Name:	**Dummy**	Case ID:	**XXX**
Age:	**16 Years**	Sample Type:	**Blood**
Sex:	**Male**	Sample Receipt Date:	**XXX**
Referring Clinician:	**XXX**	Reporting Date:	**XXX**
Test Requested:	**Whole Exome Sequencing (WES)**		

CLINICAL INFORMATION/HISTORY

Dummy, a 16-year-old male born to a non- consanguineous couple, presented with the complaints of Progressive vision loss, left eye- retinal detachment, left eye complete opacification of cornea leucomatous scar with vascularisation, right eye macula and fundus grossly tessellated, nystagmus, elongated face, high arch palate, wrist sign positive, anteverted pinna, scoliosis, pectus excavatum, left eye prepthismical eyes, high myopia, ectopia lentis, left eye- shrunken eye, right eye- low reflective membrane seen in vitreous cavity.
Age of Onset: Congenital.
Family History: Proband's younger brother is affected with similar symptoms and maternal grandmother has psychological issues.
Clinician's Suspicion: Traboulsi syndrome

RESULT SUMMARY

Likely Pathogenic variants in the *COL18A1* gene, probably causative of the reported phenotype was identified.
***Correlation with clinical profile and family history is required.**

VARIANTS RELEVANT TO INDICATION FOR TESTING

Likely pathogenic variants in the *COL18A1* gene were identified in this individual. No other variants of relevance to the indication were identified. Please see below for more detailed variant information.

Gene & Transcript	Variant	Zygosity	Location	Disorder	Inheritance	ACMG Classification
COL18A1 NM_030582.4	c.4063_4064delCT p.Leu1355Valfs*72	Heterozygous	Exon 39	Knobloch syndrome type 1 [OMIM ID: 267750]	Autosomal Recessive	Likely pathogenic PVS1, PM2 & PP5
	c.1893_1911del p.Gly632Aspfs*9	Heterozygous	Exon 10			Likely pathogenic PM2 & PVS1

Note: ***Two variants to cause the phenotype in a likely compound heterozygous inheritance have been identified in the COL18A1 gene; parental segregation analysis is recommended to confirm the same. Sanger validation of both COL18A1 variant is recommended in the proband due to low allele read depth.***

DETAILED VARIANT INFORMATION (VARIANTS RELEVANT TO INDICATION FOR TESTING)

Variant 1 - *COL18A1* Chr. 21:46930005 – Likely pathogenic:
The frameshift deletion NM_030582.4(*COL18A1*):c.4063_4064delCT (p.Leu1355Valfs*72) has been reported to ClinVar as Pathogenic with a status of (2 stars) criteria provided, multiple submitters, no conflicts (Variation ID 65410 as of 2024-01-04). The p.Leu1355Valfs*72 variant is observed in 59/194,886 (0.0303%) alleles from individuals of gnomAD All background. The p.Leu1355Valfs*72 variant is not reported in any individuals in 1kG All. This variant is predicted to cause loss of normal protein

function through protein truncation causing a frameshift mutation. The frame shifted sequence continues 72 residues until a stop codon is reached. This variant is a frameshift variant which occurs in an exon of *COL18A1* upstream of where nonsense mediated decay is predicted to occur. This variant has been previously classified as pathogenic, indicating that the region is critical to protein function. There are 7 downstream pathogenic loss of function variants, with the furthest variant being 50 residues downstream of this variant. This indicates that the region is critical to protein function. The p.Leu1355Valfs*72 variant is a loss of function variant in the gene *COL18A1*, which is intolerant of Loss of Function variants, as indicated by the presence of existing pathogenic loss of function variant NP_085059.2:p.V303* and 86 others. For these reasons, this variant has been classified as **Likely Pathogenic.**

