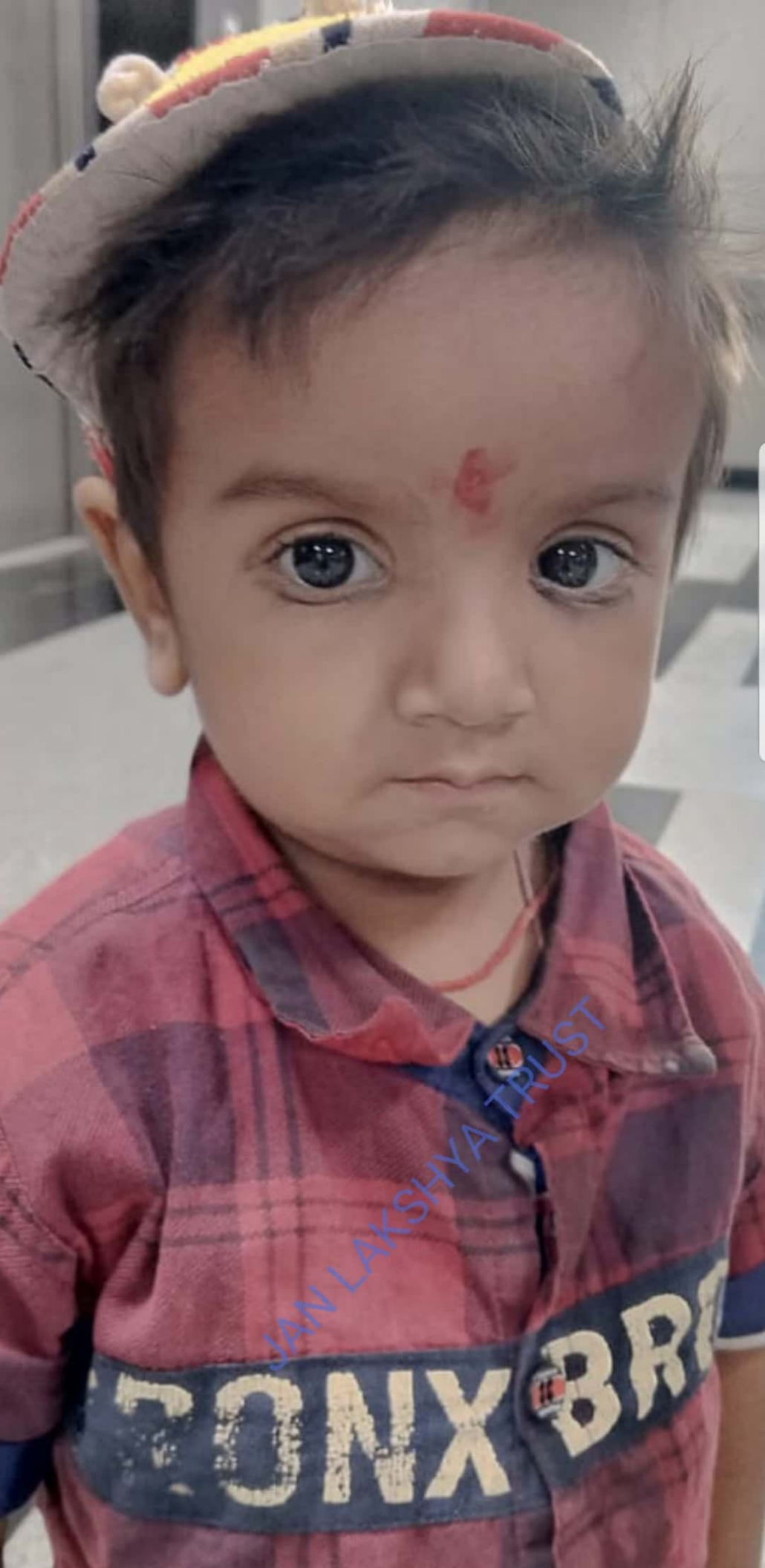
JAIN LAKSHYA TRUST

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

SMOKING IS PROHIBITED IN HOSPITAL PREMISES

बाल चिकित्सा विभाग

UHID 106656338
ABHA: 0
0
Dept No.: 20230033011608

कमरा / Room C-217 Unit I
Paediatric
सोम गुरु
Mon Thu (सोम गुरु) 04/03/2024
Queue: F49

OPR-6

O.P.D. Regn. No.

आयु Age

पता/Address

कुलदीप वेर्मा
KULDEEP VERMA
S/O NARYAN SINGH VERMA

Follow Up Patient

निदान/Diagnosis

CKD-5

दिनांक/Date	उपचार/Treatment
49	P-I (-4.182)

c/o CKD-5 / hypodysplastic kidneys ?? nephronophthisis
on followup
polydipsia/polyuria.

