सी.डी.एफ.डी. CDFD
डी एन ए फिंगरप्रिंटिंग एवं निदान केन्द्र
(जैव प्रौद्योगिकी विभाग, विज्ञान एवं प्रौद्योगिकी मंत्रालय, भारत सरकार का स्वायत्त संस्थान)
CENTRE FOR DNA FINGERPRINTING AND DIAGNOSTICS
(An autonomous Institute of the Dept. of Biotechnology, Ministry of Science & Technology, Govt. of India)
प्रयोगशाला ब्लॉक : तुलजागुडा, (एम जे मार्केट के सामने), नामपल्ली, हैदराबाद - 500 001, भारत
Laboratory Block : Tuljaguda, (Opp. M.J. Market), Nampally, Hyderabad - 500 001, India
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कार्यालय ब्लॉक : भवन 7, गृहकल्पा, 5-4-399/B, नामपल्ली, हैदराबाद - 500 001, भारत
Office Block : Bldg. 7, Gruhakalpa, 5-4-399/B, Nampally, Hyderabad - 500 001, India

BIOCHEMICAL REPORT

Name of the patient	Rushikesh Reddy	Age/Sex	16yr/M
Date of collection	18.08.16	Date of reporting	29.08.16
CDFD No	3051	Ref by	NIMS
Method of analysis	HPLC	NIMS No.	160810519

Test	Result	Normal Range
Plasma Homocysteine	**60.0**umol/L	< 14.0 umol/L
Plasma cysteine	190 umol/L	70 -250 umol/L

Suggested Clinical Correlation

Prajnya
DrPrajnya R
I/C Biochemical Genetics

Prajnya
for Dr.Ashwin Dalal
Head, Diagnostics Division

DIAGNOSTIC REPORT

SRL Diagnostics

Cert. No. M-0025

CLIENT CODE : C000058070

CLIENT'S NAME AND ADDRESS :
LATHA DIAGNOSTICS
BESIDES KAPU KALYANA HOSDAPUR, OPP. MURLIDHAR REDDY HOSPITAL.
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ONGOLE 523001
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Tel : 1-800-222-000,1-800-102-8282, Fax : 022 - 67801212
CIN - U74899DL1995PLC070603
Email : srl.mumbai@srl.in

PATIENT NAME : K RUSHIKESH REDDY

PATIENT ID :

ACCESSION NO : **0002PG003782** AGE : 16 Years SEX : Male DATE OF BIRTH :

DRAWN : 30/06/2016 00:00 RECEIVED : 02/07/2016 10:18 REPORTED : 08/07/2016 11:53

REFERRING DOCTOR : DR. MAKINENI KIRAN

CLIENT PATIENT ID :

