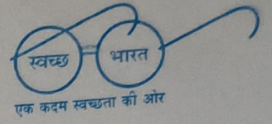




अ० भा० आ० सं० अस्पताल / A.I.I.M.S. HOSPITAL

बहिरंग रोगी विभाग / Out Patient Department

अस्पताल के अन्दर धूम्रपान मना है। SMOKING IS PROHIBITED IN HOSPITAL PREMISES



Adult Genetic. UHID: 106750656 Dept No.: 20231100001761 QR Code	कमरा / Room C 214 Unit-I ADULT GENETIC OPD मंगल, शुक्र,	OPR-6 पंजीकृत सं०/O.P.D. Regn. No. _____ पता/Address GC: 2276/23 FHM UND
शरीरमापन खतु एकक/Unit विभाग/Dep	आयु 13/10/2023 Queue: N8 Barcode Reporting 09:27:39	
माधुरी भास्कर MADHURI BHASKAR W/O RAJU BHAI PARDESHI 32Y 4M 18D / F (महिला) Add: DIST CIVIL COURT PRAYGRAJ, UTTAR PRADESH, INDIA Mob: 8765173440 New Patient General 0		

निदान/Diagnosis: **Hypertriglyceridemia ↓ evaluation**

दिनांक/Date	उपचार/Treatment										
13/10/23	Referred from Cardiology OPD on v/o <u>Hypertriglyceridemia</u> lipid profile ↓ (27/9/23) (On L ₂) • T. chol - 387 mg/dl (<200) • TG - 604 mg/dl (<150) • HDL - 30.9 mg/dl (>50) • NonHDL chol } - 356 mg/dl (<130)										
Echo (N)	On Rosuvastatin, Brilho, fenofibrate, ezetimibe H/o High myopia (-9 D) & ? Right blindness (+). APOA 5 / exon 3 / c853 c>T / hetero / (VUS) / hyper / HD = High triglyceridemia susceptibility										
<table border="1"> <thead> <tr> <th>WTG</th> <th>TG</th> </tr> </thead> <tbody> <tr> <td>Dec 21</td> <td>298</td> </tr> <tr> <td>Mar 22</td> <td>873</td> </tr> <tr> <td>Sep 22</td> <td>1098</td> </tr> <tr> <td>Sep 23</td> <td>604</td> </tr> </tbody> </table>	WTG	TG	Dec 21	298	Mar 22	873	Sep 22	1098	Sep 23	604	(TG not known) MI 46y High myopia 13y, MTP, 9y, MTP
WTG	TG										
Dec 21	298										
Mar 22	873										
Sep 22	1098										
Sep 23	604										

Adv
① Eye evaluation - RPC for Funder's evaluation
info myopia and night blindness

- ② Literature review regarding variant
- ③ fasting lipid profile for both children

Rpt 2 weeks (Mon 9am) in 214

Adult Genetic, UHID: 106750656
Dept No: 20231100001761
माधुरी भास्कर / MADHURI BHASKAR
W/O RAJU BHAI PARDESHI
32Y 11M 0D / F/(महिला)
DIST CIVIL COURT PRAYGRAJ, UTTAR
PRADESH, INDIA
Ph: 8765173440
Follow Up Patient
कमरा / Room C 214
Queue / संख्या F9
Unit-I, ADULT GENETIC OPD,
FRI मंगल शुक्र,
Reporting: 08:50:52
26/04/2024

20th May 2024
Appointment on
Metabolic Clinic
Dewi R

35

Follow up case
Seen in RPC - Myopic funder - Adv to
use glasses
Son detected to have hpl myopia
Lipid profile of both children - Normal
Ophthalmic evaluation of daughter
Already under follow up of Adult Ophthalmology
on drug therapy + dietary interventions
Reanalysis of WES data for evaluation to explain
familial hpl myopia - REANALYSIS in follow up

Appt for genetic clinic 3rd Monday 2PM
(GMC)

Mr. Dewinder Sub Registrar
Counsellor

Madhulika
डॉ० मधुलिका काबरा / Dr. MADHULIKA KABRA
आचार्य एवं विभागाध्यक्ष / Professor & Head
बाल चिकित्सा विभाग / Dept. of Pediatric
अ.भा.आ.सं./A.I.I.M.S.
नई दिल्ली / New Delhi-110029

Mr 102 w
wt 65 kg

हृदय वक्ष एवं तंत्रिका विज्ञान केन्द्र
ब० रो० वि०

अ० भा० आ० सं०, नई दिल्ली-110029

Cardiothoracic & Neurosciences Centre, O.P.D.

