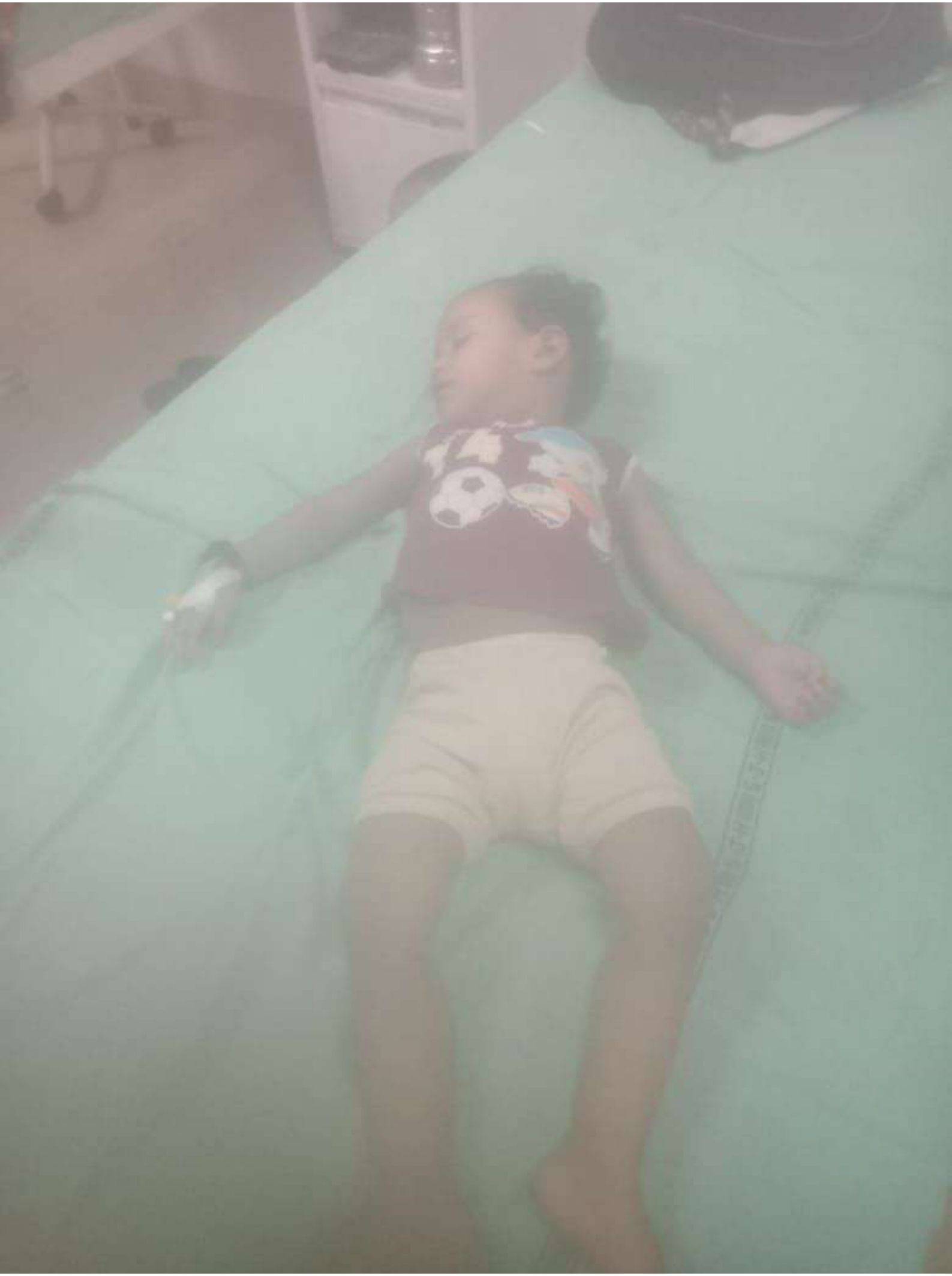


mfine
Telegynostics

LifeCell
Diagnostics

Name	[illegible] MOHD KASIM	Lab ID	[illegible]
	[illegible]	Sample Collection Date	[illegible]
Age	11 Month(s)	Sample Receipt Date	[illegible]
DOB		Reporting Date	[illegible]
Gender	Male	Location	[illegible]
Referring Physician	Dr. SITA SAHU		
Client Name	MFINE		

