
PURSUIT

Old OT genetics lab

GENETICS UNIT DEPARTMENT OF PEDIATRICS, AIIMS, NEW DELHI
Biochemical Genetics Lab Room No. 2/3 Old O.T. Block Ph: 011-26593558, 26594585

Requisition & Reporting Form for Blood Ammonia

Name: Ruah Raj Age/Sex: 14 Date: 22/2/23

Hospital Regn./UHID No: 10605070.1 Unit: II Ward/OPD/Clinic: ~~[illegible]~~ OPD

Patient's Address and contact no. Delhi, [illegible]

Clinical Summary with Important Investigations: Leopard syn I, neonatal hyperammonemia

Diagnosis:

Referring Doctor: Dr Rohit

Name ---------- Contact No. ---------- Signature Rohit.

Conditions Required:

1. OPD or Ward Patients / Patient's Attendant should be sent to lab (room no. 2, Genetic Unit, ground floor) with filled - in form prior to taking the sample.

2. Sampling for OPD Patients is done in Room No. 109 first floor Genetic Unit 10 AM – 12:30 PM

3. Patient should be **Fasting** (Can take Water) :

- Day 0 – 3 Months : 2 hrs
- 3 Months – 3 Years : 4 - 6 hrs
- 3 Years - Adult : Overnight

FOR LAB USE ONLY

Report:

Lab no.: AI-230/23

Date of Reporting: 22/2/23

Blood Ammonia : 86 µg/ dl

Normal Range (Age wise):

Day 0:	≤240 µg/ dl
Day 1 to 1 week:	≤228 µg/ dl
1 Week and Above:	30 – 86 µg/ dl

Comments: Ammonia Level Normal

Signature (Test done by)

Signature (Checked By)

Signature (In – Charge Genetic Un

Incomplete form will not be accepted. Please specify the HIV/Hepatitis status of the patient, if any.
Challan to be collected from room no.109, Genetics unit, AIIMS (Monday to Friday 10:00am to 12:30pm)

अ० भा० आ० सं० अस्पताल/A.I.I.M.S. HOSPITAL
बहिरंग रोगी विभाग /Out Patient Department

OPR-4

आयु
Age

पता/Address

निदान/Diagnosis

उपचार/Treatment

दिनांक/Date

8 वर्ष (-0.7?)

c/o GDD

२९ (-0.23)

Ht: 74cm

HC: 42cm (-4.69)

NCM

Birth: No adverse perinatal events
LSCS, not aware whether the
child cried.
Child was discharged on day 7
Was beside the mother during
the whole duration. No seizures

Developmental:
Gross: no neck holding
sits c̄ support

Fine: palmar grasp

Language: Monosyllables

Social: recognises parents
social smile

· Gibbus (+)
Mongolian spots (+) · (Rt) foot - Postaxial polydactyly
(Lt) Inverted nipple ·
(Rt) foot - Overriding toes · Broad - great toe ·

DD ++ Central hypotonia
No organomegaly
Wrist widening (+) ·

MRI - B
21. June. 2023
· LHF sequels ·
CC dysgenesis

NH3 - 32
Na - 136
K - 4.9
Cl - 99
Lactate - 31.9 (4.5 - 19)
G. ketone - Negative

Database :- 1 year / male / NCM ·
GDD c̄ macrocephaly c̄
Subtle dysmorphism c̄
Central hypotonia c̄ Syn -
(Lt) Post-axial polydactyly

· Low - B - Physiotherapy ·
· Peads Neuro CR on call → Room 5/4/32
(for DQ Assessment)
PRC ·
↓
· Genetic OPD ·
· ENT OPD ·
· forms ·
(5th July / 2023)

Plan
· MRI for discussion ✓
· Thyroid fn test ✓ ·
· Hearing evaluation
· Database search ·
· USG abdomen
Echo
· Vit D / PTH / Ca / P / ALP ·

CNC Cardiology consultation
↓
Echo ·

Dr. [signature]

Ex-1817

अ० भा० आ० सं० अस्पताल/A.I.I.M.S. HOSPITAL
बहिरंग रोगी विभाग /Out Patient Department