CV 2023/014/0013309 ₹0 Cardiology
UHID: 106750656 Cardiology

दिनांक/Date

Date 26/05/2023 MON, FRI

Name MADHURI BHASKAR 32Y /F

विभाग
Deptt.

W/O RAJU BHAI PARDESHI

Phone No. 8765173440

Consultant Room 8

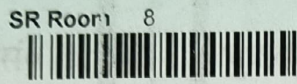
DR SANDEEP SETH

S. Agstam

DR MOHSIN

उम्र
Age

यू०एच०आ
UHID No.



लिंग
Sex

निदान
Diagnosis

? familial hypertriglyceridemia

WES → VUS detected in
APOA5 gene

No CV symptoms.

ECG: T↓ V₁—V₃

tubo: (N)

wt 93/59
ht 29

Mother
had SCD
@ 42yr age

1. T. Rosuvastatin 40mg HS. ————

2. T. Bivello 180mg BD 0

3. T. fenofibrate ~~45mg~~ 200mg qd

4. T. Eicosapentaethyl 1000mg BD. 0—0

दिनांक
Date

SMART LAB

HBAC
73/74/154

25
= ✓

Genetics OPD review

Ry. G. S. T
post report

Urine R/m -
& Urine A/C -
lipid profile - ✓

pediatric - R 27.
screenshot.

STC-260523034 106750656
 LC2605232072 106750656
 LH2605231517 106750656
 MADHURIBHASKAR

proh

Appt for
pediatric OPD
Adult Genetics
13/10/23
9 AM Rm 214

New RAK OPD
2M Kow
C wing

11/10/23
डॉ. मधुलिका काबरा / Dr. Madhulika Kabra
आचार्य / Professor
अनुवांशिकी एकक (बालरोग चिकित्सा विभाग)
Division of Genetics (Department of Pediatrics)
आ.आ.स., नई दिल्ली / A.I.I.M.S. New Delhi-29
DMC Regn. No. 1680

हृदय वक्ष एवं तंत्रिका विज्ञान केन्द्र
ब० रो० वि०
अ० भा० आ० सं०, नई दिल्ली-110029
Cardiothoracic & Neurosciences Centre, O.P.D.
A.I.I.M.S., New Delhi-110029

दिनांक/Date 106750656 / 13/10/23

विभाग
Deptl.

नाम
Name Madhuri Bhaskar

उम्र
Age 32y

ब०रो०वि०सं०
O.P.D. No.

पुत्र/पुत्री/पत्नी
S/D/W Manoj Bhaskar

लिंग
Sex f

13309/23

निदान
Diagnosis Hypertension

R8 (46)

13/10/23

Coenzyme Q₁₀ on

Ren x 6m

crankly
x 6m

दिनांक
Date

R8 26
26/4/24

CST x by
Jm
26/4/24

Patient Details

Name :	MADHURI BHASKAR	Sex / Age :	FEMALE / 32 Years	Case ID :	21107401264
Ref By :	Dr. Deepthi Saxena	Dis. Loc. :		PT. ID :	2022903396
Test Name :	ORION (WES-Whole Exome Sequencing)	Bill. Loc. :	SGPGI HOSPITAL, LUCKNOW	PT. Loc. :	

Sample Details

Registration Date & Time :	2022-11-15 16:19:33	Sample Type :	Whole Blood EDTA	Ph # Re :	
Sample Date & Time :	2022-11-15 16:19:00	Ref ID 1 :		Acc. Remarks :	
Report Date & Time :	2022-11-30 10:34:51 AM	Ref ID 2 :		PT. Loc. :	

Clinical History

Consanguinity: Absent

Clinical symptoms: features of weakness and episodes of syncopal attacks 3-4 times within a period of 4 months 1 year back. H/o difficulty in night vision with no history of chest pain or diabetes or hypertension.

Investigations done:

Serum triglycerides- 786 mg/dl, serum cholesterol- 144 mg/dl, HDL- 36 mg/dl, LDL- 58.7 mg/dl, VLDL- 157.2 mg/dl, RBS- 97mg/dl and hba1c- 5.6 %

Family history: Proband's mother passed away at 46years of age due to myocardial infarction

Clinical suspicion: Familial hypertriglyceridemia.