HBB GENE SEQUENCING FOR MUTATION ANALYSIS

Details	Remarks
Sample Type	Peripheral venous blood
Quality of Sample	Adequate
Clinical Indication	Suggestive of Beta thalassemia major and under blood transfusion
Test Requested	Beta Globinopathy

RESULTS

Variant Detected	Genotype	Allele Status	Clinical Significance
Common nomenclature: **Codons 41/42 (-TTCT)** HGVS nomenclature: **c.126_129delCTTT**	**β^0/β^0**	**Homozygous**	**Thalassemia Major**

INTERPRETATION

- **Homozygous deletion of CTTT** between codons 41 and 42 of the *HBB* gene is detected in the **provided sample**. This frameshift results in a premature stop codon at the new codon 59 terminating translation. This variant is predicted to cause loss of normal protein function either through protein truncation or nonsense-mediated mRNA decay. According to HbVar database this variant is classified as **β^0**, and the disease indication is of **Thalassemia Major**. The frequency of this variant among the affected population is found to be 13.23% in Punjabi population.

RECOMMENDATION

Kindly correlate the results with other clinical findings and Genetic counseling is recommended to understand the inheritance and risk of disease occurrence in future generations. **Parents *HBB* gene sequencing is recommended.**

CAP ACCREDITED

F-66

HISTOGENETICS
300 Executive Blvd, Ossining, New York - 10562.
Phone : 914-762-0300, Fax : 914-762-4441, Website : www.histogenetics.com
ASHI # 03-1-NY-26-2, CLIA # 33D0985173
Soo Young Yang Ph.D / Nezih Cereb M.D. Directors
Email : customerservice@histogenetics.com

ASHI ACCREDITED

HLA TYPING REPORT

Patient Information

Last Name	KASEEM	Date Of Birth	04-Apr-2023
First Name	MOHAMMAD	Histo ID	H142528
Hospital	Thalassemia Camp by Dr.Dinesh Bhurani	MR#	NA
Physician	DR DINESH BHURANI		
Diagnosis	THALASSEMIA MAJOR	Received On	10-Jun-2024
Specimen Type	Buccal Swab	Collected On	28-May-2024

A*	B*	C*	DRB1*	DRB345*	DQB1*	DQA1*	DPB1*	DPA1*
11:01:01G	40:01:01G	03:04:01G	07:01:01G	NA	03:03:02G	NA	NA	NA
33:03:01G	40:01:01G	03:04:01G	14:04:01G	NA	05:03:01G	NA	NA	NA

Typing Status : Complete

Donor(s) Results

Last Name/URD#	PARVEEN	Date Of Birth	: 16-Nov-2021
First Name	RUMAISHA	Histo ID	: H142529
Relation	Sibling	Received On	: 10-Jun-2024
Specimen Type	Buccal Swab	Collected On	: 28-May-2024
MR#	NA		

A*	B*	C*	DRB1*	DRB345*	DQB1*	DQA1*	DPB1*	DPA1*
11:01:01G	40:01:01G	03:04:01G	07:01:01G	NA	03:03:02G	NA	NA	NA
33:03:01G	40:01:01G	03:04:01G	14:04:01G	NA	05:03:01G	NA	NA	NA

Typing Status : Complete

Matching Ratio with Patient : 10/10(0/0) **Matched**

Rajiv Gandhi Cancer Institute
and Research Centre

Dated: 29-08-2024

TO WHOM SOEVER IT MAY CONCERN

This is to certify that patient Mast. Mohammad Kaseem is a diagnosed case of Thalassemia major & is currently under treatment at Rajiv Gandhi Cancer Institute and Research Center vide **CR No.348837**. He requires Allogenic Bone marrow transplant.The approximate cost of treatment at hospital is Rs 15,00,000/-(Fifteen Lakh Rupees Only).Since unforeseen emergencies & medical conditions can arise, this estimate is to be likely revised from time to time.

This letter is being issued to him to enable him to apply for Prime/Chief Minister's relief fund.