Variant 2 - *COL18A1* Chr. 21:46900020 – Likely pathogenic:
The frameshift deletion NM_030582.4(*COL18A1*):c.1893_1911del (p.Gly632Aspfs*9) has not been reported previously on a disease database like ClinVar or in the disease database literature, to our knowledge. The p.Gly632Aspfs*9 variant is not reported in any individuals in gnomAD All. The p.Gly632Aspfs*9 variant is not reported in any individuals in 1kG All. This variant is predicted to cause loss of normal protein function through protein truncation causing a frameshift mutation. The frame shifted sequence continues 9 residues until a stop codon is reached. This variant is a frameshift variant which occurs in an exon of *COL18A1* upstream of where nonsense mediated decay is predicted to occur. There are 100 downstream pathogenic loss of function variants, with the furthest variant being 773 residues downstream of this variant. This indicates that the region is critical to protein function. The p.Gly632Aspfs*9 variant is a loss of function variant in the gene *COL18A1*, which is intolerant of Loss of Function variants, as indicated by the presence of existing pathogenic loss of function variant NP_085059.2:p.V303* and 86 others. For these reasons, this variant has been classified as **Likely Pathogenic. Sanger validation of the *COL18A1* variants is recommended in the proband due to low allele read depth.**

Knobloch syndrome type 1 [OMIM ID: 267750]:
Knobloch syndrome type 1 (KNO1) is caused by homozygous or compound heterozygous mutation in the *COL18A1* gene (120328) on chromosome 21q22. Knobloch syndrome-1 (KNO1) is an autosomal recessive developmental disorder primarily characterized by typical eye abnormalities, including high myopia, cataracts, dislocated lens, vitreoretinal degeneration, and retinal detachment, with occipital skull defects, which can range from occipital encephalocele to occult cutis aplasia (summary by Aldahmesh et al., 2011).

FINDINGS UNRELATED TO PHENOTYPE

This section provides information on variants identified which are unrelated to the provided phenotype.

ACMG Secondary Findings

No clinically relevant variants associated with the ACMG recommended secondary list of genes were found in the sequence data.

Incidental Findings

No variants were detected as incidental findings in the sequenced data which may not be associated with the diagnostic indication for which the sequencing test was performed.

Carrier Status in the genes related to disease

No Pathogenic or Likely Pathogenic variants were detected.

RECOMMENDATIONS

Based on the clinical features and the observed genetic findings the following have been recommended:

1. Genetic counseling is recommended to discuss the potential clinical implications of this result.
2. **Clinical/ Genotype-phenotype correlation is strongly recommended.**
3. **Sanger validation of identified variant(s) in the proband and segregation analysis in the parents, affected sibling, and close relatives is recommended.**

4. **Re-analysis of whole exome sequencing data can be done if additional phenotype is provided, and results may change/differ on re-analysis depending on the provided phenotype.**
5. **If the clinician suspects for copy number variations as a cause of the patient's phenotype then additional testing with chromosomal microarray is recommended with better sensitivity and specificity for the detection of copy number variants.**
6. If the above results do not correlate completely with patient phenotype, additional testing is advised based on the clinician's recommendation.

REPORTED VARIANTS STATISTICS:

Gene/Transcript	Variant	Depth	Allelic Depth	Alternate Allele Fraction	dbSNP rsID
COL18A1 NM_030582.4	c.4063_4064delCT	18X	8X	0.44	rs398122391
COL18A1 NM_030582.4	c.1893_1911del	14X	10X	0.71	NA

DATA STATISTICS

Total data generated (Gb)	**6.2**
Reads that passed alignment (%)	**99.15**
Data > Q30 (%)	**91.83**

METHODOLOGY

Sequencing of the protein coding regions of approximately 41Mb of the human exome (targeting approximately 99% of regions in CCDS and RefSeq) was performed using Illumina NovaSeq platform at a mean depth of 100-150X and >90% of bases covered at 30X depth >90% in the target region. The individual's DNA was extracted and fragmented, with fragments from the coding regions of the selected gene panel targeted for amplification and sequencing. Reads from the sequence output were aligned to the human reference genome (GRCh37) using the Burrows-Wheeler Aligner (BWA). Duplicate reads identification and removal, base quality recalibration and re-alignment of reads based on indels were done using inbuilt DRAGEN bio-IT pipeline. Variants to the reference were called using the Genomic Analysis Tool Kit (GATK). The variants were annotated and filtered using the **Golden Helix VarSeq** and **Varsome** analysis workflow implementing the ACMG guidelines for interpretation of sequence variants. This includes comparison against the gnomAD population catalogue of variants in 123,136 exomes, the 1000 Genomes Project Consortium's publication of 2,500 genomes, the NCBI ClinVar database of clinical assertions on variant's pathogenicity and multiple lines of computational evidence on conservation and functional impact. All variants with minor allele frequency (MAF) of less than 1% in gnomAD database, and disease-causing variants reported in **HGMD**, in ClinVar are considered. The investigation for relevant variants is focused on coding exons and flanking +/-10 intronic nucleotides of genes with clear gene-phenotype evidence (based on OMIM information). All potential modes of inheritance patterns are considered. In addition, provided family history and clinical information are used to evaluate identified variants with respect to their pathogenicity and causality. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