Test Results and Interpretation

HETEROZYGOUS VARIANT OF UNCERTAIN SIGNIFICANCE (VUS) DETECTED: CLINICAL CORRELATION RECOMMENDED

Summary of Variants

Gene and Transcript	Exon/Intron Number	Variant Nomenclature	Zygoty	Classification	Disease	Inheritance
APOA5 (NM_001371904.1)	Exon 3	c.853C>T p.Leu285Phe [Depth-56X]	Heterozygous	Uncertain significance	{Hypertriglyceridemia, susceptibility to}	Autosomal dominant

Variant Details

APOA5

Page 1 of 11

body plan

(52)

हृदय - वक्ष एवं विज्ञानतंत्रिका केन्द्र

CARDIO-THORACIC & NEURO-SCIENCES CENTRE

अखिल भारतीय आयुर्विज्ञान संस्थान
 A.I.I.M.S. Hospital
 नाम Modhu
 एक्स-रे नम्बर _____ तिथि _____
 X-Ray No. _____ Date _____
 हस्पताल यू.एच.आई.डी. नं. _____ वार्ड/ ओ. पी. डी. _____
 Hosp. UHID No. _____ Indoor/Outdoor _____
 Examination Required _____
 चिकित्सक की जांच रिपोर्ट :
 Clinical Information :

एक्सरे - फार्म
 X-RAY REQUISITION FORM
 आयु _____ लिंग _____ आय _____
 Age _____ Sex _____ Income _____
 चिकित्सक विभाग
 Referring Unit
 रोगी स्थिति
 Ambulatory/Non

Cartha Intern Medist
Thudhu

Plu MD

किसी दवा का बुरा प्रभाव
 Any History of Allergy _____
 अन्तिम माहवारी तिथि
 LMP _____
 कोई पुराने एक्स-रे
 Any Previous X-Rays _____

[Signature]
 चिकित्सक के हस्ताक्षर
 SIGNATURE OF MEDICAL OFFICER
 रेडियाग्राफर के लिए
 FOR RADIOGRAPHERS USE

पहचान चिन्ह Identification Mark
अंगूठा निशान Thumb Impression

कमरा न. Room No.	फिल्म साइज Size & No. of Films	के.वी. एम.ए.एम. KV MAS
हस्ताक्षर Signature		

सामुदायिक नेत्र विज्ञान Community Ophthalmology

सातवीं मंजिल / 7th Floor

डा० राजेन्द्र प्रसाद नेत्र विज्ञान केन्द्र

Dr. Rajendra Prasad Centre for Ophthalmic Sciences

आ० भा० आ० संस्थान, अंसारी नगर,

A.I.I.M.S., Ansari Nagar

नई दिल्ली - ११००२९

New Delhi - 110029

Phone No.: 26593046

पंजी० सं० / Reg. No.:- _____

दिनांक / Date :- 13-10-2023

स्थान / Site :- _____

नाम/Name	पुत्र/पुत्री/पत्नी S/D/W	लिंग Sex	आयु Age	पता/Address
Madhuri Bhaskar		P	32	