BP = 90/62
Ht = 79 cm (-4.182)

3/3/24

7.93 > 150/80 < 2.18 ; N 32 660

(2 from 1.3) ↑

Vit D - 34
Ur/Cr - 68/2.4 (eGFR ~12.6)
Alb - ALP - 98
Alb - 3.6
PTH - 826.7 → (↓ from 1113)

Adv Dr Adil

1) Syp Vitcofol 7.5ml OD
2) Syp. Shelcal 5ml TDS empty stomach
3) C Calcitriol 0.25mcg twice weekly
4) Syp. Nodosis 9.5ml TDS
5) Inj Darbepoetin 20mcg s.c every 2 weeks
6) F/u in 802/8th floor MCB to meet Dr Adil @ 2:30pm

3pm
- Counselled parents at length.
- Keen to get estimate made for transplant.

Dr Saiteja / Dr. Pavni.

Dr. P SAITEJA
Senior Re...
AIIMS, New Delhi

Please do the estimate

Pass.

9

Syp. Vitcofol 7.5 ml OD.
Syp. Shelcal 5ml TDS.
Nodosis 2.5 ml TDS
Inj. Darba 25 mcg s.c. q 2 wkly

R/v 1 week.

बाल चिकित्सा विभाग

UHID: 106656338
Dept No.: 20230030011606

कमरा / Room
C-212
Unit
Paediatric

कुलदीप वेर्मा
KULDEEP VERMA
SONARYAN SINGH VERMA

Follow Up Patient

Mon,Thu
11/01/2024
Queue: F29

Ht = 79.4 cm

28/12/23
Hb = 6.30 — pallor not clinically present.
WBC = 15.48
Plt = 240
Urea/creat = 126/2.7 ←
UA = 9.5
Ca/phos = 9.1/3.9
Na/K+ = 138/4.6
Chloride = 102
ALT/AST = 20/34
ALP = 548 ↑
T.P = 7.4
Albu = 4.4 (N)
Vit. D3 = 51.50 (N)
Iron = 141 ↑
Transferrin = 215
Ferritin = 621.0 ↑
TIBC = 278
Vit B12 = 1699.0
Sr. folate = 720.
PTH = 1113.60

? CaSR hypersensitivity

K/c/o CKD-5 (CAKUT)

? ~~h/o PUV — fulguration done 12 months back~~

→ USG-KUB (15/11/23): B/L hydronephrosis
— hypodysplasia
— nephronophthisis

— currently c/o Anemia, mineral bone disease
— h/o polydipsia, awakening at night following micturition.

eGFR ... ml/min/1.73

→ Advice:
1. Syrup. Vitcofol 7.5 ml OD (दिन में एक बार)
2. Syrup Shelcal 5 ml TDS (सुबह-दोपहर-रात)
3. 1-α Vitamin D 0.25 ug twice weekly (हफ्ते में 2 बार)
4. Nodosis 2.5 ml TDS (सुबह-दोपहर-रात)
5. Inj Darbapoietin 25 mcg s.c. q 2 weekly
- Syp PCM (125 mg/5 ml) 4 ml PO. SOS
6. Review in MCB-4th floor Room 418:- 4th floor. Dietician Briefing.
7. Review in 6 wks.

Before coming
CBC
LFT, KFT
Vit D, iPTH

Dr. ALAPAN DAS
Junior Resident
Department of Paediatrics
AIIMS New Delhi

Dr. Alapan

[illegible]
[illegible]

Father is the likely donor

To get mother
father } blood group

BP - 130/84 mmHg
Wt 53.8 kg
Ht 172 cm

Recipient - Blood group

Based on blood group - Donor
CBC
RFT
LFT
FBS
HbA1c, TFT, lipid profile
Viral markers HCV
HIV
HBsAg
Urine R/M
24 hr urine protein, creatinine

Recipient - TFT, Viral markers
HbA1c, FBS, lipid profile.

R/w after Reports

Dr P SAITEJA
Senior Resident
...
AIIMS, New ...

