

सेवा में

दुस्ती

मिशन चाइल्ड केयर

ब्राह्मपुर - J-100

दिल्ली - 110034



प्रत्यक्ष

आपसे यह निवेदन है कि मैं सुशील कुमार हैं
मेरी बच्ची का नाम विशी कुमार है जो कि केवल
17 महीने का है। मेरी बच्ची को रिड की इडुई
में बिगारी बतई गई है। बिम बतई से वह
बच्चा अपने पैरों पर खड़ा होने का काम नहीं
कर पा रहा है।

इसके दिल्ली में केएम हस्पताल में उसको दिखया
जा जहां उसमें इलाज में हर महीने दस से
पन्नाइ इलाज कि दवाइयो का खर्च बताया गया
है आपसे मैं प्रार्थना है कि जैसी सहायता कि
आस मैं एक ठोने में काम करके अपना घर
पलता है

धन्यवाद

विशी कुमार का पिता



For MISSION CHILD CARE

Handwritten Signature

Auth. Sign.

20/11/18





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

शरीरमार्च खलु वर्मसाधनम्

OPR-6

एकक/Unit
विभाग/Dept.

General

 DeptSeq: 539
 P0 104049259
 Dept: Paediatrics
 Clinic No: NMC- Clinic No. 2018/NMC/399
 Unit: Unit-II
 Name: RISHI KUMAR
 Room: 3
 S/O SUSHIL KUMAR, 1Y 4M 10D, F
 N/6
 Ph: 7065924814
 Days: Friday
 VILL PINAPRA FATEHYABAD, DISTT AGRA,
 UTTAR PRADESH, INDIA
 App. Date: 16/11/2018
 Appt. ID: 
 2018111611173

कृत सं०/O.P.D. Regn. No.

पता/Address

निदान/Diagnosis

SMA Type 2

दिनांक/Date

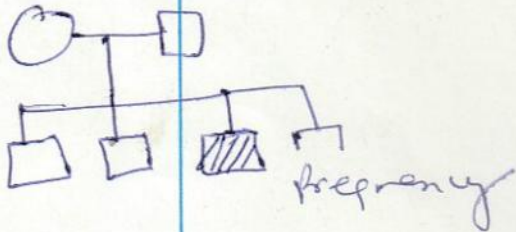
उपचार/Treatment

41

7.8K

• sitting - 14m

• PCR of SMN gene - Deletion identified in exon 7 of SMN gene



• vaccination advised - not done

Advice

• file ~~to be~~ made next visit

• PMK-OT/PT

• vaccination to be done

• follow up after 3m

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)

2y pneumococcal 1st dose
vaccine
(Prevenar) in stat. →
13

2y Hib 1st dose
in stat

2y influenza in stat

07/12/18

2pm
(RMS)

Analysis

gim
20/11/18

SENIOR RESIDENT
Department of Pediatrics
All India Institute of Medical Sciences
Ansari Nagar, New Delhi-110029

07/12/18
SMN 2a

Adv :

Physiotherapy - PT/OT (RMS)

File

- Card
↓
NMC / Inday 2pm

- Exp x 3mo

Analysis

(1) Dr. O.P. Narayan



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

शरीरमाघं खलु वर्नसावन्म

OPR-6

एकक/Unit _____

विभाग/Dept. PAEDIATRICS

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

| नाम/Name | पिता/पुत्र/पत्नी/पुत्री F/S/W/D of | लिंग Sex | आयु Age | पता/Address |
|-------------|---------------------------------------|-------------|------------|---------------|
| RISHI KUMAR | SHUSHIL KUMAR | M | 15 MONTHS | AAGRA U.P. |

निदान/Diagnosis

~~Highly Hypotonic~~
IDD (predominantly motor delay)

दिनांक/Date

उपचार/Treatment

5/04/2018

17 Feb / 0

? SMA
flppy infant.

1) predominantly motor delay - NO follow up

2) alert looks.

3) ↓ Anigravily marks (+)

No Seizures
facial smile (+)

O/E:

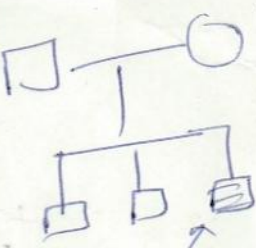
Typhoid fever - doubtful
Polymyogalax (+)

O/E - 45w

Anigravily marks ↓.

Myoclonic / slip sign (+)

No dysmorphism, hyperextensibility (+)



CIAB

SD

No meo
any

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)

Possibilities

- 1) SMA - ?
- 2) Cong myopathy
- 3) Cong myofibrillar myopathy

2

CPK & bhl

b

1. Review & CPK

Plan SMN gene deletion study.