निदान
Diagnosis

उपचार Treatment

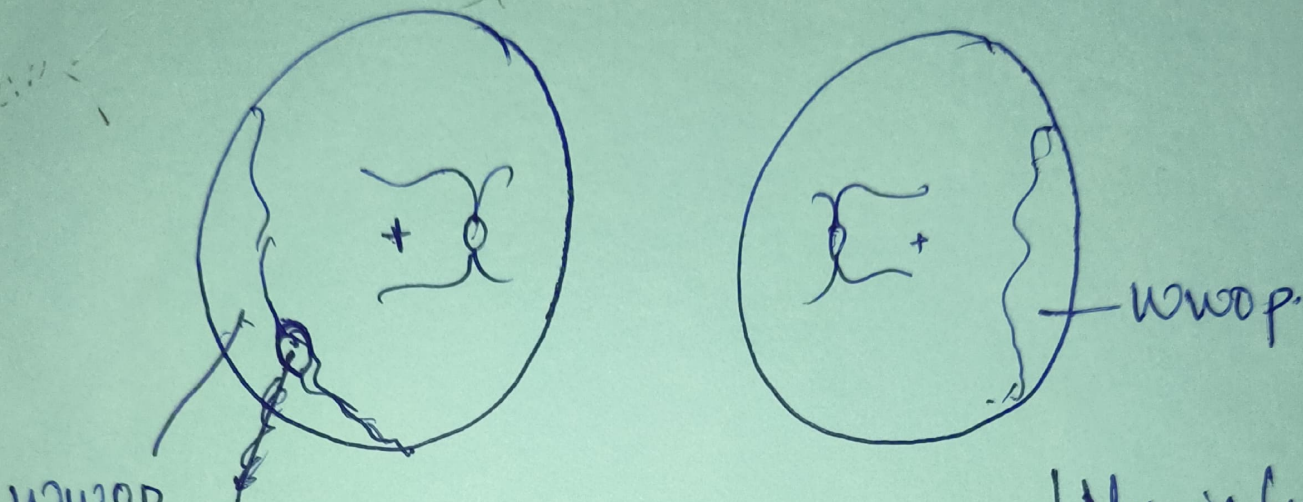
M { 5/60
5/60

NET { 17 mmHg
16 mmHg

Adv

- Ace { - 9.00 S ————— ६६६
- 10.00 S ————— ६९९

- नेत्र विशेषज्ञ की सलाह का पालन करें एवं अपनी आँखों को घरेलू दवाइयों के जोखिम से बचाएँ।
- अपनी आँखों को धूल, धुआँ, तेज धूप और गन्दे पानी से बचाएँ।
- अपनी आँखों को चोट लगने से बचाएँ।
- विशेष परिस्थितियों में सहायता के लिए नेत्र स्वास्थ्य कार्यकर्ता की सेवा का लाभ उठाएँ।



pigmented dot within area of wwop.

Fn < Myopic fundus
 CDR-0.3:1
 AR-2:3
 PR (+)
 NRR healthy

As-wop

Adv
- Use glasses.

नेत्र ईश्वरीय सर्वश्रेष्ठ उपहार है जिनका मनुष्य जीवन में दान करना परमश्रेष्ठ है। इनकी पूर्ण रक्षा कीजिए ताकि ये आपकी रक्षा कर सकें।

Eyes are God's most precious gift to man kind. Take full care of them So that they can take care of you



अखिल भारतीय आयुर्विज्ञान संस्थान, नई दिल्ली
ALL INDIA INSTITUTE OF MEDICAL SCIENCES (AIIMS)

New Delhi, null

नकदी रसीद / CASH RECEIPT

संस्थान / ALL INDIA INSTITUTE OF MEDICAL SCIENCES

अंसारी नगर, नई दिल्ली-110029 / Ansari Nagar, New Delhi-110029

दूरभाष / 26588500
Phones / 26588700

नियुक्ति पर्ची
APPOINTMENT SLIP



रसीद संख्या / Receipt No

Done By-Mr.ROHIT AGGARWAL DEOSWSC (New)

General ₹ 0.0

Print Appointment Slip

दिनांक / Date

जमाकती / Received From

Department Name: Adult Genetic/ADULT GENETIC SPECIAL CLINIC

रोगी प्रकार / Patient Type :

ओ.पी.डी./यू.एच.आई.डी. स / OPD UHID No. Reporting Time: 11:30 PM

Appointment Date: 20/05/2024

कक्ष संख्या / Room No.

के नामे / ON ACCOUNT OF

Doctor Name: Dr. Maulika Nabra

Appointment Request date

26/04/2024

Name of Patient

MRS. MADHURI BHASKAR

Appointment No

2024042608544

Sex

Female

Age

32 years 11 months

Contact Details

Mobile: XXXXXXX440

Request Mode

counter

Queue No

NI

Remarks:

Your UHID Is : 106750656.

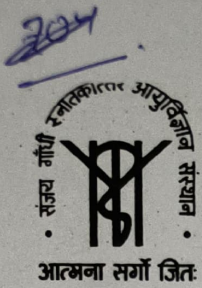
भुगतान का प्रकार / Payment Mode :

रूपये / INR (Rs.) :

रूपये शब्दों में / Rs. in Words

यह कम्प्यूटर द्वारा जारी की गई रसीद है और इसमें हस्ताक्षर और मोहर अपेक्षित नहीं है।

THIS IS COMPUTER GENERATED SLIP AND DOES NOT REQUIRE SIGNATURE AND STAMP



10008

Dr. Deepthi

OUT PATIENT RECORD

(Tue/Wed/Sat)

