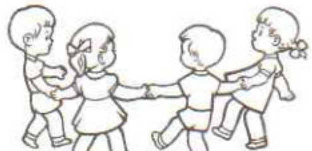
Dr Prabhas Prasun Giri

MBBS (Cal), (Hons), MD(Ped)
MRCPCH(UK), FRCPCH(UK)
Consultant Child Specialist
Reg.No.63526(WBMC)
Mobile : 9051958420 (Only for Emergency)
For Vaccination : 6290342147
Web : www.prabhaspediatrics.com
Online consultation through DOCON App



ব্যানার্জী মেডিক্যাল হল

ঘাসিয়াড়া মোড়, সোনারপুর
প্রতি মঙ্গলবার সকাল ৮টা থেকে ৯-৩০ টা
প্রতি বৃহস্পতিবার সকাল ৮ টা থেকে ৯-৩০ টা
প্রতি শনিবার বিকেল ৪ টা থেকে ৫-৩০ টা
নাম লেখানোর জন্য :
৭৯৮০০৫১৯৫০ / ৯৮৩১২৮৩১৬৭

Name : B/O Haimanti Das Wt. Sex : F Age : 4 m Date : 3/9/24

F/U c/o Bronchiolitis.
Admitted 19/8 → 22/8
Dysphagia?
min. cough
Hypotonic baby
? SMA.

℞

- Calcium vitamin D_3 + A to Z
- SMN gene deletion duplication array.
- Blood for TSH.

(signature)

Chambers

SURAKSHA DIAGNOSTIC, BALLYGUNGE
1st Floor Sunny Tower, 43, Asutosh Chowdhury Avenue, Ballygunge, Kolkata - 700019, Near Birla Mandir
Mon to Fri : 4.00 pm to 5.30 pm
Sat : 1.00 pm to 2.00 pm
2nd & 4th Sunday 12 pm
Appointment : 7595055023

SURAKSHA DIAGNOSTIC KALIKAPUR
Prince Anwar Shah Road Connector, (Beside Avishikta Apartment). Mon-Wed-Fri 8-9 am
Tues & Thurs Morning 10.30 am
For Appointment : 7579640442

EEDF(Sri Aurobinda Seva Kendra)
Jodhpur Park, Kolkata - 700 068
Saturday : 8am - 9 am
for Appointment
Call : 7439744792, 033 40171573, 033 40171520

Radiance Medical Services
13, Sarat Ghosh Garden Road, Dhakuria.Kolkata 700 031
(Near Dhakuria Railway Station)
Monday to Saturday 7pm to 8pm
Appointment & Enquiry
74397 44792 /08062280999

MedGenome Labs Limited.
Sy No 94/1C and 94/2, Tower-1,
Veerasandra Village, Attibele Hobli,
Electronic City, Phase-1,
Bangalore, Karnataka, 560100.
Tel: +91 (0)80 67154932 / 933

MEDGENOME

DNA TEST REPORT – MEDGENOME LABORATORIES

Full Name / Ref No:	B/O Haimanti Das (24-22279)	Order ID/Sample ID:	1045348/8695603
Date of Birth / Age:	4 Months	Gender:	Female
Parental Sample ID:	NA	Sample Type:	Peripheral Blood in EDTA
Referring Clinician:	Dr. P.P Giri, Institute Of Child Health, Kolkata	Date of Sample Collection:	6th September 2024
		Date of Sample Receipt:	7th September 2024
		Date of Order Booked:	8th September 2024
		Date of Report:	23rd September 2024
Test Requested:	**Spinal Muscular Atrophy (*SMN1/SMN2*) deletion/duplication analysis [MGM143]**		

CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

The patient presented with clinical indications of acute bronchiolitis and is thus being evaluated for pathogenic deletions and duplications in exons 7 and 8 of *SMN1* and *SMN2* genes.