VARIANT ASSESSMENT PROCESS

The following databases and in-silico algorithms are used to annotate and evaluate the impact of the variant in the context of human disease: 1000 genomes, gnomAD, ClinVar, OMIM, dbSNP, NCBI RefSeq Genes, ExAC Gene Constraints, VS-SIFT, VS-PolyPhen2, PhyloP,

GERP++, GeneSplicer, MaxEntScan, NNSplice, PWM Splice Predictor. Analysis was reported using the HGVS nomenclature (www.hgvs.org/mutnomen) as implemented by the VarSeq transcript annotation algorithm. The reported transcript matches those used most frequently by the clinical labs submitting to ClinVar.

LIMITATIONS

It should be noted that this test is limited to a limited number of genes and does not include all intronic and non-coding regions. This report only includes variants that meet a level of evidence threshold for cause or contribution to disease. Certain classes of genomic variants are also not covered using the NGS testing technology, including triplet repeat expansions, copy number alterations, translocations and gene fusions or other complex structural rearrangements. More evidence for disease association of genes and causal pathogenic variants is discovered every year, and it is recommended that genetic variants are re-interpreted with updated software and annotations periodically.

VARIANT CLASSIFICATION BASED ON ACMG RECOMMENDATIONS

Genetic test results are reported based on the recommendations of American College of Medical Genetics (ACMG) as described below [1]

Variant	A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).
Pathogenic	A disease-causing variation in a gene which can explain the patients' symptoms.
Likely pathogenic	A variant which is very likely to contribute to the development of disease. However, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity
Variant of uncertain significance	A variant which is difficult to classify either as pathogenic (disease causing) or benign (non-disease causing) based on current available scientific evidence.

ACMG Criteria for classifying Variants.

Very Strong (PVS1)	
PVS1	Null variant (nonsense, frameshift, canonical ±1 or 2 splice sites, initiation codon, single or multi-exon deletion) in a gene where LOF is a known mechanism of disease.
Strong (PS)	
PS1	Same amino acid change as a previously established pathogenic variant regardless of nucleotide change
PS2	De novo variant (both maternity and paternity confirmed) in a patient with the disease and no family history.
PS3	Well-established in vitro or in vivo functional studies supportive of a damaging effect on the gene or gene product.
PS4	The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls.
Moderate (PM)	
PM1	Located in a mutational hot spot and/or critical and well-established functional domain (e.g., active site of an enzyme) without benign variation
PM2	Absent from controls (or at extremely low frequency if recessive) in reputed databases.

PM3	Variant (one of the compound heterozygous), is segregating with a pathogenic variant with known phase after testing of parents.
PM4	An in-frame deletions/insertions in non-repeat region or stop-loss can alter the protein length.
PM5	A novel missense change at the same amino acid residue where a pathogenic missense variant has already been determined.
PM6	De novo, without testing in the family.
Supporting (PP)	
PP1	A variant in known gene for a disease which is co-segregating in multiple affected family members
PP2	Missense variants are a common mechanism of disease in a gene which has low benign missense variants.
PP3	A deleterious effect of the variant is predicted by multiple lines of computational evidence (conservation, evolutionary, splicing impact, etc.)
PP4	Patient's phenotype or family history is highly specific for a disease with a single genetic etiology.
PP5	Reputable source recently reported the variant as pathogenic, but the evidence is not available to the laboratory to perform an independent evaluation.

DISCLAIMER

- In accordance with the Pre-Conception and Pre-Natal Diagnostic Testing (PCPNDT) Act, 2003- Govt. of India; Lab does not disclose the gender of the fetus.
- Prenatal genetic testing or pre-implantation genetic diagnosis is not recommended for the variants reported as variants of uncertain significance (VUS).
- 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. The classification of variants can change over time and the laboratory cannot be held responsible for this. Re-analysis of variants in previously issued reports considering new evidence is not routinely performed but may be available upon request.
- Negative results do not completely exclude the risk/carrier status for these disorders tested (residual risk)
- The sensitivity of this assay to detect large deletions/duplications of more than 10bp or copy number variations (CNV) is 70-75%. The CNVs detected must be confirmed by an alternate method.
- Due to inherent technological limitations of the assay, not all bases of the exome 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 analyzed but have not been detected. The variants not detected by the assay that was performed may impact the phenotype.
- It is also possible that a pathogenic variant is present in a gene that was not selected for analysis and/or interpretation in cases where insufficient phenotypic information is available.
- 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.

