neuberg Center for Genomic Medicine
"KEDAR" Opposite Krupa Petrol Pump, Near Parimal Garden, Ahmedabad - 380 006 I Phone: +91- 79 - 4040 8181 / 6161 8181 / 2640 8181
Email: ncgm@neubergdiagnostics.com I CIN : U85195GJ2013PTC077365,2013-14 I Web: www.supratechlabs.com/orion

Limitations of genetic testing:

A negative test result does not always exclude a genetic disorder. In some cases the test may not detect a variation even though present in a protein coding area because of limitation in technology/scientific information.

The current technology does not standardly analyze intronic variants, non-variant splice nucleotides, repeat expansions and methylation abnormalities. Similarly coverage of gene promoters regions may not be uniform or universal.

The accuracy of genetic test results is dependent of the information provided with relation to biological relation, clinical history and sample collection and transport. Contamination may interfere with results. In rare cases due to insufficient DNA quantity or quality, a repeat sample may be required. The laboratory usually ensures timely dispatch of reports, however certain un-anticipated delays may occur for which the laboratory cannot be held liable.

The reports are released to your referring clinician as well as the patient/guardian (in case of minor). Since genetic test results are confidential, reports/ information regarding the results will not be released to any other person/clinician unless consent is provided by the patient.

- ☐ I have read and understood the above/have been explained the above in a language of my understanding and permit NCGM to perform the recommended genetic analysis.
- ☐ I understand that the data derived from my genetic testing may be stored indefinitely as a part of the laboratory database. This data is always stored in de-identified form.
  I understand my de-identified data may be used for research collaborations as well as scientific presentations and publications to further existing medical knowledge.
- ☐ I do NOT consent to the reporting of incidental findings.

Signature:

Name:

Relationship to patient:

Clinician Signature:

Clinician Name :

Date, time and place:

Neuberg Center for Genomic Medicine
"KEDAR" Opposite Krupa Petrol Pump, Near Parimal Garden, Ahmedabad - 380 006 I Phone: +91- 79 - 4040 8181 / 6161 8181 / 2640 8181
Email: ncgm@neubergdiagnostics.com I CIN : U85195GJ2013PTC077365,2013-14 I Web: www.supratechlabs.com/orion