Dr. Rohan Halder
MD, DM
Haemato-Oncology

DR. ROHAN HALDER
MD, DM (Clinical Hematology)
PDCL (Hemato Oncology)
Consultant (Hemato Oncology)
RGCI&RC, Sec-5, Rohini, Delhi-85
Regn No: AM-37

Dr.Dinesh Bhurani
MD, DM, FRCPA
Haemato-Oncologist

Chromatogram Report

ver. V1.05 2024/03/08 13:29:19
HPLC 760
ID 20240308125000007 SL 0001 - 07
Sample No.
Patient ID
Name
Comment

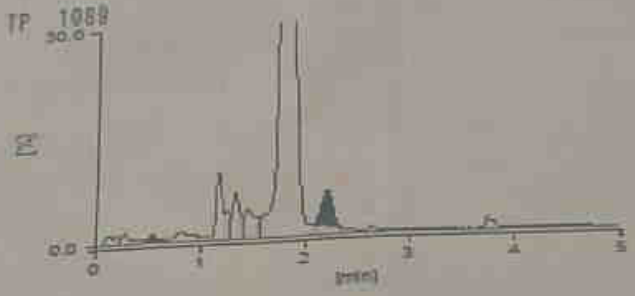
M/o Md Kalam
MID-255079
Age - 24 yrs

CAL18 F Y =1 2864X + 0 2973
A2 Y =1 3260X + 0 5582

Name	%	Time	Area
F	1 2	0 68	7 42
A0	78 0	1 08	897 56
A2	6 0	2 23	47 59
E*			
D*			
S*			
C*			

Total Area 1150 60

HbF 1 2 % HbA2 5 0 %



[Unknown Peak]

Name	%	Time	Area
P00	0 8	0 13	9 53
P01	1 0	0 27	11 24
P02	0 1	0 64	1 40
P03	1 7	0 81	19 26
P04	5 1	1 18	58 11
P05	3 2	1 34	36 52
P06	3 2	1 45	36 77

Chromatogram is suggestive of β Thalassemia trait

BIOCHEMISTRY LABORATORY REPORT
भारत सरकार/GOVERNMENT OF INDIA
CLINICAL BIOCHEMISTRY UNIT
DEPARTMENT OF BIOCHEMISTRY
V.M.M.C. & Safdarjung Hospital, New Delhi

24/03/08 13:57:10

Patient Name & CCR MRD& OPD/Ward	MD KASEEM/401
Sex	Male
Age	Year
Sample	Serum
Collection Date & Time:	91
Reviewing:	

	Result	Units	Ref. Range & (Methodology)
SODIUM	136	mEq/L	136-145 (ISE Indirect)
POTASSIUM	5.4	mEq/L	3.5-5.1 (ISE Indirect)
UREA	14	mg/dL	17-43 (Urease, UV/GLDH)
CREATININE	0.12	mg/dL	M- 0.9-1.3 , F- 0.6-1.1 (Jaffe Kinetic)
T.BIL	1.50	mg/dL	0.3-1.2 (Vanadate Oxidation)
AST(SGOT)	43	U/L	M <35, F <31 (UV Kinetic without PLP)
ALT(SGPT)	25	U/L	M < 45, F < 34 (UV Kinetic without PLP)
ALP	233	U/L	M- 53-128, F- 42-98 (PNPP-AMP Buffer)
CALCIUM	9.8	mg/dL	8.6-10.2 (Arsenazo III)
PHOSPHORUS	6.0	mg/dL	2.5-4.5 (Phosphomolybdate UV)

Remarks:

Sign. of Tech. :

Sign of Doctor:

Dr. KAVITA
PG Resident
Department of Bioch
VMMC & Safdarjung H
New Delhi

Date & Time of Report 24/03/08 13:57:10

ATIONAL
Diagnostic Centre

WAY PROFESSIONALLY

Facilities Available

Laboratory Test Report

ME :MAST.KASEEM	LAB : NO: 5724
GE: 11 MONTH	SEX :MALE
EFF. : S.J.H	DATE : 08/03/2024

VIRAL MARKER

ANTI HCV

RESULT : NON-REACTIVE

Hepatitis C is a liver disease caused by hepatitis C (HCV) virus which is found in the blood of person having disease. Hepatitis C is caused by viral infection, which is primarily as a result of blood transfusion or improper needlepuntures.
This risk of post transfusion hepatitis was estimated 8 to 12 % with 90% of post transfusion hepatitis being caused by NANB hepatitis agent. Other reports estimated that 5 to 10 % transfused individuals will develop acute NANB (Non-A Non-B) hepatitis, with 40 to 60 % progressing to become chronic NANB hepatitis and carriers. Recently the post transfusion NANB hepatitis agent, which also spreads through non-transfusion routes was definitely named hepatitis C virus. Screening for hepatitis C is important for public health management.