- The mutations have not been validated/confirmed by Sanger sequencing.
- Incidental or secondary findings (if any) that meet the ACMG guidelines [2] can be given upon request.
- The report shall be generated within turnaround time (TAT), however, such TAT may vary depending upon the complexity of test(s) requested. Laboratory under no circumstances will be liable for any delay beyond aforementioned TAT.
- It is hereby clarified that the report(s) generated from the test(s) do not provide any diagnosis or opinion or recommend any cure in any manner. Laboratory 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. Laboratory hereby disclaims all liability arising in connection with the report(s).
- In a very few cases genetic tests may not show the correct results, e.g., because of the quality of the material provided to the laboratory. In cases where any test provided by the laboratory fails for unforeseeable or unknown reasons that cannot be influenced by the laboratory in advance, the laboratory shall not be responsible for the incomplete, potentially misleading, or even wrong result of any testing if such could not be recognized by the laboratory in advance.
- This is a laboratory developed test and the development and the performance characteristics of this test was determined by the laboratory.

REFERENCES

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Conditions for Reporting

1. It is presumed that the specimen belongs to the patient named or identified, such verification being carried out at the point of generation of said specimen.
2. A test might not be performed due to following reasons:
   a. Specimen quantity not sufficient (Inadequate collection/spillage during transit).
   b. Specimen quality not acceptable (Hemolysis/clotted/lipemic).
   c. Incorrect sample type.
   d. Test canceled either on request of patient or doctor.
3. In any of the above cases a fresh specimen will be required for testing and reporting.
4. The results of the tests may vary from lab to lab, time to time for the same patient.
5. The reported results are dependent on individual assay methods, equipment, method sensitivity, specificity and quality of the specimen received.
6. Partial representation of the report is not allowed.
7. The reported tests are for the notification of the referring doctor, only to assist him/her in the diagnosis and management of the patient.
8. Report with status "Preliminary" means one or more tests are yet to be reported.
9. This report is not valid for Medico Legal Purpose.
10. Applicable Jurisdiction will be of "Delhi" for any dispute/claim concerning the test(s) & results of the test(s).

Name:	**Dummy**	Case ID:	**XXX**
Age:	**19 Years**	Sample Type:	**Blood**
Sex:	**Male**	Sample receipt date:	**XXX**
Referring Clinician:	**Dr. XXX**	Reporting date:	**XXX**
Test Requested:	**Whole Genome Mitochondrial NGS analysis**		

CLINICAL HISTORY:

Dummy, a 19-year-old male came for the genetic evaluation for LHON.

Clinician's Suspicion: ?LHON (Leber hereditary optic neuropathy).

VARIANTS RELEVANT TO INDICATION FOR TESTING

A *likely pathogenic* variant in the *MT-ND4* gene was identified in this individual. No other variants of relevance to the indication were identified. Please see below for more detailed variant information.

Gene & Transcript	Variant	Zygosity	Location	Disorder	Inheritance	ACMG Classification
MT-ND4	c.1019G>A p.Arg340Hisfs*121	Homoplasmy	Exon 1	Leber hereditary optic neuropathy [OMIM ID: 535000]	Mitochondrial Inheritance	Likely pathogenic PM2, PM1, PP5, & PM5

DETAILED VARIANT INFORMATION (VARIANTS RELEVANT TO INDICATION FOR TESTING)

***MT-ND4* Chr. MT:11778 – Likely pathogenic:**

The missense variant *MT-ND4*(*MT-ND4*):c.1019G>A (p.Arg340Hisfs*121) causes a change at the same amino acid residue as a previously established pathogenic variant. This variant was found in ClinVar (Variant 9708) with a classification of Pathogenic and a review status of 3 stars, reviewed by an expert panel. The p.Arg340Hisfs*121 variant is not reported in any individuals in gnomAD All. The p.Arg340Hisfs*121 variant is not reported in any individuals in 1kG All. There is a small physicochemical difference between arginine and histidine, which is not likely to impact secondary protein structure as these residues share similar properties. 2 variants within 6 amino acid positions of the variant p.Arg340Hisfs*121 have been shown to be pathogenic, while none have been shown to be benign. For these reasons, this variant has been classified as **Likely Pathogenic.**