अखिल भारतीय आयुर्विज्ञान संस्थान, नई दिल्ली

All India Institute Of Medical Sciences, New Delhi

UHID:	100656338	Sex :	Male
Patient Name :	Mr KULDEEP VERMA	Sample Received Date :	28-Dec-2023 20:26 PM
Age :	2Y 8m	Department :	Paediatrics
Lab Name:	Dept of Laboratory Medicine	Lab Sub Centre:	Smart Lab New OPD Block
Reg Date :	28-Dec-2023 16:33 PM	Sample Collection Date:	28-Dec-2023 13:51 PM
Recommended By:		Lab Reference No:	231336078S

Sample Details : LC2812231775 Sample Type : Serum

Report

BIOCHEMISTRY

Test Name (Methodology)	Result	UOM	Reference

Dr. Sudip Kumar Datta (Biochemistry & Immunoassay) | Dr. Tushar Sehgal (Hematology & Coagulation) | Dr. Suneeta Meena (Serology) | Dr. Devendra Kr. Verma (Laboratory Medicine) 29-Dec-2023 11:22

IMMUNOASSAY

Test Name (Methodology)	Result	UOM	Reference
Intact PTH (ECLIA)	1113.00	pg/mL	15 - 65

-----End of Report----

Dr. Sudip Kumar Datta (Biochemistry & Immunoassay) | Dr. Tushar Sehgal (Hematology & Coagulation) | Dr. Suneeta Meena (Serology) | Dr. Devendra Kr. Verma Laboratory Medicine) 29-Dec-2023 11:22

UHID: 106656338
Dept No.: 20230030011808

C-212

Paediatric

शिशुरोग

कुलदीप वेर्मा
KULDEEP VERMA

Mon Thu (सोम गुरु)
09/10/2023

Queue: F66

Follow Up Patient

F/u/c CKD 4-5/anemia, RMD
No h/o reduced urine output

7 kg

BP – 80/50 mmHg
Ht – 79 cm

- 41 E ... 4 ...
- { CBC, KFT, 25 OH vitamin D, PTH, VBG/ICE } MCB 4C
  - 4 pm 4C ward
  - Langa
- Syp Calcifol 7.5 ml OD
- Syp Shelcal 5 ml BD
- Vit D3 drops 2 ml P/O OD
- Syp. Nodosis 2.5 – 2.5 – 2.5 ml
- Inj Darbepoetin 25 μg S/C 1-2 weekly.

F/u in 6 weeks.

Currently accepting well

↓

NO c/o loose stools

Vitals stable

Adv-

- Syp. Zinc (5ml/20mg) 5ml PO OD X 14days.
- ORS ad lib.
- Tab. folic acid (5mg) 1tab PO stat
  f/b 1mg/day X 4days.
- Syp. M.V. 2.5 ml PO BD
- Danger signs explained
- Review SOS in casualty
- F/u in ORD for [illegible]
- F

Pandya

Sd/-

Syp. PCM (125/5) 4ml SOS. (T>100.4F)

Syp. Ondem (2mg/5ml) 5ml @6hrly (2days)

DS/ ward
VBG at 4pm
↓
to decide on Syp Nodoeis VDY

Adv . ○ दोनों

→ Syp Vitcofol 5ml od

Syp Vit D_3 2ml od ○ दोनों

Syp Shelcal 5ml - 5ml - 5ml सुबह दोपहर शाम

Inj Daelrpoietin 25 ug s/c 1/2 weeks.

. Diet as advised - 215 .

Syp Nodoeis -

. Review after 1 month with CBC, RFT, LFT,

Diet:-

- Poor oral intake ~ ½ bowl at once.
- 8-9 times breastfeeding

Adv - ↑ portion intake.
Diet plan given & counselled.
k/c in two.

अ० भा० आ० सं० अस्पताल/A.I.I.M.S. HOSPITAL

बहिरंग रोगी विभाग /Out Patient Department

शरीरमाद्यं खलु धर्मसाधनम्

[...]ITAL PREMISES

OPR-6

कक्ष/ Room

UHID: 106656338
Dept No: 20230030011608

14
Unit-I
Paediatric
Queue No: N4
01/05/2023

कुलदीप वेर्मा
KULDEEP VERMA
2Y M 16D / M(पुरुष)
SONARYAN SINGH VERMA
Add: VILL HINOTI POST LAKHONI TASHIL JIRAPUR HANOTI RAJGARH, MADHYA PRADESH Pin 0 INDIA

सोम,गुरु
Mon,Thu (सोम,गुरु)