RESULTS*

PATHOGENIC VARIANT CAUSATIVE OF THE SUSPECTED PHENOTYPE WAS IDENTIFIED

Sl. No.	Gene Exons†	Deletions /Duplications	MLPA probe ratio (Dosage quotient)#	Copy number	Disease (OMIM)	Inheritance	Classification
1	*SMN1* (Exon 7)	Homozygous deletion	Exon 7 (0.00)	0	Spinal muscular atrophy	Autosomal recessive	Pathogenic
2	*SMN1* (Exon 8)		Exon 8 (0.00)	0			
3	*SMN2* (Exon 7)	-	Exon 7 (1.01)	2	-	-	-
4	*SMN2* (Exon 8)		Exon 8 (0.94)	2			

CLINICAL CORRELATION AND VARIANT INTERPRETATION

Homozygous deletion of exons 7 and 8 in the *SMN1* gene was detected within the detection limits of MLPA, in the subject (Fig.1). No deletions or duplications were detected in the exons 7 and 8 of the *SMN2* gene. The subject has gene copy number ratio of *SMN1*:*SMN2* of 0:2. Functional absence of *SMN1* gene due to homozygous deletions is reported to be pathogenic in 95% of SMA cases [1]. Hence, **this deletion is pathogenic and must be carefully correlated with clinical symptoms.**

RECOMMENDATIONS

Genetic counselling is advised.

Screening of parents is recommended to determine their carrier status.

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BACKGROUND

Spinal muscular atrophy (SMA) is characterized by degeneration of lower motor neurons in the spinal cord, causing progressive paralysis of the limbs and trunk, followed by muscle atrophy. SMA is one of the most frequent autosomal recessive diseases, with a carrier frequency of 1 in 38 and is the most common genetic cause of childhood mortality [4]. The phenotype is extremely variable, and patients are classified as SMA type I to III based on age at onset and clinical course. There are two (highly similar) genes playing a pivotal role in SMA: *SMN1* and *SMN2*. These two genes can only be distinguished by single nucleotide differences in exon 7 and 8. *SMN2* is much less efficient in making the SMN protein; therefore, it is the *SMN1* gene which is the determinant factor in SMA. Of these, greater than 96% are homozygous for the deletion of exons 7 and 8 of this gene. Genetic analysis for this deletion provides an efficient diagnosis for this disorder.

TEST METHODOLOGY

Copy number changes in exons 7 and 8 of the *SMN1* & *SMN2* genes were identified by hybridizing with MLPA (Multiplex Ligation-dependent Probe Amplification) probes. Each MLPA probe consists of two hemi-probes that bind to adjacent sites on the target sequence. Upon ligation and subsequent PCR amplification, each distinct MLPA probe (specific to distinct target regions) generates an amplicon with a unique length which is separated and quantified by capillary electrophoresis. Heterozygous deletions within target sequences will prevent efficient probe binding and give a 35-50% reduced relative peak area of the amplification product specific to that probe set. Copy number differences of various exons between test and control DNA samples can be detected by analyzing the MLPA peak patterns.

***Genetic test results are reported based on the recommendations of American College of Medical Genetics (Richards CS et al., Genet Med, 2015), as described below:**

Variant	A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).
Pathogenic	A disease-causing variation in a gene which can explain the patients' 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.
Benign	A variant which is known not to be responsible for disease has been detected. Generally, no further action is warranted on such variants when detected.
Likely Benign	A variant is not expected to have a major effect on disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion.
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.

ǂ the exon numbering is based on the *SMN1* mRNA reference sequence NM_000344.3 and *SMN2* mRNA reference sequence NM_017411.3 nomenclature respectively in the NCBI GenBank database.

MLPA ratios (dosage quotient) of below 0.7 or above 1.3 are indicative of a deletion (copy number change from two to one) or duplication (copy number change from two to three), respectively. A dosage quotient of 0.0 indicates a homozygous deletion, 0.35 to 0.65 indicates heterozygous deletion, 1.35 to 1.55 indicates heterozygous duplication and 1.7 to 2.2 indicates homozygous duplication. A MLPA ratio (dosage quotient) between 0.80 to 1.20 indicates a normal copy number status

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MEDGENOME

DISCLAIMER

- MLPA cannot detect any changes that lie outside the target sequence of the probes and will not detect most inversions or translocations. Even when MLPA did not detect any aberrations, the possibility remains that biological changes in that gene or chromosomal region do exist but remain undetected.
- The MLPA test will not detect the point mutations in the *SMN1* and *SMN2* genes.
- A point mutation or polymorphism in the sequence detected by a probe, which results in reduced probe binding efficiency, can also cause a reduction in relative peak area. Therefore, single exon deletions detected by MLPA should always be confirmed by other methods like multiplex PCR or sequencing.