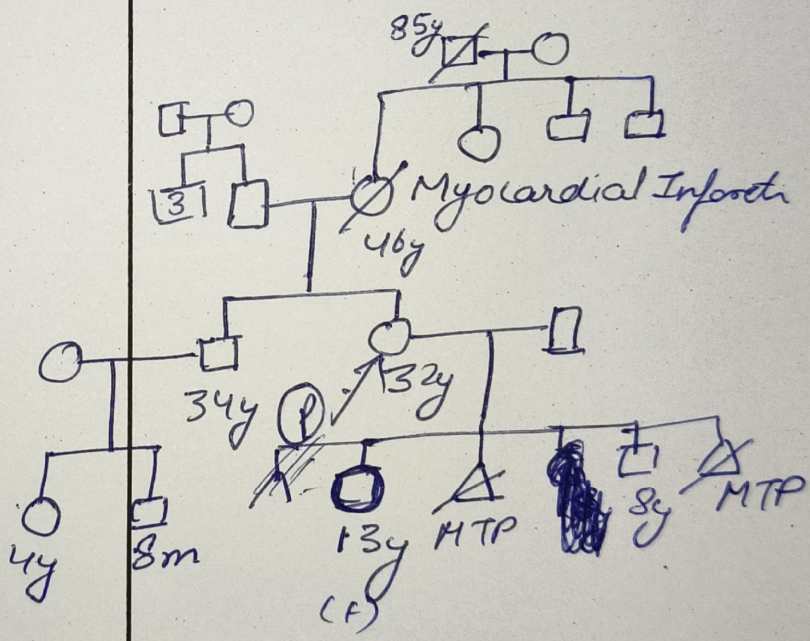
DEPARTMENT OF MEDICAL GENETICS
SANJAY GANDHI POSTGRADUATE INSTITUTE
OF MEDICAL SCIENCES, LUCKNOW- 226014

: 2494076 (OPD)
Tel. : 2494325 (Office)
: 2494348/2494334

Name Madhuri Bhaskar CR No. 20229083 Diagnosis
32y IF 96

Date Clinical Condition Advice

08/11/2022



H/O weakness, syncope attacks 3-4 times within 3-4 months on exertion - 1 year back.

H/O Night blindness

No DM/HTN

H/O hypertriglyceridemia

No xanthoma

wt: 59.7kg

Tg 786 mg/dl, s. Cholesterol 144 mg/dl

HDL 36 mg/dl, LDL 58.8 mg/dl

VLDL 157.2, #

RBS 97 mg/dl, HbA1C 5.6

Please bring this Card at each visit.

02/11/22

Date	Clinical Condition	Advice
	<p>△ hypertriglyceridemia.</p> <p>also family history.</p> <p>Referred from cardiology.</p>	
		<p>Adv- whole exome sequencing.</p> <p>to look for monogenic causes. (test not available in SCP62)</p> <p>- If any genetic cause is identified, then risk of recurrence to siblings and offspring. ~ 50%.</p> <p>- Lipid profile of at-risk relatives can be done.</p> <p>- A/c with reports after 2 months (Tue/Wed).</p>

J. Saxena

Date	Clinical Condition	Advice
17/10/23	<p>Brother's lipid profile (Mayer Rajubhai, Pardihi)</p> <p>TGA 386.3 (<200)</p> <p>T. Cholesterol 190.6</p> <p>HDL 36.5, NonHDL 154.1, LDL 76.84</p> <p>VLDL 77.26, Chol/HDL 5.22 (<4.00)</p> <p>LDL/HDL 2.11, HDL/LDL 0.48</p>	
	<p>Father's lipid profile</p> <p>Chol. 183, TGA 81, HDL 73, LDL 93</p> <p>VLDL 16, Chol/HDL 2.5, LD/HDL 1.2</p>	
		<p><u>WES</u>: Heterozygous missense variant in APOA5 gene</p> <p>APOA5 (NM_001371904.1): c.853C>T (p. Leu 285Phe)</p>
		<p><u>VUS</u>, OMIM phenotype: Hypertriglyceridemia, susceptibility to CAD.</p>
		<p>Key variant likely causative of the phenotype has been detected.</p>
		<p>Adv- Reanalysis of raw data.</p>
		<p>- follow up in March, 2023</p>

X