# All-India Institute of Medical Sciences Ansari Nagar, New Delhi-29 (RESEARCH SECTION)

Ref. No. 24/Prop./Ped. /NG/18-19/RS

Dated: 08.10.2018

### Subject: Purchase of Whole Exome sequencing for the Deptt. of Peadiatics, AIIMS, New Delhi-29 on proprietary basis- <u>Inviting comments thereon.</u>

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The request has been received from Dr.Neerja Gupta, Assistant Professor, Deptt. of Pediatrics, AIIMS to purchase the subject item from M/s MedGenome Labs Pvt. Ltd. on proprietary basis. The proposal submitted by M/s MedGenome Labs Pvt. Ltd. and Performa Invoice and Departmental PAC certifications are attached.

The above documents are being uploaded for open information to submit objections, commets, if any, from any manufacturer regarding proprietary nature of the equipment/item within issue of 15 days giving reference **No. 24/Prop./Ped./NG/18-19/RS**. The comments should be received by office of Stores Officer (RS), Research Section at AIIMS on or before **24/12/2018 upto <u>12:00 p.m.</u>**, failing which it will be presumed that any other vendor is having no comment to offer and case will be decided on merits.

**STORES OFFICER (RS)** 

<u>Encl</u>: Related documents enclosed. 1. PAC Certificate enclosed. 2. Performa Invoice

### 4 ALL INDIA INSTITUTE OF MEDICAL SCIENCES ANSARI NAGAR, NEW DELHI-110029 PROPRIETORY/SPECIFIC BRAND GOODS CERTIFICATE 1. Item/Type/Model No. required Whole Exome Sequencing alongwith specification. 2. Is the item a spare parts attachment No or accessory for an existing equipment. 3. Name of the manufacturers/supplier M/s MedGenome Labs Pvt. Ltd. of the item proposed by the Indentor. Are they sole providers/sold distributors of the item. Yes for our specifications. 5. Is there any other item with similar/ Yes, similar items are available with other laboratories equivalent specification available in as well. However, after trying out all available options, the market to meet the job requirement other companies, the result from M/s MedGenome Labs envisaged. If the answer is yes, why the Pvt. Ltd. were found to be most satisfactory. same can't be procured. Demanding The functional advantages/cost effectiveness officer should bring out comparative of the recommended items areof the recommended terms are-The Technology Platform: Illumina is the sole manufacture of HiSeqX and HiSeq 4000. MedGenome is the sole service provider of sequencing on HiSeqX and HiSeq 4000. There is no equivalence specification for HiSeqX and HiSeq 4000. functional advantages/cost effectiveness of the recommended item from these offered by other. HiSeqX and HiSeq 4000. Quality Certification: The lab is CAP/NABL certified. participating the proficiency testing as a part of the quality program. The exome are CAP certified while the NABL certification have whole exome as a scope of

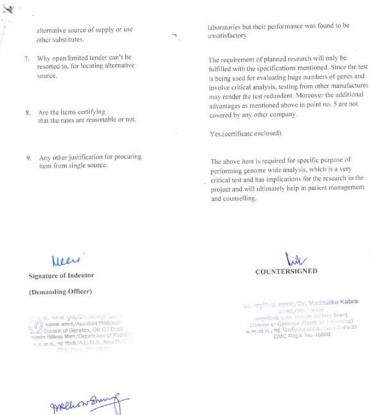
Lab Location: Bangalore based. Test will be performed in India. Lab has extensive experience in reporting minor allele frequency from Indian population.

the test.

Add on confirmation: In addition to Exome sequencing, company will offer CNV analysis on the Exome data performed for the candidate genes. Sanger confirmation for ONE VARIANT if identified will be part of the project. Minor allele frequency from Indian population for the variant identified would be given along with the data

6. What were the efforts made to locate

We have experience of getting the test done in other



Dr. Madhumita Roy Chewonury Senior Scientist Division of Genetics, Department of Paciatries All India Institute of Madical Sciences New Dethi - 110020

## ALL INDIA INSTITUTE OF MEDICAL SCIENCES ANSARI NAGAR, NEW DELHI-110029

### Specifications

- DNA isolation from blood Exome capture (Sure Select V-5 Capture Kit.)
- Library preperation,
- Cluster generation,
  Cluster generation,
  NGS Sequencing using 2x 150bp reads @ 100x (10GB) on Illumina Hiseq or XTen platforms.
  Bioinformatics analysis with illumina HiseqXten
  Phred quality score of Q30, for 85% of data.
  Output should be inform of raw reads (fastq format).

- The charges should be inclusive of Sanger validation for 1 variant.
   An Indian lab that has extensive experience in Exome processing and reporting minor ellele frequency from Indian population
- Test should be performed in India. Samples deportation and testing out of Indian Territory are not authorized.
- The performing lab should have NABL & CAP certification, as part of proficiency programme.
  The performing Lab should have extensive experience. Essentially over ten thousand exomes processing and in reporting minor allele frequency from Indian population.
- . The service provider should use ultra-throughput sequencing system, HiSeqX (part of X ten) for higher data generation and to bring cost effectiveness.

plus.