Jeevana Prahar sha.A	Manjunath V	Dr. Rammurthy Anjanappa	Dr. Pragya Gupta MBBS, MD Path. PDF Molecular Geneticist (TMCK)
Senior Genome Analyst	Senior Manager (Lab Operations)	Senior Scientist (Lab Operations)	Senior Molecular Pathologist & Clinical Head

REFERENCES

1. Yoon S, Lee CH, Lee KA. Determination of *SMN1* and *SMN2* copy numbers in a Korean population using multiplex ligation-dependent probe amplification. Korean J Lab Med. 2010; 30(1):93-6.
2. Ogino S, Wilson RB. Spinal muscular atrophy: molecular genetics and diagnostics. Expert Rev Mol Diagn. 2004;4(1):15-29.
3. Prior TW et al, Homozygous *SMN1* deletions in unaffected family members and modification of the phenotype by *SMN2*. Am J Med Genet A. 2004.
4. Nilay M, Moirangthem A, Saxena D, Mandal K, Phadke SR. Carrier frequency of SMN1-related spinal muscular atrophy in north Indian population: The need for population-based screening program. Am J Med Genet A. 2021 Jan;185(1):274-277. doi: 10.1002/ajmg.a.61918. Epub 2020 Oct 14. PMID: 33051992.

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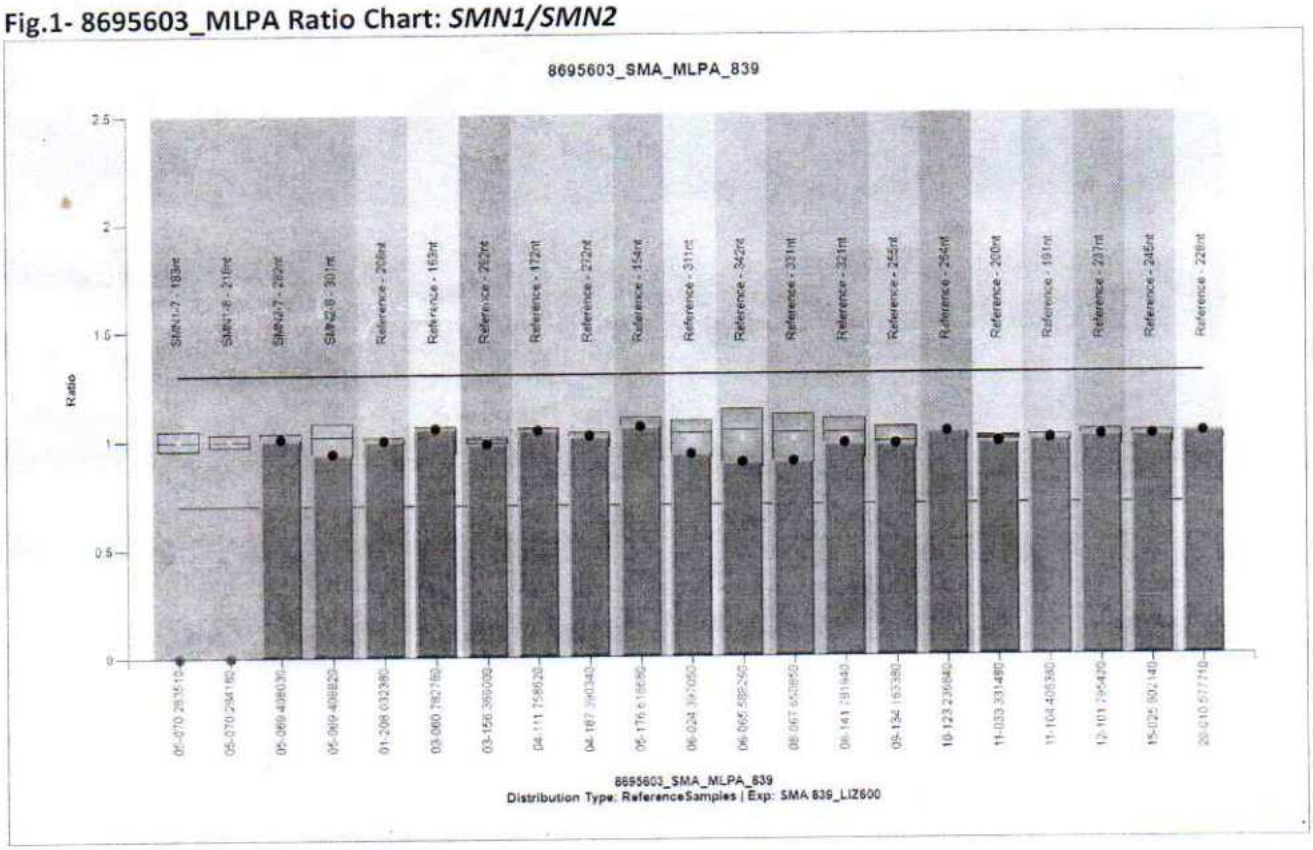
MEDGENOME