Leber hereditary optic neuropathy [OMIM ID: 535000]:

Leber optic atrophy, also known as Leber hereditary optic neuropathy (LHON), can be caused by mutation in multiple genes encoded by the mitochondrial genome (mtDNA). Leber hereditary optic neuropathy (LHON) typically presents in young adults as bilateral painless subacute visual failure. The peak age of onset in LHON is in the second and third decades of life, with 95% of those who lose their vision doing so before age 50 years. Very rarely, individuals first manifest LHON in the seventh and eighth decades of life. Males are four to five times more likely to be affected than females, but neither sex nor mutational status significantly influences the timing and severity of the initial visual loss (GeneReviews).

.0001 LEBER OPTIC ATROPHY -MTND4, LHON11778A: The allele changes the highly conserved arginine at amino acid 340 to a histidine (R340H). This allele accounts for over 50% of Leber hereditary optic neuropathy (LHON; 535000) cases among Caucasians and over 90% of the cases in Asians. The mutation has not been observed in random population controls, may be either homoplasmic or heteroplasmic within families, and has been shown to have arisen multiple times on different mtDNA haplotypes in association with the disease (Wallace et al., 1988; Singh et al., 1989). In families harboring this mutation, approximately 33 to 60% of the maternal relatives are affected and of these, about 80% are males. Visual recovery is seen in only 4% of cases. Chinnery

et al. (2001) analyzed 17 independent pedigrees that harbored the 11778G-A mutation. They made the following observations: (1) The frequency of blindness in males was related to the mutation load in the individual's blood. (2) Mothers with 80% or less mutant mtDNA in blood were less likely to have clinically affected sons than mothers with 100% mutant mtDNA in their blood. (3) Within individual lineages, changes in mutation load from one generation to the next were largely determined by random genetic drift. Phasukkijwatana et al. (2006) examined 30 unrelated pedigrees of Thai or Chinese origin with LHON and the 11778G-A mutation. Compared to Caucasian and Japanese populations with the same mutation, the pedigrees in the study showed a lower male-to-female ratio (2.6:1) of affected persons and a higher prevalence of blood heteroplasmy (37% of the pedigrees contained at least 1 heteroplasmic 11778G-A individual). The estimated overall penetrance was 37% for males and 13% for females (OMIM).

RECOMMENDATIONS

Based on the clinical features and the observed genetic findings the following have been recommended:

1. Please note that the genetic information obtained from the patient's genomic DNA was analyzed for regions of the mtDNA, and mutations in regions other than these regions have not been assessed.
2. Genetic counseling is recommended for the patient to discuss the potential clinical implications of this result.
3. **Clinical/ Genotype-phenotype correlation is strongly recommended.**
4. **Sanger validation of identified variant(s) in the proband and segregation analysis in the parents, affected and unaffected family members and close relatives is recommended.**
5. If the above results do not correlate completely with patient phenotype, additional testing is advised based on clinician's recommendation.

METHODOLOGY

DNA extraction is done by QIAamp Blood mini kit. Mitogenome is amplified in two amplicons using long range PCR kit. Amplified Amplicons were pooled in equimolar concentration and then the whole genome library was prepared by QIASeq FX DNA Library Kit. Mito-WGS library was sequenced on Novaseq 6000 to generate with 2X150 bp chemistry to generate 0.5 GB of data. The sequence reads were pre-processed to remove adapter contamination and low-quality bases using the fastp tool. The high-quality trimmed reads were mapped on rCRS mitochondrial sequence using BWA-MEM. Duplicated reads were removed using Mark Duplicate followed by a variant recalibration module incorporated in the GATK tool. Variant calling was performed using GATK mutect2 mitochondrial mode in the setting of ploidy 1. Mitochondrial haplogroup assignment and contamination check was done using haplocheck. Mitochondrial variant annotation was carried out using Varseq & further checked using web server MSeqDR mvTool.