New Patient General ₹ 0 Reporting: 8:00 AM-9:00 AM

एकक/Unit
विभाग/Dept.
नाम/Name

O.P.D. Regn. No. Rea 8

LGH010523403 106656338
LC0105232024 106656338
KULDEEPVERMA

106656338

निदान/Diagnosis: CKD / Marked ↑ PUV r/o

दिनांक/Date	उपचार/Treatment
19 SDS: Wt/Ht - 3.03 Wt/age - 5.3 Ht/age - 6.5 8.4 / 17,700 / 27/60 < 2.93 Urea/Creat 94/2.6 UA 10.8 Ca/P: 9.5/6.4 OT/PT: 37/22 TP/Alb: 7/4.7 Vit D3: 66 PTH GFR: 17 (DTPA (26/4/23) Iron replete! SAT 135. Transf 181. Ferritin 595. TIBC 744. Vitamin B12 >2000	6.4 kg – Recurrent episodes of fever c/o poor urine stream since birth HR = 68; BP – crying; Capillary refill; Do not have small cuff Referred from Paeds Sx. h/o polyuria / polydipsia CKD unclear etiology. Suspected PUV on MCU. However, USG r/o small kidneys, no hydronephrosis or hydroureter. – Marked failure to thrive + anemia + MBD. Has undergone MCU twice and cystoscopy once. 1st MCU: c/o prominent UB on c narrow urethra. 2nd MCU: and abn urethra on 2nd MCU. Cystoscopy r/o PUV. eGFR: – Currently on a catheter o/e – Frontal + Parietal bossing – Pallor ++. – SDS < Wt, Ht eGFR ~10.8 ml/min/1.73 m² ↑ Adv: – Stop Metholamine. – USG KUB here or Maninder: r/o Nephronophthisis, ADTKD PTH. Magnesium. Uric Acid. Urea. Creatinine. HbA1c. ★ (27) today

CLEAN AND GREEN AIIMS / एम्स का यही संकल्प, स्वच्छता से काया कल्प
अंगदान-जीवन का बहुमूल्य उपहार/ORGAN DONATION - A GIFT OF LIFE
O.R.B.O., AIIMS, 26588360, 26593444, www.orbo.org Helpline - 1060 (24 hrs service)

मेरा अस्पताल My Hospital meraaspatal.nhp.gov.in

अखिल भारतीय आयुर्विज्ञान संस्थान, नई दिल्ली
All India Institute Of Medical Sciences, New Delhi

UHID:	106656338	**Sex :**	Male
Patient Name :	**Mr KULDEEP VERMA**	**Sample Received Date :**	04-May-2023 18:17 PM
Age :	2Y	**Department :**	Paediatrics
Lab Name:	Dept of Laboratory Medicine	**Lab Sub Centre:**	Smart Lab New OPD Block
Reg Date :	04-May-2023 17:16 PM	**Sample Collection Date:**	04-May-2023 14:55 PM
Recommended By:		**Lab Reference No:**	2312366420

Sample Details : LC0405231892 **Sample Type : Serum**

Report

BIOCHEMISTRY

Test Name *(Methodology)*	**Result**	**UOM**	**Reference**
Urea *(Urease/GLDH)*	**90**	mg/dL	17 - 49
Creatinine *(Jaffe compensated)*	**2.3**	mg/dL	0.2 - 0.4
Uric Acid *(enzymatic colorimetric)*	**10.5**	mg/dL	3.4 - 7.0
Calcium *(5-Nitro-5'-methyl-BAPTA)*	9.4	mg/dL	8.8 - 10.8
Phosphorus *(molybdate UV)*	**4.8**	mg/dL	2.5-4.5
Sodium *(Ion Selective Electrodes)*	**134**	mmol/L	135 - 145
Potassium *(Ion Selective Electrodes)*	3.5	mmol/L	3.5-5.1
Chloride *(Ion Selective Electrodes)*	100	mmol/L	98-107
Bilirubin (T) *(Colorimetric diazo)*	0.53	mg/dL	0 - 1
Bilirublin (D) *(Diazo Gen.2 Jendrassik-Graf)*	**0.52**	mg/dL	0 - 0.2
Bilirubin (I) *(Calculated)*	0.01	mg/dL	0 - 0.9
ALT *(IFCC without pyridoxal phosphate)*	**262**	U/L	0 - 26
AST *(IFCC without pyridoxal phosphate)*	**56**	U/L	<=40
ALP *(IFCC)*	**710**	U/L	142 - 335
Total protein *(Biuret)*	7.3	g/dL	6.0 - 8.0
Albumin *(BCG)*	5.1	g/dL	3.8 - 5.4
Globulin *(Calculated)*	**2.3**	g/dL	3.0 - 3.7
A/G ratio *(Calculated)*	**2.2**		0.8-2.0

Dr. Sudip Kumar Datta (Biochemistry & Immunoassay) | Dr. Tushar Sehgal (Hematology & Coagulation) | Dr. Suneeta Meena (Serology) | Dr Hemang (Biochemistry & Immunoassay)
04-May-2023 21:22