Test Report Status **Final**	Results		Biological Reference Interval	Units
APLA TOTAL				
CARDIOLIPIN ANTIBODIES, SERUM				
CARDIOLIPIN IGG	1.6		Normal Range : < 12 Equivocal Range : 12.0-18.0 Positive Results : >18.0	GPL/mL
CARDIOLIPIN IGM	1.4		Normal Range : < 12 Equivocal Range : 12.0-18.0 Positive Results : >18.0	MPL/mL
ANTI Ç2 GLYCOPROTEIN 1 IGM, SERUM				
ANTI Ç2 GLYCOPROTEIN 1 IGM	3.9		< 20 (Negative) > or = 20 (Positive)	RU/mL
METHOD : ENZYME IMMUNOASSAY				
ANTI Ç2 GLYCOPROTEIN 1 IGG, SERUM				
ANTI Ç2 GLYCOPROTEIN 1 IGG	2.0		< 20 (Negative) > or = 20 (Positive)	RU/mL
METHOD : ENZYME IMMUNOASSAY				
PTT & MIXING STUDIES, PLASMA				
PTT (TEST)	**71.5**	High	30.0 - 43.0	SECONDS
PTT CONTROL (NORMAL POOLED PLASMA)	33.1		30.0 - 43.0	SECONDS
METHOD : CLOT-BASED ASSAY				
PTT (TEST+CONTROL) 1:1	46.0			SECONDS
METHOD : CLOT-BASED ASSAY				
DRVV SCREEN TIME, PLASMA				
DRVV SCREEN (TEST)	**53.30**	High	32.82 - 48.90	SECONDS
METHOD : CLOT-BASED ASSAY				
DRVV SCREEN CONTROL	38.30		32.82 - 48.90	SECONDS
METHOD : CLOT-BASED ASSAY				
DRVV SCREEN RATIO	**1.39**	High	0.82 - 1.22	RATIO
METHOD : CLOT-BASED ASSAY				
DRVV CONFIRM, PLASMA				
DRVV CONFIRMATORY (TEST)	**46.10**	High	27.59 - 34.55	SECONDS
METHOD : CLOT-BASED ASSAY				
DRVV CONFIRMATORY (CONTROL)	34.00		27.59 - 34.55	SECONDS
METHOD : CLOT-BASED ASSAY				
DRVV CONFIRM RATIO	**1.36**	High	0.93 - 1.17	RATIO
METHOD : CLOT-BASED ASSAY				
NORMALISED RATIO [DRVV SCREEN RATIO / DRVV CONFIRM RATIO]	1.02		0.82 - 1.14	RATIO
LUPUS ANTICOAGULANT				

Comments

Lupus Anticoagulant Assay : Prolonged APTT seems to be due to reason(s) other than Lupus Anticoagulant.

Advised : Repeat APTT on a fresh sample, if clinically indicated.

Test Method(s)
CARDIOLIPIN ANTIBODIES, SERUM-Enzyme Immuno-Assay.
ANTI Ç2 GLYCOPROTEIN 1 IGM, SERUM-
ANTI Ç2 GLYCOPROTEIN 1 IgM

Antiphospholipid syndrome (APS), also known as Hughes syndrome, is an autoimmune condition associated with reccurring vascular thrombosis and pregnancy morbidity. APS is divided into primary APS where the condition occurs alone, and secondary APS where the condition occurs in association with another autoimmune disease such as Systemic Lupus Erythematosus (SLE). Antibodies targeting cardiolipin, Ç2-glycoprotein-1 (Ç2GP-1) and phosphatidylserine are commonly found in patients with the clinical symptoms of APS. Anti B2 glycoprotein antibodies can be of IgG. IgM and IgA isotype.

Test utility:
Å Autoantibodies to B2-glycoprotein (Apolipoprotein H) are associated with arterial and venous thrombosis as well as recurrent spontaneous abortion.
Å IgG and /or IgM class antibodies against Ç2GP-1B2 occur with prevalence of 30%-60% of APS cases.
Å Ç2GP-1 antibodies are only found in case of autoimmune diseases, whereas antibodies against cardiolipin can be detected in APS and in certain infections (syphilis, Borreliosis, AIDS, hepatitis, tuberculosis). Detection of these antibodies provide a serological aid for the differentiation of autoimmune diseases from infections.

Limitation :
Positive test results alone are not diagnostic and must be interpreted in conjunction with the patientõs clinical presentation and other serological markers. Patients must be tested at least twice, minimum 12 weeks apart to demonstrate antibody persistence.
ANTI Ç2 GLYCOPROTEIN 1 IGG, SERUM-
ANTI Ç2 GLYCOPROTEIN 1 IgG

Antiphospholipid syndrome (APS), also known as Hughes syndrome, is an autoimmune condition associated with reccurring vascular thrombosis and pregnancy morbidity. APS is divided into primary APS where the condition occurs alone, and secondary APS where the condition occurs in association with another autoimmune disease such as Systemic Lupus Erythematosus (SLE). Antibodies targeting cardiolipin, Ç2-glycoprotein-1 (Ç2GP-1) and phosphatidylserine are commonly found in patients with the clinical symptoms of APS. Anti B2 glycoprotein antibodies can be of IgG. IgM and IgA isotype.