Hepatitis Cis spread primarily by exposure to human blood. Hepatitis C has been transmitted between Sex partners and among household members. HCV cannot be spread by food or water.

****END OF REPORT****

DR. ASHWIN BANGA (M.D)
MD. CONSULTANT MICROBIOLOGIST

LifeCell Diagnostics

SAFDARJUNG HOSPITL QWALYSEVO

Sex
Date of birth
Species

MD KASEEM 25365
MD KASEEM 25365

ABO Group Rh

Determination n° 1 — O+
Sample n° MD KASEEM 25365
Date of analysis 3/8/2024 3:15:53 PM
Validated by :
Validation date :

Microplate n°

Phenotyping Rh Kell

Determination n° 1 — D+C+c+E-e+ K-
Sample n° MD KASEEM 25365
Date of analysis 3/8/2024 3:15:53 PM
Validated by :
Validation date :

Microplate n°

A.B.S

Determination n° 1
Sample n° MD KASEEM 25365
Date of analysis 3/8/2024 3:14:40 PM
Validated by : Non validated
Validation date :

Microplate n°

Analysis	Date of analysis	Consumables	Barcode	Batch number	Expiry date
DuoLys2H	3/8/2024 3:15:53 PM	BromeLine	108050942303	800	9/8/2024
DuoLys2H	3/8/2024 3:15:53 PM	MagneLys	117080354632	705	3/31/2025
DuoLys2H	3/8/2024 3:15:53 PM	HemaLys A1	204804011472	48	4/1/2024
DuoLys2H	3/8/2024 3:15:53 PM	HemaLys B	224804011473	49	4/1/2024
ScreenLys	3/8/2024 3:14:40 PM	NanoLys	164620554351	482	5/31/2025
ScreenLys	3/8/2024 3:14:40 PM	ScreenDiluent	170350117671	635	3/31/2025
ScreenLys	3/8/2024 3:14:40 PM	HemaScreen 1	310803220006	08	3/22/202
ScreenLys	3/8/2024 3:14:40 PM	HemaScreen 2	320803220555	09	3/22/20
ScreenLys	3/8/2024 3:14:40 PM	HemaScreen 3	330803220555	09	3/22/20

Qwalys Data Software 2.0.5.6

LifeCell Diagnostics

CLINICAL BIOCHEMISTRY UNIT
DEPARTMENT OF BIOCHEMISTRY
VMMC & SAFDARJUNG HOSPITAL, NEW DELHI

24/03/08 14:26:49

Patient Name & CCR MRD# OPD/Ward	MD KASEEM/401
Sex	Male
Age	Year

Serum
91
Collection Date & Time:

Test	Result	Units	Ref. Range & (Methodology)
	0.533	mg/dL	0-0.2 (DPD-Diazo)
	1.0	mg/dL	0.2-1.0 (Calculated)

Remarks:

Sign. of Tech. :

Sign of Doctor: Dr. KAVIYA K
PG Resident
Department of Biochemistry
VMMC & Safdarjung Hospital
New Delhi 110029

Date & Time of Report 24/03/08 14:26:49

विकृति विज्ञान विभाग
DEPARTMENT OF PATHOLOGY (LAB MEDICINE)
वर्धमान महावीर मेडीकल कालेज एवं सफदरजंग अस्पताल
VARDHMAN MAHAVIR MEDICAL COLLEGE & SAFDARJANG HOSPITAL
नई दिल्ली - 110029
NEW DELHI – 110029

Lab No. : 15609

Name of Patient : KASEEM	Age : A	Gender : M	Registration No/UHID No. : 5079
OPD/Unit : PEAD	Ward/Unit: Bed No. :	Type of Primary Sample: Serum	
Date of Collection : 18/3/24		Date of Reporting : 19/3/24	

Report:

Name of Test	Value	Biological Reference Range (for normal adults)
Parathyroid Hormone	25.0	11-81 pg/ml