SEROLOGY

Test Name *(Methodology)*	**Result**	**UOM**	**Reference**
HIV Combo (HIV 1, 2) *(ECLIA)*	0.24	COI	< 1.0 Non Reactive ≥ 1.0 Reactive
Anti HAV IgM *(ECLIA)*	0.29	COI	< 1.0 Non Reactive ≥ 1.0 Reactive

Attention: Please collect blood samples by puncturing the rubber cap of the vacutainers. Manual opening of caps and filling it must be avoided strictly. Lab reports are subjected to pre-analytical errors due to inappropriate patient preparation, phlebotomy practices, storage and transport. Please inform SMART Lab in case of any discrepancies with the expected results on the same day on Ext.no. 2526

NAME OF THE PATIENT: Kuldeep AGE /SEX: 2y/M UHID : 10665238

VITALS (DATE/ TIME)	11/5/23 9pm	11pm					
HEART RATE	148	136					
RESPIRATORY RATE	expiry	44					
SP02	98%	98% RA.					
BLOOD PRESSURE	expiry	88/52					
PERIPHERIES	Warm	Warm					
PP/CP	2s/2s	2s/2s					
CRT	<3sec	<3sec					
GCS AND PUPILS	irritable	Sleepy					
IONOTROPES							
U.O(ML/KG/HR)							
INVESTIGATION							
HB							
PLATELATES							
TLC(N/L/E/M/B)							
PH							
PCO2							
P02							
HC03							
LACTATE							
NA/K/CA/PO4							
UREA/R							
AST/ALT							
RBS							
INTERVENTION IF ANY							

1) - Inj DARBEPOIETIN 25 μg ½ vial every 2 weeks (2 हफ्ते में 1 बार)
2) - Syr Shelcal 5 ml TDS ⇒ दिन में 3 बार

empty stomach खाली पेट — ē meals खाने के साथ — ē meals खाने के साथ #

3) - Remove Foley's catheter

4) - Syr VIROFOL 5 ml OD (5ml दिन में 1 बार)
5) - Syr Vitamin D_3 1 ml OD (1ml दिन में 1 बार)] Ⓔ

F/u in 1 week Monday.

Please review ē Dr Manisha Jana # 8 CT Room today. @ 3 pm.

U/S KUB
RK - 2.5 cm } no hydronephrosis
LK - 3.6 "
CMD reduced/lost
echogenicity increased.
Small cysts (~2-3 mm size), <10 in number
F/s/o nephronophthisis
Manisha
1.5.23

4th floor mother child block

pH 7.29
HCO_3 12.7
BE -12.5
Na 137
K 3.0
Cl 112
Glu 95.

HbA1c 5.65%.
Urea (116)
Creat (2.7) UA (11.6)
Ca 9.9 Mg 3.4
P (6.7) T.bil (2.25)
SAP 756 D 2.05
Alb 5.3
PTH (1399) SGOT 847 SGPT 756

Diet as advised.
Adv (add) Syr NODOSIS 2 ml TDS × 3 months
- P/casualty: CMO, please remove foley's catheter.
- Syr Azimax Duo (200/5 ml) 4 ml BD × 5 days.
- Syr VIROFOL 5 ml OD × 3 months
- Syr VITAMIN D_3 2 ml OD × 3 m.
- Syr Shelcal 5 ml TDS × 3 m (empty stomach, ē meal, ē meal)
- Inj Darbepoietin 25 μg ½ vial every 2 weeks.

अ० भा० आ० सं० अस्पताल/A.I.I.M.S. HOSPITAL
बहिरंग रोगी विभाग /Out Patient Department
अस्पताल के अन्दर धूम्रपान मना है।/SMOKING IS PROHIBITED IN HOSPITAL PREMISES

OPR-6

एकक/Unit
विभाग/Dept.
नाम/Name
Regn. No.
पता/Address

UHID: 106656338
Dept No: 20230030011808
कमरा/ Room
14
Unit-I
Paediatric
Queue No: F30
04/05/2023
कुलदीप वेर्मा
KULDEEP VERMA
2Y M 19D / M(पुरुष)
SIONARAYAN SINGH VERMA
Add: VILL HINOTI POST LAKHONI TASHIL JIRAPUR HANOTI RAJGARH, MADHYA PRADESH. Pin 0. INDIA
Mon,Thu
Follow Up... General: 8-0 Reporting: 8:00 AM-9:00 AM