Test utility:
Å Autoantibodies to B2-glycoprotein (Apolipoprotein H) are associated with arterial and venous thrombosis as well as recurrent spontaneous abortion.
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Å Ç2GP-1 antibodies are only found in case of autoimmune diseases, whereas antibodies against cardiolipin can be detected in APS and in certain infections (syphilis, Borreliosis, AIDS, hepatitis, tuberculosis). Detection of these antibodies provide a serological aid for the differentiation of autoimmune diseases from infections.

Limitation :
Positive test results alone are not diagnostic and must be interpreted in conjunction with the patientõs clinical presentation and other serological markers. Patients must be tested at least twice, minimum 12 weeks apart to demonstrate antibody persistence.
PTT & MIXING STUDIES, PLASMA-
LUPUS ANTICOAGULANT SCREEN, PLASMA

Lupus anticoagulants (LA) are autoantibodies that prolong phospholoipid based clotting assays in vitro but cause increased tendency to venous and arterial thrombosis in vivo. LA are directed against phospholipid - containing protein constituents of cells and plasma. They are an integral part of the APLA syndrome and can be associated with conditions such as SLE and other connective tissue disorders, HIV and other infections and malignancies. LA could also be drug induced and transient.

The results of two separate and independent screening tests for LA, namely dilute APTT and RVVT, need to be confirmed by neutralization assays using an excess of phospholipids. The presence of LA in the sample is confirmed if the RVVT normalized ratio is greater or equal to 1.14. Positive results need to be confirmed 12 weeks later to rule out transient LA. The result is to be interpreted in the context of the clinical setting. The patient should not be on oral anticoagulant therapy or heparin at the time of testing.

AUTOIMMUNE IFA

SLE DIAGNOSTIC PROFILE

ANTI-NUCLEAR AB-IFA, HEP2, SERUM		
ANTINUCLEAR ANTIBODIES	NEGATIVE	NEGATIVE
DSDNA (WITH TITRES)		
DNA	NEGATIVE	NEGATIVE

(Also refer to "CONDITIONS OF REPORTING" on the reverse.)

SRL Diagnostic

Cert. No. M-0025

SRL Diagnostics

CLIENT CODE : C000058070

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PATIENT NAME : K RUSHIKESH REDDY PATIENT ID :

ACCESSION NO : **0002PG003782** AGE : 16 Years SEX : Male DATE OF BIRTH :

DRAWN : 30/06/2016 00:00 RECEIVED : 02/07/2016 10:18 REPORTED : 08/07/2016 11:53

REFERRING DOCTOR : DR. MAKINENI KIRAN CLIENT PATIENT ID :

Test Report Status **Final**	Results	Biological Reference Interval	Units
ANTINEUTROPHIL CYTOPLASMIC AB(ANCA), SERUM			
P-ANCA	NEGATIVE	NEGATIVE	
C-ANCA	NEGATIVE	NEGATIVE	

Test Method(s)
ANTI-NUCLEAR AB-IFA, HEP2, SERUM-The Immunofluoresence assay is the Gold standard method for ANA testing. A negative ANA test virtually rules out a diagnosis of Systemic Lupus Erythematosus but a positive test may be indicative of a number of autoimmune connective tissue diseases such as Scleroderma, Rheumatoid Arthritis and Sjogren's syndrome.When correlated with the Clinical history & physical examination ,it identifies almost all pts. With SLE (Senstivity < 95 %). Population studies show positive ANA in approximately 1-5 % of healthy subjects. False positive results for ANA can be seen in pts. Taking certain medications like ï hydralazine , isoniazid , procainamide etc.ANA test carried out by Immunofluorescence assay using HEP-2 slide (Tissue culture substrate) is more sensitive and specific than ANA carried out by enzyme immunoassay.