APPENDIX-1

SMN1/SMN2 -MLPA Result Figure

Fig.1- 8695603_MLPA Ratio Chart: *SMN1/SMN2*



.....End of Report.....

CAP Test ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

Institute of Child Health

11, Dr. Biresh Guha Street, Kolkata - 700 017
Phone : 2290-5686, Fax : (033) 2289-3242
Email : ichcal@yahoo.com, Web : www.ichcalcutta.org
Online OPD Booking : www.ichkolkata.org

SPECIAL BIOCHEMISTRY

Name	B/O HAIMANTI DAS	Registration No.	2400022279
Age	4-Mn.20-Days	Admission No.	0
Sex	F	Bed No.	0
Referred By Dr.	PRABHAS PRASUN GIRI	Contact No.	8777062393
Reporting Date	6/9/2024	Collection Date	6/9/2024

Sample : BLOOD

Test Parameters	Result	Unit	Reference Interval
Serum Thyroid Stimulating Hormone (TSH) (Enhanced Chemiluminescence Immunometric Immunoassay))	2.49	µIU/ml	0 - 23mns:0.5885-6.880, 2 - 12yrs:0.7291-4.402, 13 - 21yrs:0.4557-4.160, >= 22yrs:0.4001-4.049(M & Non Pregnant Fe), Pregnant 1st trimester:0.1298-3.120, 2nd trimester: 0.2749-2.652, 3rd trimester:0.3127-2.947

---End of Report---

Prof. S. Basu
M. Sc., Ph. D.
Professor & HOD
Dept. of Biochemistry,
Deputy Director, ICH

Dr. A. Saha
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MD (Biochemistry)
Consultant Biochemistry

Dr. J. (Raman) Chowdhury
MBBS(CAL), DTM&H(CAL),
DNB (Pathology)
HOD&Director,Dept.of Pathology

Dr. K. S. Ray
MBBS(CAL),
MD (Pathology)
Consultant Pathologist

Dr. K. Sarkar
MBBS(CAL),
MD (Pathology)
Consultant Pathologist

Dr. A. Chatterjee
MBBS(CAL),
MD (Microbiology), CPHIC
Consultant Microbiologist

Instrument Name: VITROS 5600

Report Prepared By: C003

Dr Prabhas Prasun Giri

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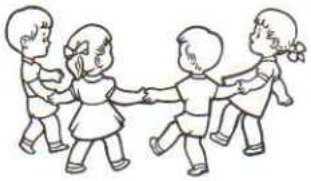

ব্যানার্জী মেডিক্যাল হল
ঘাসিয়াড়া মোড়, সোনারপুর

প্রতি মঙ্গলবার সকাল ৮টা থেকে ৯-৩০ টা
প্রতি বৃহস্পতিবার সকাল ৮ টা থেকে ৯-৩০ টা
প্রতি শনিবার বিকেল ৪ টা থেকে ৫-৩০ টা

নাম লেখানোর জন্য :
৭৯৮০০৫১৯৫০ / ৯৮৩১২৮৩১৬৭

Name : B/O Hainanti Das Wt. 6kg Sex : F Age : 5m 11d Date : 28/9/24

Hypotonic Body.

SMN gave nutrition boost.

currently no chest infection.

Rx

→ Attend SMA clinic of Peerless Hospital

↓ Dr. Sanjukta Sir.

