Neuberg DIAGNOSTICS | CENTER FOR GENOMIC MEDICINE

TEST REQUISITION FORM

ORION Intelligent Genomics

MOLECULAR GENETIC TESTING

PATIENT DETAILS
(In BLOCK letters)

Full Name

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

E-mail ID Contact No.

Height Weight Blood Type

REFERRING CLINICIAN
(In BLOCK letters)

Clinician Name

Hospital

E-mail ID Contact No.

SAMPLE DETAILS

Sample Type

☐ Blood (4 ml EDTA) ☐ Amniotic Fluid (20 ml) ☐ CVS (10-15 ug) ☐ DNA [1000 ng (20 ul x 50 ng)] [Source : .....]

☐ Dried Blood Spot ☐ Others -

☐ Prenatal Sample: Gestational age ☐☐ wks ☐☐ days
(* Maternal cell contamination is mandatory for any molecular tests - AF/CVS/POC/Cord Blood)

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

TEST REQUESTED

NGS based Tests (Tick appropriately: ☐ Single ☐ Duo ☐ Trio)

☐ ORION Single gene
(Requested for..... gene)

☐ ORION Focus _____
(Pre designed disease specific gene panel)
*Please contact lab for gene list & panel details

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

☐ Scale up to ORION
(In case of Orion single gene negative report)

☐ ORION Plus _____
(Phenotype based Whole Exome + CNV Analysis + Mitochondrial Genome Sequencing)

☐ Mitochondrial Genome Sequencing

☐ Whole Genome Sequencing

Non NGS based Tests

☐ MLPA (Requested for..... gene)
* Please contact lab for kit availability

☐ Digital PCR

☐ Sanger Sequencing
(Requested for..... gene / ..... variant)

☐ Microarray ☐ 315K- Cytoscan Optima (Detects deletions upto 1Mb and duplications upto 2Mb in size)
☐ 750K- Deepdive (Detects deletions and duplications upto 200kb in size)

☐ Others

Neuberg Centre for Genomic Medicine (NCGM)
Near GTPL House, Opp. Armedia, Sindhu Bhavan Road, Bodakdev, Ahmedabad 380059
Phone: +91-6357244307, 079-61618111 | Email: contact@ncgmglobal.com | Web: www.ncgmglobal.com

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TEST REQUISITION FORM

CLINICAL DIAGNOSIS

Clinical Details / Pedigree :

(Please provide detailed clinical information including age of onset of symptoms, disease progression, current status, response to treatment, presence of consanguinity, family history and relevant investigations performed.)

(Relevant documents can be emailed to gc.ncgm@supratechlabs.com)

Details of additional samples of other family members sent

	Name	DOB / Age	Relationship (with patient)	Affected (Yes / No)	Details
1)					
2)					
3)					
4)					

Name:

Signature:

Relationship to Patient:

Date, Time and Place:

Clinician Name & Signature:

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CONSENT/ASSENT FORM

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.

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

Test Categories

a) **ORION (Single gene) :** Analysis is limited to protein coding regions of the gene of interest only.

b) **ORION Focus*:** Testing of a pre-designed set of disease specific genes.

c) **ORION:** A customized phenotype based analysis on a whole exome backbone. Only protein coding regions of genes including nuclear mitochondrial 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 this may have to be validated by another non-NGS technology.

d) **ORION Plus:** A customized phenotype based analysis on a whole exome backbone. Only protein coding regions of genes including nuclear mitochondrial 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 this may have to be validated by another non-NGS technology. Mitochondrial Genome sequencing is included in the analysis.

e) **Scale upto ORION:** In case of a negative ORION single gene report the test can be scaled upto ORION.

f) **Whole Genome:** Testing of coding as well as non coding portions of all the genes (approx 22,000) irrespective of co-relation with human disease.

g) **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.

h) **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 & Test Results

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

b) Only variants related to the patient phenotype are reported. Benign and likely benign variants are not reported.

c) 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-connect the laboratory for any new updates periodically, especially before contemplating prenatal testing or screening of "at risk" relatives.

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

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 uncertain 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 variation:** Though the test analyzes phenotypically significant copy number variations, they may be reported as variants of unknown significance until confirmed by an alternative validated by an alternative Non-NGS test methodology.

e) **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.

Limitations of Genetic Testing

a) 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.

b) 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.

c) The accuracy of genetic test results is dependent of the information provided with relation to biological relation, ship clinical history and sample collection and transport. Contamination may interfere with results.

d) In rare cases due to insufficient DNA quantity or quality, a repeat sample may be required.

e) The laboratory usually ensures timely dispatch of reports, however certain unanticipated 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/have been explained the above in 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/sample may be used for research collaborations as well as scientific presentations and publications.
- ☐ I do NOT consent to the reporting of incidental findings.

Name: Signature:

Relationship to Patient: Date, Time and Place:

Clinician Name & Signature:

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