GENES STUDIED

MT-ND1, MT-ND2, MT-ND3, MT-ND4L, MT-ND4, MT-ND5, MT-ND6, MT-CYB, MT-CO1, MT-CO2, MT-CO3, MT-ATP6,MTATP8, MT-RNR2, MT-RNR1, MT-RNR2, MT-TA, MT-TR, MT-TN, MT-TD, MT-TC, MT-TE, MT-TQ, MT-TG, MT-TH, MT-TI, MT-TL1, MT-TL2, MT- TK, MT-TM, MT-TF, MT-TP, MT-TS1, MT-TS2, MT-TT, MT-TW, MT-TY, MT-TV.

IMPORTANCE

1. These test results should be interpreted by the referring clinician only in conjunction with the patient's clinical history, other test results and any previous analysis of appropriate family members.
2. Only phenotype-related Pathogenic and Likely Pathogenic variations reported in the Mito Map database as well as literatures are reported. Haplogroups are not analyzed. A list of variants other than the above is available on request.
3. The classification and interpretation of all the variants in this assay reflects the current state of scientific understanding at the time this report was issued. In some instances, the classification and interpretation of such variants may change as new scientific information comes to light.

LIMITATIONS

1. This report is for research purposes only, not for use in clinical diagnostic or therapeutic applications.
2. This test has not been validated by the FDA, NABL or CAP, and it has been determined by the accrediting bodies that such validation is not required at this time.
3. The analysis is based on the clinical summary provided by the clinician.
4. DNA studies do not constitute a definitive test for the selected condition(s) in all individuals. It should be realized that there are possible sources of error. Errors can result from trace contamination, rare technical errors, rare genetic variants that interfere with analysis, recent scientific developments, and alternative classification systems. This test should be one of many aspects used by the healthcare provider to help with a diagnosis and treatment plan, but it is not a diagnosis itself.
5. The significance/classification of the variant(s) may change based on the genetic testing in the parents and other family members.
6. This test was developed, and its performance characteristics were determined by Redcliffe Life Sciences. It has not been cleared or approved by the FDA.
7. The classification of variants of unknown significance can change over time and Redcliffe Life Sciences cannot be held responsible for this. Please contact Redcliffe Life Sciences later to inquire about any changes.
8. Intronic variants are not assessed using this method.
9. Large deletions of more than 10 bp or copy number variations/chromosomal rearrangements cannot be assessed using this method.
10. Certain genes may not be covered completely, and few variants could be missed. Variants not detected by the assay that was performed may impact the phenotype.
11. The variants have not been validated by Sanger sequencing.
12. Incidental or secondary findings (if any) that meet the ACMG guidelines can also be given upon request.
13. Variants that have coverage below ≤ 20 are not taken into consideration.

---------- End Of Report ----------

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   c. Incorrect sample type.
   d. Test canceled either on request of patient or doctor.
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9. This report is not valid for Medico Legal Purpose.
10. Applicable Jurisdiction will be of "Delhi" for any dispute/claim concerning the test(s) & results of the test(s).

Disclaimer: Method given in report are only indicative and can be changed depending upon type of machine and kit available at time of testing.

Not all tests at all locations are under NABL scope. Availability of tests under NABL scope varies from lab to lab.

Terms and Conditions of Reporting

1. The presented findings in the Reports are intended solely for informational and interpretational purposes by the referring physician or other qualified medical professionals possessing a comprehensive understanding of reporting units, reference ranges, and technological limitations. The laboratory shall not be held liable for any interpretation or misinterpretation of the results, nor for any consequential or incidental damages arising from such interpretation.

2. It is to be presumed that the tests performed pertain to the specimen/sample attributed to the Customer's name or identification. It is presumed that the verification particulars have been cleared out by the customer or his/her representation at the point of generation of said specimen / sample. It is hereby clarified that the reports furnished are restricted solely to the given specimen only.

3. It is to be noted that variations in results may occur between different laboratories and over time, even for the same parameter for the same Customer. The assays are performed and conducted in accordance with standard procedures, and the reported outcomes are contingent on the specific individual assay methods and equipment(s) used, as well as the quality of the received specimen.

4. This report shall not be deemed valid or admissible for any medico-legal purposes.

5. The Customers assume full responsibility for apprising the Company of any factors that may impact the test finding. These factors, among others, includes dietary intake, alcohol, or medication / drug(s) consumption, or fasting. This list of factors is only representative and not exhaustive.