TITRE
ANA positivity of greater than or equal to 1:160 titre is of clinical significance in diagnosis of Collagen Vascular Disorders. Upto 40 % of elderly subjects with chronic non-rheumatological illness have ANA positivity usually at low titre (1: 40 - 1:160)

PATTERN
The ANA pattern seen on immunofluorescence staining helps in determination of the antibody specificities which need to be confirmed by immunoblot techniques.

The positivity seen on fluorescence indicates
1+ positivity = Minimum Immunofluroscence of no significance.
2+ Positivity = Mildly positive, clinically insignificant.
3+ Positivity = Significant positive, needs clinical correlation.
4+ Positivity = Strong positive, highly suggestive of collagen vascular disease.
A titre estimation helps to monitor response to treatment.

Please refer to the following test codes for specific antibody determination by IMMUNOBLOT
1220 : Sm(SMITH) antibody # 1215 : U1SNRNP antibody
1204 : SSA antibody # 1205 : SSB antibody
1007 : SSA & SSB antibodies
1235 : Scl - 70 antibody # 1208 : Jo - 1 antibody

PLEASE NOTE: ALL ANA RESULTS WILL BE REPORTED WITH FINAL END POINT TITRE VALUE.
DSDNA (WITH TITRES)-High titres of anti-DsDNA are associated with SLE, hence its testing should be reserved only for pts. with positive ANA . Not all pts. of SLE have positive anti DsDNA ,therefore a negative anti -DsDNA does not exclude the diagnosis of SLE. The titre of the anti-n DNA may decrease with successful therapy and increase in acute recurrence of the disease. Also, DNA-anti-DNA immune complexes play a role in the pathogenesis of SLE through the deposit of the complexes in the kidney and other tissues. For these reasons, the detection and quantitation of anti-n DNA is diagnostically and therapeutically helpful in patients suspected or known to have SLE. Prevalence of anti-DsDNA antibodies among healthy people is nil or very low.
ANTINEUTROPHIL CYTOPLASMIC AB(ANCA), SERUM-Anti-Neutrophilic Cytoplasmic Antibody testing is useful in the diagnosis and treatment of autoimmune mediated vasculitides. The diagnosis is made in correlation with the Clinical history , biopsy findings & ANCA positivity .Positive ANCA with cytoplasmic pattern (C - ANCA) is seen in the cases of Wegener's Granulomatosis. The perinuclear pattern (P - ANCA) is seen in Polyarteritis Nodosa..The final diagnosis should be on the basis of criteria as given by the American College of Rheumatology Or by 2012 revised Chapel Hill consensus conference criteria.

SPECIALISED CHEMISTRY - CARDIAC MARKER

HOMOCYSTEINE, SERUM/ PLASMA				
HOMOCYSTEINE	>65.00	High	With folate supplementation < 12 Without folate supplementation < 15	Õmol/L

METHOD : CHEMILUMINESCENCE,COMPETITIVE IMMUNOASSAY

Comments

****

NOTE : RECHECKED FOR SERUM HOMOCYSTEINE.
PLEASE CORRELATE CLINICALLY.

****

Test Method(s)
HOMOCYSTEINE, SERUM/PLASMA-Homocysteine is an intermediary in the sulfur-amino acid metabolism pathways, linking the methionine cycle to the folate cycle. Inborn

Dalal
s Division

NOSTIC REPORT

SRL Diagnostics

Cert. No. M-0025

SRL Diagnostics

CLIENT CODE : C000058070

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PATIENT NAME : K RUSHIKESH REDDY PATIENT ID :

ACCESSION NO : **0002PG003782** AGE : 16 Years SEX : Male DATE OF BIRTH :

DRAWN : 30/06/2016 00:00 RECEIVED : 02/07/2016 10:18 REPORTED : 08/07/2016 11:53

REFERRING DOCTOR : DR. MAKINENI KIRAN CLIENT PATIENT ID :

Test Report Status **Final**	Results	Biological Reference Interval	Units

errors of metabolism that lead to homocysteinemia or homocysteinuria include cystathionine beta-synthase deficiency and various defects of methionine re-methylation. Genetic defects in vitamin cofactors (vitamin B6, B12, and folate) and nutritional deficiency of B12 and folate also lead to abnormal homocysteine accumulation. As Homocysteine is an indicator of acquired Folate or Vitamin B 12 deficiency, serum Folic acid or Vitamin B12 deficiency should be ruled out in case of abnormal homocysteine level.