OPR-6

Unit
Dept
Name
Address

120

GC 221/22 UNB
F.M.M

Diagnosis

Date	Treatment
26	8516
	FUC last seen on 24.06.22
(DOB = 17.07.2021)	Database
	1yr male child / NCM / Predominantly motor delay (DQ ~ 50%)
	c̄ megalencephaly c̄ subtle dysmorphism
	c̄ central hypotonia c̄ (R) foot post axial poly syndactyly.
24.06.22	(N) cognition (N) fine motor
· Vit D →	115.5 ng/ml → No Hx of vomit's / altered sensorium feeding problem, polyuria / polydypsia
iPTH →	18 pg/ml
Urea/Cr. =	32/0.2
Ca/PO4 =	9.8/6.0
	GRC MRI discussion (30.06.22)
	· Thalami & brainstem → darker than (N)
	· Gyration abnormalities < (L) frontal, (R) Parietal
2D Echo → (N)	· Prominent cerebellum
USG Abd → (N)	· Crowding of post. Fossa
	(N) - corpus callosum

O/E

Vitals stable

RE open Rim x Rem
not bulging

R/L pupil reacting to light

CNS

Motor - Bulk (N)
Tone (N) in R/L Limb
DTR brisk (2+)
Power 3/5

Sensory: reacting to touch, pain

No dysdiadochokinesia

F/A
CVS } NAD
RS

NCCT - Ventricles slightly dilated
Mild cerebral atrophy

Adv

~~PMR~~ MRI / fundus

opthal evaluation - RPC } PR C
hearing evaluation - ENT } (DROM)

R/w repeat of MRI

- VBG
- Serum electrolytes
- Venous lactate
- Urine ketone
- Serum Ammonia

10/ 6yr

MRI Discussed: No holoprosencephaly
Brainstem (N)
prominent cerebellum
No gyral asymmetry
Corpus callosum - thinned

LH, FSH, TSH → pending

TSH → 1.0253 mIU/ml
(↓) LH → 0.11 mIU/ml
FSH → 0.34 mIU/ml

Adv.

[CBC] → hematology
[LH, FSH, TSH] → Central lab

c/D/w Dr Mounika ma'am.

- Some features matching c̄ PTCH1.
- Segregation analysis to be planned in followup.
- To repeat (ABG) - [MCB SC] - Dr Rohit (SR genetics).

- To f/u in genetics clinic after 3 months
Appt Room 30

- physiotherapy
- occupational therapy] (PRC).
L Ped Neuro

Soumalya
SR
26/June/23
26/June/23
Genetic cli.

SHREE SARAN JANCH GHAR
HOSPITAL ROAD NEAR PATHTANKI, HAJIPUR
One of the Largest Referral Bolld Testing Lab in Hajipur
Mob. : 6207096610
9608413639

REPORT

PATIENT NAME	:RUDRA KUMAR	REF.NO.	:M-24915
AGE	:10 MONTH	DATE	:Mon 23-05-2022
SEX	:Male	REF.BY DR.	:ROSHAN (M.D)

TEST NAME	NORMAL VALUE	FINDING
TOTAL LEUCOCYTE COUNT	(4000-11000/cumm)	8.200 /cumm
DIFFERENTIAL LEUCOCYTE COUNT		
Neutrophils	40-75 %	44 %
Lymphocytes	20-50 %	46 %
Eosinophils	01-06 %	09 %
Monocytes	02-08 %	01 %
Basophils	00-01 %	00 %
HAEMOGLOBIN	(M-15±2.5, F-14±2.5)	9.9 gm/dl

PROCESSED AT :
THYROCARE
CHOUHATTA, OPPOSITE
DARBHANGA HOUSE,
ASHOK RAJPATH ROAD, PATNA -
800 004

Thyrocare
The Trust. The Truth.

Corporate office : Thyrocare Technologies Limited, ✉ B-37/1, TTC MIDC, Turbhe, Navi Mumbai - 400 703
✆ 022 - 3090 0000 / 6712 1400 ✆ 9870666333 ✉ wellness@thyrocare.com ⊕ www.thyrocare.com

REPORT

NAME : RUDRA KUMAR (10M/M)
REF. BY : DR ROSHAN MD
TEST ASKED : TSH

SAMPLE COLLECTED AT :
(8441021334),SHARAN TAHCH GHAR PARE TAHVI
NEAR UDPAD THANA HAJIPUR VAISHALI BIHAR,
844101

TEST NAME	TECHNOLOGY	VALUE	UNITS	REFERENCE RANGE
THYROID STIMULATING HORMONE (TSH)	C.M.I.A	0.68	µIU/ml	0.35 - 4.94

Please correlate with clinical conditions.