निदान/Diagnosis

दिनांक/Date	उपचार/Treatment
(24) Ht - 68 LC0405231892 106656338 KULDEEPVERMA	6.5 kg Kee other card Pmo personally to Fistula to be removed + Diet as advised 4ml सुबह 4ml शाम x 5 दिन ✓ — Syp Azimax 4 — 4 x 5 days 2ml सुबह 2ml दुपहर 2ml रात ✓ — Syp Modom 2ml — 2 — 2ml x 3 days 5ml दिन में 1 बार ✓ — Syp Virofol 5ml OD. 2ml दिन में 1 बार ✓ — Syp Vitamin B3 2ml OD. ✓ — Syp Shelcal 5ml — 5ml — 5ml x TP सुबह 5ml, शाम 5ml, दुपहर 5ml ✓ — Inj Darbepoietin 25 µg ½ [illegible] every 2 weeks S/C KCB. urea flow. TSH 10am 4C [illegible] Anti HAV, IgM, Anti HCV, SGOT, SGPT, HBsAg, Bil T/D, Anti HBsAb + Child exome sequencing — not done [illegible] cystic kidney disease

अ० भा० आ० सं० अस्पताल/A.I.I.M.S. HOSPITAL
बहिरंग रोगी विभाग /Out Patient Department
अस्पताल के अन्दर धूम्रपान मना है। SMOKING IS PROHIBITED IN HOSPITAL PREMISES

स्वच्छ भारत — एक कदम स्वच्छता की ओर

OPR-6

UHID: 106656336
Dept No: 20230030011606

C-212
Unit-I
Paediatric
Queue No: F35
17/07/2023

कुलदीप वेर्मा
KULDEEP VERMA
2Y 3M 2D / M(पुरुष)
S/O NARYAN SINGH VERMA
Add: VILL HINOTI POST LAKHONI TASHIL JIRAPUR HANOTI RAJGARH, MADHYA PRADESH, Pin 0, INDIA

सोम,गुरु
Mon,Thu (सोम,गुरु)

Follow Up... General F0 Reporting: 8:00 AM-9:00 AM

बा०रो०वि० पंजीकृत सं०/O.P.D. Regn. No. ____

आयु Age	पता/Address

निदान/Diagnosis CKD 4-5 / Hypodysplastic or nephronophthisis / Anemia ; MBD / -

दिनांक/Date	उपचार/Treatment
Ht = 79 cm 16/7/2023 Hb = 6.5 TLC = 760 Platelet = 2.33 N/L/M/E = 31.8/57.9/10.3/01 SGOT/SGPT = 57/3.4 Bil T = 0.11, D = 0.19 Ur/Cr = 112/ ~~[illegible]~~ Ca = 8.53 Uric acid = 10.36	7.1.6 CKD / unknown cause : Genetic ? nephronophthisis ? Hypodysplastic kidney Short stature + frontal bossing + pallor. Counselled · Palliative care plan Syr VITROFOL 7.5 ml OD - Drops Vitamin D_3 2 ml OD - Syr Shelcal 5 ml TDS × 3 mo → Inj DARBEPOIETIN 25 µg s/c 1/2 weeks - Syr Nodosis 2.5 ml - 2.5 ml - 2.5 ml - Nutrition re-emphasized Hearing required 2.17.7 3.00 MCB 8th floor 802 / 823 · 4:30pm CARUT.

शरीरमाद्यं खलु धर्मसाधनम्

अ० भा० आ० सं० अस्पताल/A.I.I.M.S. HOSPITAL
बहिरंग रोगी विभाग /Out Patient Department
अस्पताल के अन्दर धूम्रपान मना है।/SMOKING IS PROHIBITED IN HOSPITAL PREMISES

Surgical O.P.D.

शरीरमाद्यं खलु धर्मसाधनम्

OPR-6

एकक/Unit
विभाग/Dept.
नाम/Name

Regn. No.
पता/Address

बाल शल्यचिकित्सा
UHID: 106856338
Dept No: 20230220002007
कक्ष / Room
G-31
Unit-I
Paediatric Surgery OPD
Queue No: F33
29/04/2023
कुलदीप वेर्मा
KULDEEP VERMA
2Y M 14D / M(पुरुष)
S/ONARYAN SINGH VERMA
Add: VILL HINDTI POST LAKHONI TASHL JIRAPUR HANDTI RAJGARH, MADHYA PRADESH, Pin 0, INDIA
Follow Up... General F0 Reporting: 8:00 AM-9:00 AM

प्रमुख गोस्वामी
अपर आचार्य
बाल शल्य चिकित्सा OPD

निदान/Diagnosis PUV

दिनांक/Date	उपचार/Treatment
50 [redacted] e'14	c/o straining during micturition. MCU (19/4/23) ↳ Trabeculated bladder post urethra dilated BC 120 GFR (26/4/23) - 17 Hgm (17/4/23) - 8.4 | 1770 | 293k U/C - 94/2.6 Na/K - 137/3.5 ALP - 425. Ferritin - 595 Ped nephro - 1/5/23 USG KUB - 3/5/23. Adw Dr. Prabudh Goel Adv · Catheterisation (35) - PAC · Repeat Hgm / GFR on 5/ ... f/u in PSOPD on 6/5/23 for OT
Appointment on 6/5/23 for Deptt./Clinic Cont A-Z vitakal	