(Method used - Electrochemiluminescence Sandwich Immunoassay; Measuring range: 5.5 – 2300 pg/ml)

REMARKS: - Kindly Correlate Clinically

PLEASE NOTE:

1. Always interpret these values in conjunction with clinical history, serum Calcium and Phosphorus levels.
2. Elevated serum PTH along with normal serum Calcium may be seen in cases of secondary hyperparathyroidism like in case of Vitamin D deficiency.
3. The above reference range applies to healthy subjects and may vary in case of chronic kidney disease and renal failure.
4. PTH is secreted in a pulsatile manner with a circadian rhythm characterized by nocturnal rise.
5. Kindly enquire if the patient has been exposed to animal antigens, whether in the environment or as part of treatment or imaging procedures, as these patients may have circulating antihuman antibodies which may interfere with the assay reagents to produce unreliable results.

Lab. Technologist

Pathologist

fine
[illegible]gnostics

16, Vijayaraghava Road, I Lane, T.Nagar, Chennai - 600017
CIN : [illegible]
support@[illegible].co, [illegible]
Toll Free Number - [illegible]

LifeCell Diagnostics

Master MOHD KARIM HASNAYAN 12 Months Male DR. RITA MONI [illegible]	Lab ID Sample Collection Date Sample Receipt Date Reporting Date Location	[illegible] [illegible]-03-2024 15:4[illegible] [illegible]-03-2024 09:00 [illegible]-04-2024 17:3[illegible] DELHI	CRM : 5000[illegible]

Edison ES, Shaji RV, Devi SG, Moses A, Viswabandhya A, Mathews V, George B, Srivastava A, Chandy M (2008) Analysis of b globin mutations in the Indian population: presence of rare and novel mutations and region-wise heterogeneity. ClinGenet 73:331–337.
http://globin.bx.psu.edu/hbvar/menu.html

DR. JAYAKRISHNA MSc. PhD
[illegible]

DR. CHIRSTU PADHIAR
M.B.B.S [illegible]

[illegible]
This is an Electronically Authenticated Report.
Master MOHD KARIM

ATIONAL
agnostic Centre
WAY PROFESSIONALLY

Facilities Available

D. :MAST.KASEEM | LAB : NO: 5724

: 11 MONTH | SEX :MALE

F. : S.J.H | DATE : 08/03/2024

VIRAL MARKER

HBs Ag

RESULT : NON-REACTIVE

Hepatitis B is serious disease of the liver caused by hepatitis B Virus(HBV). HBV attacks And destroys the liver.

Hepetitis B is surface antigen of the hepatitis B virus. The is detected between one and four months following exposure to the Hepatitis B virus. The presence of HBsAg indicates acute phase of infection or chronic infection.some patients may develop chronic liver disease and be HBsAg positive but have no history of acute hepatitis.HBsAg is also Positive in 'chronic carreiers'. Some people who have hepatitis B have no symptoms and may not know they are infected, others who are infected with HBV never fully recover and carry virus in their blood for rest of their lives. These people are known as carrier.

*****END OF REPORT*****

DR. ASHWIN BANGA (M.D)
M.D. CONSULTANT MICROBIOLOGIST
REG. NO.4897

Methodology:

Sanger sequencing based DNA analysis was performed to identify the mutations in the coding regions and reported mutations in the non- coding regions of β Globin gene (HBB). DNA was isolated from the sample received using a commercial kit that works on the silica- membrane- based DNA purification. Mutation surveyor software or software V5.1.1 is used for identifying the mutations and the identified sequence alterations are reported in accordance with the Human Genome Variants Society (HGVS) nomenclature and annotated using one or more of the following database/s- ClinVar, OMIM, HGMD and SwissVar.

Understanding the results:

Mutation Type	Possible Genotype *	Clinical significance#
β Type	$\beta^+\beta^+$ $\beta^+\beta^0$ $\beta^+\beta$ $\beta\beta$	Thalassemia Intermedia/Major Thalassemia Major Thalassemia Minor Normal
β^+ Type	$\beta^+\beta^0$ $\beta^+\beta^+$ $\beta^0\beta$ $\beta\beta$	Thalassemia Major Thalassemia Major Thalassemia Minor Normal

*β type- Normal functional haemoglobin, β^+type Mutation: Reduced ability to produce β Globin protein with reduced functional capacity for hemoglobin, β^0type mutation: No β Globin will be produced and dysfunctional haemoglobin.