Dr Sardul

SR SENIO RESI
Department of Pediatric
All India Institute of Medical Sciences
Ansar Nagar, New Delhi-110029

05/10/18

Review (K102)

SMN gene deletion study

old of block

(ATM @ 10:55)

- ① Referred to PMR for OTPT
- ② Muscl clinic, Fri-2PM

b

1. Review & SMN study

Muscle clinic
RN (3) Friday
2PM

SMACT,
16/11/18
Adv

- ① Inj: Pneumovax
↓
PCV 13 1st dose in stat
- ② Inj: Hib 1st dose in stat
- ③ Inj: Influenza, instat



अखिल भारतीय आयुर्विज्ञान संस्थान, नई दिल्ली-110029

ALL INDIA INSTITUTE OF MEDICAL SCIENCES, NEW DELHI -110029

आपातकालीन विभाग



(DEPT. OF EMERGENCY MEDICINE)

UHID No:104049259

आपातकालीन नं.(Emergency No): 2018/030/0113798

दिनांक DATE: 06/10/2018

समय TIME: 09:37:17 AM

NON-MLC

नाम NAME: **BABY RISHI KUMAR**

आयु AGE : 1 years 3 months

लिंग/SEX : F

S/O : SUSHIL KUMAR

पता ADDRESS:

मकान संख्या H.NO:

VILL PINAPRA FATEHYABAD

गली / मुहल्ला STREET/MOH: DISTT AGRA

शहर/प्रखंड CITY/BLOCK:

पिन PIN:

राज्य STATE:

UTTAR PRADESH

दूरभाष सं. PHONE NO:

मोबाइल MOBILE NO:

7065924814

स्थान Location:

Paediatrics Emergency

द्वारा BROUGHT BY: Relative : FATHER

Criticality: Red / Yellow / Green

Triage: Responsive/ Unresponsive HR /min BP mmHg RR /min spO2 %

Shifted to Paeds/ Main/ New Emergency

Presenting Complaints

Primary Assessment (ABCDE) : Assessment Pentagon

| Airway | Circulation | Disability |
|--|-----------------------------|---|
| Open & stable : Yes/No If No..... | HR...../min | GCS..... |
| Breathing: RR/min Efforts: Normal/Poor/increased | CFT.....secs. | Pupil size...../min |
| Auscultation: Air entry: Normal/poor/Differential | BP.....mmHg | Pupillary Reactions..... |
| Added sounds: None/Stridor/Wheeze/Crackles | Peripheral pulse: Poor/Good | Motor activity: •Normal & Symmetrical/Asymmetrical/ Posturing/Flacidity/Seizure |
| SpO2 on Room air..... | Central pulse:Poor/Good | Blood Sugar.....mg/dl |
| | Skin temp: Warm/cool | Exposure: Temp..... Colour:Normal/pallor/cyanosis /mottled Any other skin lesions..... |
| | Others | |

Diagnosis



All India Institute of Medical Sciences

Genetics Unit, Department of Pediatrics, Ansari Nagar, New Delhi - 110029
Tel: 011-26594585, 26593558 Fax: 011-26588663, 26588411

SPINAL MUSCULAR ATROPHY GENETIC ANALYSIS REPORT

Name: Rishi Kumar
Age: 17 months
Sex: *Male*

Referred by:
Lab No.: SMA-2283
Date of sample collection: 12-10-18
Type of sample: EDTA blood
Quality of sample: Adequate

Reason for referral: Molecular analysis for SMA

Results:


| Name | Type of Sample | Test Performed | Result |
|-------------|----------------|--|--|
| Rishi Kumar | Blood | PCR/RFLP for exon 7 of <i>SMN</i> gene | Deletion identified in exon 7 of <i>SMN</i> gene |

Interpretation: A homozygous deletion in exon 7 of *SMN* gene was identified. This confirms the diagnosis of SMA in Rishi Kumar.

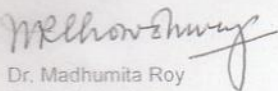
Comments: In the DNA sample extracted from blood, Exon 7 of *SMN* gene was amplified by PCR & RFLP was carried out using *DraI* restriction enzyme.

Notes: Because of their complexity and their potential implications for other family members, all genetic tests should be accompanied by genetic counselling. Although all precautions are taken during molecular genetic tests, the currently available data indicates that the technical error rate for all types of molecular DNA analysis is approximately 2-3%. It is important that the clinicians or individuals requesting molecular genetic diagnostic tests are aware of these data before acting upon these results. The results assume that all the patient information provided is correct.


TEST DONE BY:


Dr. Amita
Scientist II
Division of Genetics
Department of Pediatrics

CHECKED BY:


Dr. Madhumita Roy
Senior Scientist
Division of Genetics
Department of Pediatrics

REVIEWED AND AUTHORISED BY:


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Professor of Pediatrics and officer-in-charge, Division of Genetics
Department of Pediatrics

Dr. Neerja Gupta
Assistant Professor,
Division of Genetics
Department of Pediatrics