Other factors that may influence and increase plasma homocysteine include:
Age, Smoking, poor diet/cofactor deficiencies, Chronic kidney disease/renal disease & Hypothyroidism

A high level of homocysteine makes a person more prone to endothelial injury, which leads to vascular inflammation, which in turn may lead to atherogenesis, which can result in ischemic injury. Abnormally high levels of homocysteine in the serum, above 15 Ômol/L, is a medical condition called hyperhomocysteinimia which has been claimed to be a significant risk factor for the development of a wide range of diseases, include thrombosis, neuropsychiatric illness, and fractures.

Patients taking methotrexate, nicotinic acid, Theophylline, nitrous oxide, S-adenosyl-methionine and L-dopa can have falsely elevated serum HCY levels.

Heterophilic antibodies in human serum can react with reagent immunoglobulins, interfering with in vitro immunoassays. Patients routinely exposed to animals or to animal serum products can be prone to this interference and anomalous values may be observed.

CLIENT CODE : C000058070

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CLIENT PATIENT ID :

Test Report Status **Final** **Results**

MOLECULAR BIOLOGY

FACTOR V MUTATION DETECTION

FACTOR V LEIDEN MUTATION	NOT DETECTED

Test Method(s)
FACTOR V MUTATION DETECTION-
DNA Sequencing

The Factor V Leiden point mutation, besides its well-accepted relevance for thromboembolic events, is also being associated with an increased risk for juvenile stroke, myocardial infarction in young women and traumatic conditions. It accounts for >90% of cases with APC-resistance. Between 20-50% of patients with unexplained venous thrombo embolism have this defect.
A single point mutation in the Factor V gene (1,691 G to A substitution) predicts synthesis of a Factor V molecule with arginine at amino acid residue 506 versus glutamine (wild type). This R506Q substitution prevents a peptide bond in coagulation molecule from being cleaved by activated protein C (APC).

Clinical Utility
--Evaluation of at-risk individuals and for individuals with a history of Venous thrombosis in order to initiate anticoagulant therapy. This includes individuals at increased risk of pregnancy-associated and oral contraceptive related complications.
--In addition, individuals that are positive for the Factor V Leiden mutation can be advised of the risk of thrombotic events for other family members.

Interpretation
Results are reported as:
HOMOZYGOUS , HETEROZYGOUS or MUTATION NOT DETECTED .
This assay is specific for Factor V R506Q mutation. Heterozygote carriers of the Factor V Leiden polymorphism have increased risk of thrombosis of 5 to 10 fold, without other concomitant risk factors, and an additive synergistic risk of VTE when other thrombophilic risk factors, particularly oral contraceptive, are simultaneously present.
Homozygotes have a 50 to 100 fold increased risk of having a hypercoagulable state.
This direct mutation analysis will not detect individuals with thrombophilia caused by mechanisms other than Factor V R506Q mutation.
All the results should always be correlated with clinical status and history of the patient.

Limitations
PCR is a highly sensitive technique common reasons for paradoxical results are contamination during specimen collection, selection of inappropriate specimens and inherent PCR inhibitors in the specimen.

References:
1. Lancet (1996) 347:58.
2. American Journal of Pathology (1997) 108-427-433.