Method :

TSH - Fully Automated Chemi Luminescent Microparticle Immunoassay

Please note above printed references are applicable only for ADULT

Refer below said table for < 18 years reference range

TEST	1 - 3 D	4 - 30 D	31 - 60 D	61 D - 12 M	1 - 5 Y	6 - 10 Y	11 - 14 Y	15 - 18 Y
TSH	0.1-9.2	0.2-8.5	0.2-7.8	0.30-5.9	0.4-4.8	0.5-4.7	0.5-4.6	0.5-4.5
T3	41.7-272.1	48.2-272.1	54.7-272.1	76.8-272.1	89.2-246.7	87.2-218.1	86.6-199.8	85.3-188.8
T4	4.9-15.8	5-15.3	5.2-14.8	5.7-13.3	5.7-11.7	5.4-10.7	5.2-10	5.1-9.6
FT3	1.5-5.3	1.6-5.2	1.6-5.1	1.8-4.8	2-4.5	2.1-4.4	2.3-4.4	2.3-4.3
FT4	0.84-2.08	0.85-1.98	0.85-1.89	0.89-1.62	0.89-1.48	0.85-1.46	0.84-1.45	0.84-1.45

-- End of report --

Sample Collected on (SCT) : 24 May 2022 16:20
Sample Received on (SRT) : 24 May 2022 21:42
Report Released on (RRT) : 25 May 2022 01:32
Sample Type : SERUM
Labcode : 2405107903/BR121
Barcode : Y9815636

Dr.Caesar Sengupta MD(Micro) Dr Ritika Gupta MD(Path)

636/33

DEPARTMENT OF OTORHINOLARYNGOLOGY & HEAD-NECK SURGERY

ALL INDIA INSTITUTE OF MEDICAL SCIENCES, NEW DELHI

Requisition form for audiometric evaluation

Name: Rudra Arj

Age/Sex: 4yr/M

UHID: 106050701

Otoscopy

Weber

Rinne

ABG

Diagnosis

Requested by

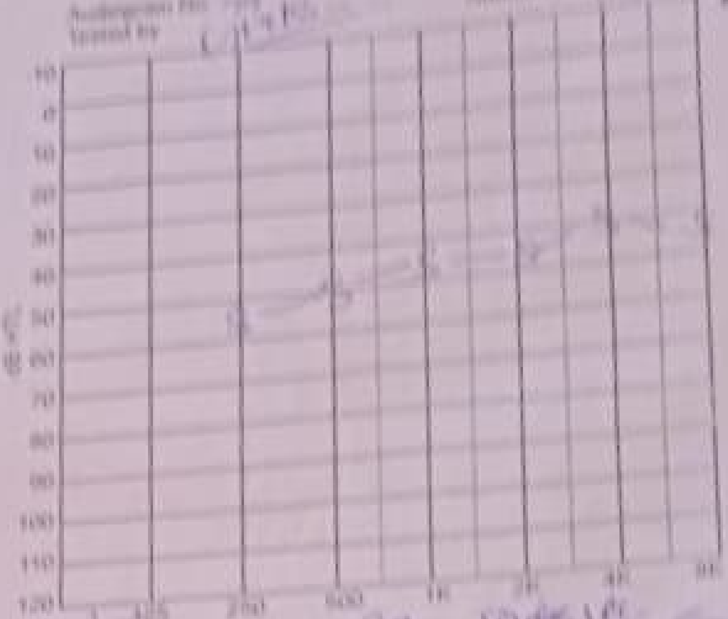
Date

Date: 4/1/23

Audiometer

Audiogram No.