CLEAN AND GREEN AIIMS / एम्स का यही संकल्प, स्वच्छता से काया कल्प
अंगदान-जीवन का बहुमूल्य उपहार/ORGAN DONATION - A GIFT OF LIFE
O.R.B.O., AIIMS, 26588360, 26593444, www.orbo.org Helpline - 1060 (24 hrs service)

Pradhan Mantri Jan Arogya Yojana
Ayushman Bharat
PM-JAY
(pmjay.gov.in)

मेरा अस्पताल
My Hospital
merahospital.nhp.gov.in

JAN LAKSHYA TRUST

UHID: 106656338
Dept No: 20230303011609

C-212
Unit: Paediatric
Queue No: F36
12/06/2023
सोम,गुरु
Mon,Thu (सोम,गुरु)

कुलदीप वेर्मा
KULDEEP VERMA
2Y 1M 27D / M(पुरुष)
S/ONARYAN SINGH VERMA
Add: VILL HINOTI POST LAKHONI TASHIL JIRAPUR HANOTI RAJGARH, MADHYA PRADESH, Pin 0, INDIA
Follow Up... General F0 Reporting: 8:00 AM-9:00 AM

OPR-6

ब०रो०वि० पंजीकृत सं०/O.P.D. Regn. No. _____

आयु Age	पता/Address

निदान/Diagnosis

CKD

दिनांक/Date	उपचार/Treatment
15/6/23	7.K USG KUB Small R & L Kidneys (2.5 cm) (3.6cm) CMD - loss small cysts (~ 2-3mm) <10 number F/s/o nephronophthisis Clinical exome → no pathogenic/ likely pathogenic variant causative of reported phenotype. MCU mild irregular contour, dilated posterior urethra with abrupt narrowing @ junction of membranous & bulbar urethra • Recurrent fever episodes with pool stream of urine, pallor, failure to thrive MCU - dilated posterior urethra

Hb - 7.1
TLC - 10,500
N23 L63
Plt → 2.43L
Urea/ 51.7/ Creat 3.08
Na/K - 134.6/3.98
OT/PT - 11/31.
HIV - Neg.
HBsAg

Wt/Age -5.3z
Ht/Age -6.5z
wt/Ht -3.03z

QUANTITATIVE DATA:

Parameters	Left kidney	Right kidney
GFR (ml/min)	4.95	2.5
2 min diff. (% of relative function)	--	--
T_{max} (mins)	--	--
Retention (%)	--	--

CLINICAL IMPRESSION:

1. **Almost bilateral non-functioning kidneys.**
2. **Scan pattern is compatible with chronic parenchymal dysfunction leading to chronic renal failure pattern.**
3. **Poor visualization of bladder activity.**

Thanks for the referral.

Checked By : -- 13742 DR G N MAHAPATRA

Dr. G. N. Mahapatra
Sr. Consultant and Director,
Department of Nuclear Medicine & PET-CT

Note: This report is purely for diagnostic correlation with the other reports and clinical data of the patient by the treating physician.This cannot be used for medico legal purposes.For any clarification, corrections and missed findings the referring Physician may kindly call 9962784274/9820801633 directly to the above signatories. Direct enquiry by patient/relatives will not be entertained.

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 / 990, Web: www.medgenome.com

MEDGENOME

Average sequencing depth (x)	Average on-target sequencing depth (x)	Percentage target base pairs covered		
		0x	≥5x	≥20x
453	163.30	0.07	99.82	99.50

Total data generated (Gb)	7.45
Total reads aligned (%)	99.99
Reads that passed alignment (%)	92.21
Data ≥Q30 (%)	96.71

$The classification of the variants is done based on American College of Medical Genetics as described below [PMID: 25741868].

Variant	A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).
Pathogenic	A disease causing variant in a gene which can explain the patient's symptoms has been detected. This usually means that a suspected disorder for which testing had been requested has been confirmed.
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 has been detected, but it is difficult to classify it as either pathogenic (disease causing) or benign (non-disease causing) based on current available scientific evidence. Further testing of the patient or family members as recommended by your clinician may be needed. It is probable that their significance can be assessed only with time, subject to availability of scientific evidence.

*The transcript used for clinical reporting generally represents the canonical transcript (MANE Select), which is usually the longest coding transcript with strong/multiple supporting evidence. However, clinically relevant variants annotated in alternate complete coding transcripts could also be reported.