Chambers

SURAKSHA DIAGNOSTIC, BALLYGUNGE
1st Floor Sunny Tower, 43, Asutosh Chowdhury Avenue, Ballygunge, Kolkata - 700019,Near Birla Mandir
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Sat : 1.00 pm to 2.00 pm
2nd & 4th Sunday 12 pm
Appointment : 7595055023

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EEDF(Sri Aurobinda Seva Kendra)
Jodhpur Park, Kolkata - 700 068
Saturday : 8am - 9 am

for Appointment
Call : 7439744792, 033 40171573, 033 40171520

Radiance Medical Services
13, Sarat Ghosh Garden Road, Dhakuria.Kolkata 700 031
(Near Dhakuria Railway Station)
Monday to Saturday 7pm to 8pm
Appointment & Enquiry
74397 44792 /08062280999

Dr. Apurba Ghosh
M.B.B.S. , M.D. (PAED), DCH (CAL),
F.R.C.P. (Ireland), M.R.C.P. (U.K), F.R.C.P.C.H. (U.K.), FIAP (INDIA)
Registration No. 40061 of WMC
E-mail : apurbaghosh@yahoo.com
Mobile : 98300 52887 Not for appointment (No phone calls after 9:30 p.m)

8777062393 M

Residence :
7, Central Park, Jadavpur
Kolkata - 700 032
Tel : 9830052887

29/9/24

B/O Haimanti Das (F) 5m/13d/6 Kg

D·O·B - 16/4/24

B·wt - 3·280kg

Ht - 41·3 Cms

C/O

- cough & cold -

hypotone baby

SMA

C-5

SpO2 - 97%

HR - 179

- Azithral (100) Susp
3 ml once daily x 6d
- Nasoclear Nasal drop
1 drop in each nostril
- cont. vit D_3 drop
- Physiotherapy

HPLC DONE :

HPLC NOT DONE :

Peerless Hospitex Hospital And Research Center Limited

360, Panchasayar, Kolkata - 700 094, Phone : 033-4011 1222, 2462 2394/2462/0071-73

E-mail : ph.enquiry@peerlesshospital.com ● Website : www.peerlesshospital.com

CIN - U85110WB1989PLC046938

OUT PATIENT CLINIC

UHID: MR/24/047333 Visit ID: OP/24/161196 Invoice No IO/24/619376 Date: 30/09/2024 Sex: Female

Name: Baby of Haimanti Das Hridika Das Age: 5 Months 14 Days

Category: GENERAL

Doctor: Dr. Sanjukta De **Visit Count : 1 (No Booking) Queue:11**

Address: GHASIARA MADHYA PARA SONARPUR SONARPUR 700150 Mob-1: 9088941929 Mob-2:

Chief Complaints / History :

Complaints of Dr. P. P. Giri

Homozygous deletion.

Drug Allergy : (Give Details)

Current Medication :

Immunisation Status

Govt schedule

Examination :	Pulse	BP	Weight (in Kg)	Height (in cm)	BMI	Nutritional Screening
			6 kg.	65.5 cm		Normal ☐
						At Risk ☐
						Malnourished ☐
						Any Special Need ☐

Investigations Requested / Recently Done :

Chest Clear

Alert, Interactive

Inflevactra 0.5 OR Varipex 4 - 0.1

Provisional Diagnosis / Impression :

SMA – Type I

Plan of Care / Treatment :

Adv

1. Physiotherapy – Kijal.
2. Multidisciplinary care.
3. Cure SMA – Contact

Follow Up :

Mousmita Ghosh;

9432888613

Dr. Sanjukta De

MBBS, DCH(Cal), DNB(Paediatrics), DCH(London), MRCPCH(UK), FRCPCH(UK), Dip Allergy(UK)

Senior Consultant Clinical Director (Pediatrician & Neonatologist)

Registration No. 52551 WBMC

Second OPD Visit within 15 days for showing of investigation results for the same Consultant for the same problem will not be chargeable

For Home collection of pathological samples please contact 6292235660.