#Thalassemia minor: Patient with one normal copy of the β-Globin gene and may show mild to no symptoms for Thalassemia, Thalassemia major: Patient with both abnormal copies of the β Globin gene and may show acute symptoms for Thalassemia, Thalassemia intermedia- Patient with both abnormal copies of the β Globin gene and may show acute symptoms for Thalassemia at adult stage.

Disclaimer:

1) The results specifically pertains to the examined sample and may not accurately represent the fetal chromosome constitution in instances of confined placental mosaicism or when the sample is contaminated with maternal cells.
2) Despite taking all necessary precautions during DNA-based tests, the technical error rate for all types of DNA analysis is approximately 2%. Therefore, it is crucial to interpret all results within this context before taking any action based on these results.

CAP ACCREDITED

LifeCell Diagnostics

Lab ID		
Sample Collection Date		
Sample Receipt Date		
Reporting Date		
Location	DELHI	

The report is generated within a specific timeframe known as the turnaround time (TAT) once received the sample received at the lab.
However, the actual TAT may differ based on the complexity of the requested test(s) and information provided along with the sample.
LifeCell is not responsible for delays due to incomplete information and or any technical requirements during sample handling
and/during reporting.

4) In certain rare cases, genetic tests may not provide accurate results, for example: when the quality of the sample given to Lifecell is not
optimal. If a test performed by Lifecell fails due to unforeseen or unknown reasons beyond our control, Lifecell cannot be held responsible
for any incomplete, potentially misleading, or incorrect results that were not foreseeable beforehand.

5) Lifecell Pvt. Ltd has validated the test and determined its performance characteristics in accordance with the CAP/ACMG and NABL
guidelines. All investigations have their limitations which are imposed by the limits of sensitivity & specificity of individual assay
procedures as well as the quality of the specimen received by the laboratory.

6) Clinical interpretation of given test result should be evaluated within the context of the patients medical history and other diagnostic
laboratory test results.

7) The present report comprises genetic analysis of the sample provided. It is important to note that this report cannot be accurately interpreted
without information regarding clinical features and other laboratory reports, investigations. The information contained herein should not be
considered a substitute for professional medical advice or diagnosis. It is highly recommended to consult with a qualified healthcare
professional or medical specialist for a comprehensive evaluation and interpretation of your specific medical condition.

8) Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations caused
by the presence of pseudogenes, repetitive, or homologous regions.

9) The mismatch between HPLC report/Capillary report and HBB gene analysis can occur due to several reasons such as the presence of rare
or novel mutations, technical errors in the laboratory, or variations in the methodology used for analysis.

10) This test does not distinguish between the alleles present in cis and trans forms. Classification of haemoglobin disorders by genetic data
alone may result in incomplete conclusions which may significantly impact overall disorder classification and expected phenotype. The test
results does not mean that the risk of carrying or developing Thalassemia/sickle cell anemia is not present and or caused by other known or
unknown variations in the genome.

11) Correlation with haemoglobin electrophoresis, red blood cell indices and clinical/family history is required. This test will detect greater than
90% of variants in HBB gene excluding large insertions and deletions, which may be a rare phenomenon.

12) Any change in primer binding site can interfere with the results and or allele dropout cannot be ruled out using this experiment. Benign
variants will be provided upon request. The current assay doesn't detect the delta beta inversion deletion mutation or any other mutation on
Delta, Gamma and Alpha gene.

13) This document is for clinical interpretation and not for medico-legal purpose.

14) As per the PRE-NATAL DIAGNOSTIC TECHNIQUES (REGULATIONS & PREVENTION OF MISUSE) AMENDMENT ACT 2002,
sex determination shall not be done for all prenatal samples.

References:

- Colah R, Gorakshakar A, Nadkarni A, Phanasgaonkar S, Surve R, Sawant P, Mohanty D, Ghosh K (2009) Regional heterogeneity of b-thalassemia mutations in the multi ethnic Indian population. Blood Cells Mol Dis 42:241–246.

This is an Electronically Authenticated Report.

CAP ACCREDITED