Variants annotated on incomplete and nonsense mediated decay transcripts will not be reported.

*The *in-silico* predictions are based on Variant Effect Predictor (v104), [SIFT version - 5.2.2; PolyPhen - 2.2.2; LRT version (November, 2009); CADD (v1.6); Splice AI; dbNSFPv4.2] and MutationTaster2 predictions are based on NCBI/Ensembl 66 build (GRCh38 genomic coordinates are converted to hg19 using UCSC LiftOver and mapped to MT2).

Diseases databases used for annotation includes ClinVar (updated on 5082021), OMIM (updated on 5082021), HGMD (v2021.3), LOVD (Nov-18), DECIPHER (population CNV) and SwissVar.

LIMITATIONS

- Genetic testing is an important part of the diagnostic process. However, genetic tests may not always give a definitive answer. In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology. Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in {my/my child's} family may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results.

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 / 990, Web: www.medgenome.com

MEDGENOME

- The sensitivity of NGS assay to detect copy number variants (CNV) is 70-75%. We recommend discussing alternative testing methodology options with MedGenome Tech Support (techsupport@medgenome.com) as required. In case clinician is suspecting CNV as an important genetic etiology, alternate tests like microarray/ MLPA or qPCR may be considered after discussing with the MedGenome TechSupport team.

Sandhya Nair, Ph.D
Sr. Manager - Variant Interpretation

Balaji Rajashekar, Ph.D
Director - Clinical Bioinformatics

Dr. Vibha Jain, MBBS, DNB (Obs & Gynae), DNB-SS (Medical Genetics); PGDHHM, FICCM
Consultant - Clinical Geneticist

APPENDIX

TEST METHODOLOGY

Targeted gene sequencing: Selective capture and sequencing of the protein coding regions of the genome/genes is performed. Variants identified in the exonic regions are generally actionable compared to variants 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 depth of >80-100X on Illumina sequencing platform. We follow the GATK best practices framework for identification of germline variants in the sample using Sentieon [Sentieon]. The sequences obtained are aligned to human reference genome (GRCh38) using BWA aligner [Sentieon, PMID:20080505] and analyzed using Sentieon for removing duplicates, recalibration and re-alignment of indels [Sentieon]. Sentieon haplotype caller is then used to identify variants in the sample. The germline variants identified in the sample is deeply annotated using VariMAT pipeline. Gene annotation of the variants is performed using VEP program [PMID: 20562413] against the Ensembl release 99 human gene model [PMID: 29155950]. In addition to SNVs and small Indels, copy number variants (CNVs) are detected from targeted sequence data using the ExomeDepth method PMID: 22942019]. This algorithm detects CNVs based on comparison of the read-depths in the sample of interest with the matched aggregate reference dataset.

Clinically relevant mutations in both coding and non-coding regions are annotated using published variants in literature and a set of diseases databases : ClinVar,OMIM, HGMD, LOVD, DECIPHER (population CNV) and SwissVar [PMID: 26582918, 18842627, 28349240, 21520333, 19344873, 20106818]. Common variants are filtered based on allele frequency in 1000Genome Phase 3, gnomAD (v3.1 & 2.1.1), dbSNP (GCF_000001405.38), 1000 Japanese Genome, TOPMed (Freeze_8), Genome Asia,HmtDB and our internal Indian population database (MedVarDb v2.1) [PMID: 26432245, 32461613, 11125122, 26292667, 33568819, 31802016, 22139932]. Non-synonymous variants effect is calculated using multiple algorithms such as PolyPhen-2, SIFT, MutationTaster2 and LRT. Clinically significant variants are used for interpretation and reporting.

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 / 990, Web: www.medgenome.com

MEDGENOME

- In a very few cases genetic test may not show the correct results, e.g. because of the quality of the material provided to MedGenome. In case where any test provided by MedGenome fails for unforeseeable or unknown reasons that cannot be influenced by MedGenome in advance, MedGenome shall not be responsible for the incomplete, potentially misleading or even wrong result of any testing if such could not be recognised by MedGenome in advance.
- Variants of uncertain significance (VUS) which are mentioned in the report need to be further correlated with the clinical phenotype, reports of other investigations, segregation analysis in the parents or affected/unaffected family members. MedGenome shall not be responsible for the inappropriate interpretation/ communication/ clinical actions/ reproductive decisions based on the VUS reported. The classification of VUS may change as the clinical phenotype evolves or more information is available in the scientific literature/ annotated databases.
- This is a laboratory developed test and the development and the performance characteristics of this test was determined by MedGenome.

END OF REPORT