Tested by



Reliability / Remarks: SRT: 50 dB HL; Unreliable

INVESTIGATION REQUIRED

- Pure Tone Audiometry ☑
- Speech Audiometry ☐
- Free Field Audiometry ☑
- Special Tests
  - Difference Limen (DL) ☐
  - SISI ☐
  - Tone Decay ☐
  - ABLB ☐
- Tinnitus Matching/Masking ☐
- BERA
- Hearing Aid Trial ☐
- Real Ear Measurement ☐
- Others ☐

AUDIOMETRY KEY

	R	L
A.C. Unmasked	O	X
No response	O	X
A.C. Masked	△	☐
No response	△	☐
B.C. (Mastoid) Unmasked	<	>
No response	<	>
B.C. (Mastoid) Masked	[	]
No response	[	]

B.C. (Forehead) Unmasked V

Free Field S

Date

Audiometer

Audiogram No.

Tested by

Reliability / Remarks :

Immittance		RT	LT
Type			
Compliance			
Pressure			
Reflex	IPSI		
	CONTRA		

दिनांक - Date | उपचार - Treatment

(E8) - Dilate BE

Fn (| CDR 0.3:1,
NRR healthy,
Fr sharp

Adv.

(E8) Refraction ⊥ E/O Atropine

25/6/22

−0.50 / +1.00 | ±0.00 / +1.50

F/U c̄ reports/SOS

−1.00 −1.50 × 180

−0.50 −1.50 × 180

अखिल भारतीय आयुर्विज्ञान संस्थान, नई दिल्ली
ALL INDIA INSTITUTE OF MEDICAL SCIENCES, NEW DELHI
Department of Lab Medicine (Emergency and Ward)

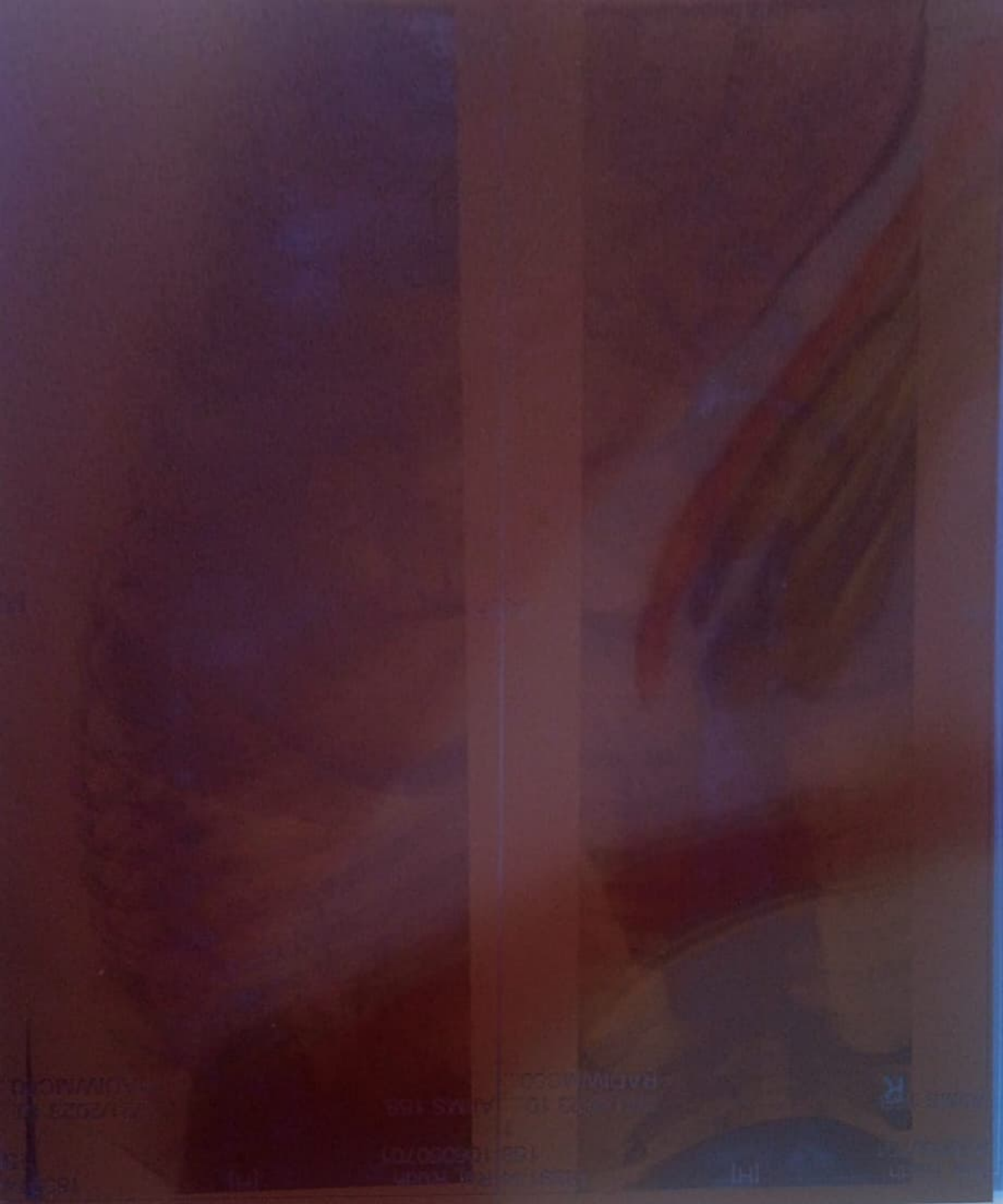
UHID:	[illegible]	Sex :	Male
Patient Name :	**Mr RUDH RAJ**	Sample Received Date :	23/03/2023 04:59 PM
Age :	1 year 7 months 1 day	Department :	Paediatrics
Unit Name :	Unit-II	Unit Incharge :	
Lab Name:	Lab Medicine	Lab Sub Centre:	
Reg Date :	24/06/2023 08:54 AM	Sample Collection Date:	23/03/2023 12:46 PM
Report Generated Date:	23/03/2023 01:33 pm	Dept / IRCH No:	2023030003955
Recommended By :	Dr. Dilip SR Pasi	Lab Reference No:	575