Note : This test is standardized and validated at SRL Limited

****End Of Report****

Please visit www.srlworld.com for related Test Information for this accession

Dr. Simi Bhatia, MD
Lab Director-Mumbai Ref Lab

Dr. A Dasgupta, MD, PhD
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Dr. Kshama P, MD
Biochemist

Dr. Firoz Ahmad,PhD
Research Scientist and Sen Manager - R&D

gene CŌRE

Clinical Exome

Your Test Results

Case Number:	19086768
Patient Name:	K. Rushikesh Reddy
Age/Sex:	20 Yrs/Male
Patient Location:	Secunderabad
Hospital Name:	KIMS Hospital
Physician Name:	Dr. Pavani
Date & Time of Accessioning:	23/07/2019 12:38 Hrs
Date & Time of Reporting:	05/09/2019 17:42 Hrs

INTERPRETATION

Most infants and children with this conditionhave an unusually small head size (microcephaly), delayed development, and intellectual disability. Less common features of the condition include eye problems and a blood disorder called megaloblastic anemia. Megaloblastic anemia occurs when a person has a low number of red blood cells (anemia), and the remaining red blood cells are larger than normal (megaloblastic). The signs and symptoms of methylmalonic acidemia with homocystinuria worsen begins in adolescence or adulthood, the signs and symptoms usually include psychiatric changes and cognitive problems. Affected individuals can exhibit changes in their behavior and personality; they may become less social and may experience hallucinations, delirium, and psychosis. In addition, these individuals can begin to lose previously acquired mental and movement abilities, resulting in a decline in school or work performance, difficulty controlling movements, memory problems, speech difficulties, a decline in intellectual function (dementia), or an extreme lack of energy (lethargy). Some people with methylmalonic acidemia with homocystinuria whose signs and symptoms begin later in life develop a condition called subacute combined degeneration of the spinal cord, which leads to numbness and weakness in the lower limbs, difficulty walking, and frequent falls.

The detected in-frame variant c.766_771delGCCCCC (p.A256_P257del) in MMACHC gene (Sequencing coverage of coding region of gene is 100%; located on chr1:45974801 CCCGCC; Read depth: 71x), frequency has been found to be low in the general population (0.328% ExAC South Asian Frequency, 0.314% gnomAD South Asian Frequency) [6-9]. Mutations on this gene have been associated with Methylmalonic aciduria and homocystinuria, cblC type[1]. Protein length change as a result of an in-frame deletion/insertion in a non-repeat region [3,4]. The detected variant has recently been reported as disease causing in HGMD[10] and Uncertain significance (Last evaluated: Jun 14, 2016) in ClinVar[11]. According to the ACMG guidelines for variant interpretation, this variant is being classified as VUS[2].

Diagnostic Findings not related to phenotype:

No significant variant was detected in this individual.

Comments: The above mentioned result must be interpreted in context with the thorough clinical phenotype of the proband.

RECOMMENDATIONS

Genetic counselling is recommended.

Targeted testing of affected and unaffected individuals for c.766_771delGCCCCC (p.A256_P257del) in MMACHC gene is recommended.

CORE Diagnostics offers targeted analysis for family members at risk for carrying the variant identified in this individual.

Dr. Avshesh Mishra, Ph. D., Molecular Scientist

Dr. Shivani Sharma, Pathologist
Reg. No. 1906

CŌRE DIAGNOSTICS™

Your Test Results

Case Number:	19086768
Patient Name:	K. Rushikesh Reddy
Age/Sex:	20 Yrs/Male
Patient Location:	Secunderabad
Hospital Name:	KIMS Hospital
Physician Name:	Dr. Pavani
Date & Time of Accessioning:	23/07/2019 12:38 Hrs
Date & Time of Reporting:	05/09/2019 17:42 Hrs