For Doctor Appointment Please Contact 033-40333333 /24622462 from 8:00AM-7:00PM

④ Lepicol D_3 60ml
0.5ml once a day × 6 months

⑤ Calciol D syrup
2.5ml once a day × 2 months

⑥ Coulcii A-2

⑦ Carnisure syrup
2ml once a day × 6 months

SMA

Risdeplain — 6 tab/ oral both

Nusinersin — IT

Gene therapy — IV

SMA

Type I

Type II

Type III

सत्यमेव जयते

GOVERNMENT OF WEST BENGAL
DEPARTMENT OF HEALTH & FAMILY WELFARE
IPGMER & SSKMH-CENTRE of EXCELLENCE
A.J.C Bose Road Kolkata-20

OPD Patient Card
NEONATOLOGY MEDICINE 41
Paid Rupees : 2

SSKM/RG2401881208

Name : HRIDIKA DAS	**Age :** 0 Yrs. 5 Months 0 Days	**Gender :** Female
Reg. No. : SSKM/RG2401881208	**Card No. :** SSKM/OR2401609101	**Reg. Date :** 04-10-2024
Visit No. : 1	**Visit Date :** 04-10-2024 12:15PM	**Day :** Friday
Token No. : 6378	**Department :** NEONATOLOGY MEDICINE	**Room No. :** 0
Doctor : Dr.S Mukherjee/ Dr.R Mukherjee/ Dr.A. Saha/ Dr. U. Mondal/ Dr.N.Sarkar		
Health Id :		**Health Id Number :**

Visit No : 2	Visit No : 3	Visit No : 4
Visit Date : Tm.	Visit Date : Tm.	Visit Date : Tm.
Department :	Department :	Department :
Doctor :	Doctor :	Doctor :
Token No :	Token No :	Token No :

Clinical Notes	Advice
5 months 18 days.	
Wt- 6.1 kg Progressive weakness of lower limbs, upper limbs with respiratory distress.	Home based Compremensive Early Intervention Hearing & Vision Assessment. To be enrolled under NPRD. Immunisation as per schedule.
Surgical Term, AFD, girl, born by vaginal delivery Birth Wt- 3250gram.	
Antenatal & Perinatal history - uneventful.	
Clinical exam- OFC - Unremarkable except mild respiratory distress & atypical posture.	
Chest- b/L air entry good & equal.	
CNS Neuro exam- Cranial nv- normal	
Low tone - lower limb > upper limb, Neck & neck, gen LL > UL.	
DTR - Knee, ankle, biceps & brachioradialis Could not be elicited.	

User Name : Bimal Paramanick (Gynae)

10/04/2024 12:21 PM

1 of 1

Genetic test - Homozygous deletion in SMN1 gene exon 7 & 8.
SMN2 gen 2 Copy number present.

Reporting time-4p.m.

National Neurosciences Centre, Calcutta

Peerless Hospital Campus (2nd Floor) 360, Panchasayar, Kolkata - 700 094
Ph. (033) 2432 0777 / 0999 / 0748
E-mail: nnccalcutta@gmail.com / marketing.nnc@live.com
Web: www.nnccalcutta.in (West Bengal Society Registration No. S/97602)

SPECIALITY OPD (Dr. Sanjukta De Consultant Pediatrician)

UHID : NUHID/129612	Visit ID :OPD/24-25/10268	Invoice No : OPD/2024-25/10265	Date : 04-10-2024
Patient Name : Baby. HRIDIKA DAS			Age : 5 Mnth19 Days
Address : SONARPUR, GHASIARA, , KOLKATA - 700150, WEST BENGAL, INDIA			Sex : Female
Category : GENERAL			Mobile:: 9088941929
Doctor : Dr. ASHIS DAS			

Pulse BP Weight (in Kg)

Diagnosed case of SMA - Type-1

Prospective case of Disease modifying Rx

Best option would be Zolgensma (Single I/V Rx) c Risdiplam
Next option Intrathecal Spinraza (Repeated inj.)
3rd / Oral Risdiplam
Lifelong treatment

- The child is alert
- Hypotonic
- ↓↓ Spont. movements
- Absent DTR

- To Continue Supportive Rx

Cure SMA

Dr. ASHIS DAS
MD. (Med), DM (Neurology)
Consultant Neurologist
National Neurosciences Centre Calcutta
Reg. No.- 50837

4/10/2024

Dr. ASHIS DAS
Consultant Neurologist
MD (Med), DM (Neurology)