Sample Details : WC-220223545

Report

Test Name	Result	Comment	Normal Range
Urea	35 **mg/dL**		• 15 - 46 mg/dL
Creatinine	0.3 **mg/dL**		• 0.66 - 1.25 mg/dL
Uric Acid	2.8 **mg/dL**		• 3.5 - 8.5 mg/dL
Calcium	9.2 **mg/dL**		• 8.4 - 10.2 mg/dL
Phosphorus	5.6 **mg/dL**		• 2.5 - 4.5 mg/dL
Sodium	145 **mmo/L**		• 137 - 145 mmol/L
Potassium	5.4 **mmo/L**		• 3.5 - 5.1 mmol/L
Chloride	116 **mmo/L**		• 98 - 107 mmol/L
Bilirubin (T)	0.2 **mg/dL**		• 0 - 1.4 mg/dL • 0.2 - 1.3 mg/dL • 1 - 10.5
Bilirubin (D)	0.10 **mg/dL**		• 0 - 0.6 mg/dL • 0 - 0.3 • 0 - 0.6
Bilirubin (I)	0.1 **mg/dL**		• 0.6 - 10.5 mg/dL • 0 - 1.1 • 0.6 - 10.5
ALT	23 **U/L**		• < 50 U/L • < 35 U/L
AST	57 **U/L**		• 17 - 59 U/L
ALP	313 **U/L**		• 38 - 126 U/L
Total protein	7.1 **gm/dl**		• 6.3 - 8.2 gm/dl
Albumin	4.3 **gm/dl**		• 3.5 - 5 gm/dl
Globulin	2.8 **g/dL**		• 3 - 3.7 gm/dl
A/G ratio	1.54		• 0.8 - 2

Over All Comment : Kindly Correlate Results Clinically.



MedGenome Labs Ltd.
3rd Floor, Narayana Nethralaya Building, Narayana Health City,
#258/A, Bommasandra, Hosur Road, Bangalore - 560 099, India.
Tel : +91 (0)80 67154989 / 980, Web: www.medgenome.com

MEDGENOME

Sakthivel Muragan, Ph.D
Vice President -
Lab Operations

Balaji Rajashekar, Ph.D
Director - Clinical Bioinformatics

Dr. Mallikarjun Patil DNB(Medical Genetics), MD(Pediatrics),DCh
Consultant- Senior Clinical Geneticist

APPENDIX

TEST METHODOLOGY

Targeted gene sequencing: Selective capture and sequencing of the protein coding regions of the genome/genes is performed. Mutations identified in the exonic regions are generally actionable compared to variations that occur in non-coding regions. Targeted sequencing represents a cost-effective approach to detect variants present in multiple/large genes in an individual.