COMMENTS

Data analysis: Only variants (SNVs/small indels) in the coding region and the flanking intronic regions (±10bp) with a minor allele frequency (MAF) <5% are evaluated. All reported variants meet internal quality control standards. The clinically relevant mutations are annotated using published variants in literature and a set of disease databases- ClinVar (2018), OMIM (2017), GWAS, HGMD (2018.1) and SwissVar. Common variants are filtered based on Minorallele frequencies in 1000Genomes (phase3v5b), dbSNP151, NHLBI Exome Sequencing Project (ESP), Exome Aggregation Consortium (0.3.19), genomeAD (2.0.1), EmVclass 2018-Q2. Only non-synonymous and splice site variants found in clinical exome panel consisting of 5200+ genes were used for clinical interpretation. Non- synonymous variants effect is calculated using multiple algorithms such as PolyPhen-2 (v2.2.2), SIFT (2016), MutationTaster2. Silent variants that do not result in any change in amino acid in the coding region are not reported.

Variants: The classification and interpretations of variants may change over time as more information about the gene association and clinical findings with respect to individual's clinical phenotype becomes available. Only variants in gene associated with the phenotype observed in this individual, or thought to be clinical relevant for the proband are reported here. These include pathogenic or likely pathogenic variants in genes described in the ACMG Recommendation for Reporting of Incidental Findings in Clinical Exome as requested by the patient[4]. Visit ACMG guideline[1] for current classification of variants.

Variants that have a population frequency greater than expected given the prevalence of the disease in the general population are considered to be benign variants. Silent variants are not reported unless known to be pathogenic or other evidence suggests potential disruption of splicing.

Note: This analysis cannot detect single and multi-exon deletions and duplications. Pathogenic variants may also be present within regions which were not analyzed (e.g. introns, promoter and enhancer regions, and long repeats). Variants are evaluated by their reported frequency[5-8].

Genetic test results are reported based on ACMG guidelines as described below:

Variant	A change in a gene that can either be disease causing (pathogenic) or not disease causing (benign)
In/Dels	It represents the insertions and deletions of bases in the genome.
Pathogenic	A genetic alteration that increases an individual's susceptibility or predisposition to a certain disease or disorder. Also known as disease causing mutation.
Likely pathogenic	A variant which is most likely to play a role in the development of the disease, however scientific knowledge is currently insufficient to prove this.
Variant of Unknown Significance (VUS)	A variant that is difficult to classify as either pathogenic or benign based on currently available scientific evidence. Further testing of patient's family members as recommended to establish the significance of the detected variant in the family. The significance can be established only with time and availability of scientific evidence.

Dr. Avshesh Mishra, Ph. D., Molecular Scientist

Dr. Shivani Sharma, Pathologist
Reg. No. 1906

1. Lerner-Ellis JP *et al.* (2009); Hum Mutat. 30(7):1072-81.

2. Richards S *et al.* (2015); Genet Med.17(5):405-24

3. Kumar P *et al.* (2009); Nat Protoc. 4(7):1073-81

4. Adzhubei IA *et al.* (2010); Nat Methods. 7(4):248-9

5. Kalia SS *et al.* (2017); Genet Med. 19(2):249-255

6. 1000 Genomes: browser.1000genomes.org/

7. NCBI dbSNP: www.ncbi.nih.gov/dbSNP

8. Exome Variant Server:evs.gs.washington.edu/EVS/EVS

9. Exome Aggregation Consortium (ExAC): exac.broadinstitute.org/

10. HGMD: www.hgmd.cf.ac.uk/ac/

11. ClinVar:www.ncbi.nlm.nih.gov/clinvar/

12. Li H *et al.* (2010); Bioinformatics. 26(5):589-95.

13. NHLBI: https:/esp.gs.washington.edu/drupal

14. ENSEMBL: www.ensembl.org/

15. Green RC *et al.* (2013); Genet Med. 15(7):565-74

16. OMIM: www.omim.org

17. GWAS: www.ebi.ac.uk/gwas/

18. dbSNP: www.ncbi.nlm.nih.gov/projects/SNP/

19. MutationTaster2: www.mutationtaster.org/

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