DNA extracted from blood was used to perform targeted gene capture using a custom capture kit. The libraries were sequenced to mean >80-100X coverage on Illumina sequencing platform. We follow the GATK best practices framework for identification of variants in the sample using Sentieon (v201808.07) [3]. The sequences obtained are aligned to human reference genome (GRCh38.p13) using Sentieon aligner [3, 4] and analyzed using Sentieon for removing duplicates, recalibration and re-alignment of indels [3]. Sentieon haplotype caller has been used to identify variants which are relevant to the clinical indication. Gene annotation of the variants is performed using VEP program [5] against the Ensembl release 99 human gene model [6]. In addition to SNVs and small Indels, copy number variants (CNVs) are detected from targeted sequence data using the ExomeDepth (v1.1.10) method [7]. This algorithm detects rare CNVs based on comparison of the read-depths of the test data with the matched aggregate reference dataset.

Clinically relevant mutations were annotated using published variants in literature and a set of diseases databases - ClinVar, OMIM (updated on 11th May 2020), GWAS, HGMD (v2020.2) and SwissVar [8-12]. Common variants are filtered based on allele frequency in 1000Genome Phase 3, gnomAD (v3.0), EVS, dbSNP (v151), 1000 Japanese Genome and our internal Indian population database [13-16]. Non-synonymous variants effect is calculated using multiple algorithms such as PolyPhen-2, SIFT, MutationTaster2 and LRT. Only non-synonymous and splice site variants found in the clinical exome panel consisting of 6120 genes were used for clinical interpretation. Silent variations that do not result in any change in amino acid in the coding region are not reported.

Average sequencing depth (x)	Average on-target sequencing depth (x)	Percentage target base pairs covered		
		0x	≥5x	≥20x
318	119.49	1.73	96.87	93.46

Total data generated (Gb)	11.67
Total reads aligned (%)	99.99
Reads that passed alignment (%)	92.60
Data ≥Q30 (%)	97.03

classification of the variations is done based on American College of Medical Genetics as described below [1].

ant	A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).

CA
ACCREDIT

डॉ० राजेन्द्र प्रसाद नेत्र विज्ञान केन्द्र / Dr. Rajendra Prasad Centre for Ophthalmic Scien

A.I.I.M.S., NEW DELHI-110029

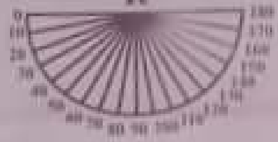
REFRACTION SLIP O.P.D. No. [illegible]

[illegible]

नाम/Name Radh Roy

Dx & Vn

R



L

	Sph.	Cyl.	Axis	Vision	Sph.	Cyl.	Axis	Vision
दूर Distance	-1.00	-1.50	180		-0.50	-1.50	180	
निकट Near	—	—			—			

निर्देश/Remarks Constant use

तिथि/Date 25/4/2[illegible]

नेत्र विशेषज्ञ/EYE SURGEO

KILKARI TRUST
Regd. No.464

KILKARI TRUST

You Think, You Care, You give.

Mob.: 8588981217

Ref. No.: .....

Date : 6-3-2023

सेवा मे;
संस्थापक महोदया,
किलकारी ट्रस्ट
नई दिल्ली,

महोदया,

मेरा बच्चा रुद्र राज जो की 11 महीना का है। वह मानसीक रूप से बीमार है। वह सही से खड़ा भी नहीं हो पाता है, उसके रीढ़ में तीवक्रता है। उसका सर बहुत बड़ा है। वह और बच्चा से बिलकुल अलग है। इसका इलाज AIIMS में चल रहा है, मैं आर्थीक रूप से कमजोर हुँ। मैं अब सक्षम नहीं हुं इलाज करवाने मे। अगर आपके ओर से मदत मील जाता। जीवन भर रुद्र गुलाम रहेगा।

कृपया आर्थीक रूप से मदत करे।

प्रार्थी पिता
संजीव कुमार

KILKARI TRUST Reg. No. 464 DELHI