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melinom

Dr. Madhumita Roy Chewdhury Senior Scientisi Division of Genetics. Department of Pediatrics all India Institute of Medical Sciences

stered Office: edGenome Labs Ltd. rd Floor, Narayana Nethralaya Building, Narayana Health City, # 258/A, Bommasandra, Hosur Road, Bangalore - 560 099, India.	- MEDGENOME
T: +91(0) 80 6715 4901 www.medgenome.com July 10, 2018	illumina
Proprietar	y Letter
To Whom It M	ay Concern
We, Illumina Singapore Pte. Ltd., a subsidiary of Il principal place of business at 11 Biopolis Way # 09-05	

principal practice of obtaines at 11 objoins way in 0-0-0 means imaginary to 2007, which is established with reputable manufacturers of Illumina Sequencing & Arrays Systems and the consumables for the Sequencing and Arrays Systems. Hereby, confirm that the following products are solely manufactured by Illumina, Inc., U.S.A. and /or Illumina Singapore Pte. Ltd. and is Proprietary technology of Illumina, Inc., U.S.A.

This is also to certify that M/s Medgenome Pvt. Ltd., is currently the only commercial service provider in India that has HiSeq 4000 & HiSeq X Ten in house.

CAT NO	DESCRIPTION
SY-401-4001	HiSeq <sup>®</sup> 4000 Sequencing System The Illumina HiSeq 4000 Sequencing System is a dual flow cell sequencing instrument. System includes workstation computer, touch screen monitor, HiSeq Control Software, installation kits and standards, installation and training, and 12 months warranty (including parts and labor).
SY-412-1001	HiSeq X <sup>w</sup> Sequencing System (as part of HiSeq X Ten) HiSeq X is an ultra-high throughput sequencing system, which is sold in quantities of at least 10 units (HiSeq X Ten = collection of 10 HiSeq X). The system is designed for whole genome sequencing application only.

Yours faithfully,

Name: Tan Kah Ling, Mavis Title: Senior Director, Finance, Asia Pacific For and behalf of: Illumina Singapore Pte. Ltd. heen

Illumina Singapore \* 11 Biopo

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Way: \* 00063/Hass: \* File: \* Tel +85.6773.0186 \* Fax +65.6774.0386 \* www.illumina.com





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	oratory	Madaanoma Lah	Private Limited S	iDF 17, 1st Floor, C-Block C	SFZ
Lau	oratory	Cochin, Kerala			
Acc	creditation Standar	d ISO 15189: 2012			
Cer	tificate Number	MC-2497	1	Page 1 of 7	
Val	idity	18.12.2017 to 17.	12.2019	Last Amended on	
SI.	Product / Material of Test	Specific Test Performed	Test Method	Range of Testing / Limits of Detection	CV%
			GENETICS		
1.	Blood/	JAK2 gene analysis -	PCR and DNA	Qualitative within limits	NA
1.00	Genomic DNA	2 exons (12 & 14)	sequencing	of Sanger sequencing	
2.	Blood/ FFPE/ Genomic DNA	PDGFRA gene analysis - 3 exons (12, 14 & 18)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
3.	Blood/ Genomic DNA	TP53 gene analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
4.	Blood/ Genomic DNA	Congenital Adrenal Hyperplasia CYP21A2 (21-0H) gene analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
5.	Blood/	VHL gene analysis	PCR and DNA	Qualitative within limits	NA
6.	Genomic DNA Blood/ Genomic DNA	Cystic fibrosis (CFTR) del508	sequencing PCR and DNA sequencing	of Sanger sequencing Qualitative within limits of Sanger sequencing	NA
7.	* FFPE / Genomic	mutation analysis K/T gene analysis - 4	PCR and DNA	Qualitative within limits	NA
1.	DNA	exons (9, 11, 13 & 17)	sequencing	of Sanger sequencing	
8.	Blood/RNA	Imatinib resistance (ABL kinase) gene analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
9.	Blood/ Genomic DNA	Factor V Leiden mutation analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
10	Blood/	(exon 10) MTHFR gene	PCR and DNA	Qualitative within limits	NA
10.	Genomic DNA	analysis - 2 exons (5 & 8)	sequencing	of Sanger sequencing	
11.	Blood/ Genomic DNA	Spinal Muscular Atrophy (SMN1)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA

ilad	MRA S	(A Constituent E	Accreditation and Calibration Board of Quality Court FACCRED	n Laboratories		
Lab	oratory	Medgenome Lab Cochin, Kerala	s Private Limited, S	DF 17, 1st Floor, C-Block C	SEZ,	
Acc	reditation Standar	d ISO 15189: 2012				
Cer	tificate Number	MC-2497	1	Page 2 of 7		
		18.12.2017 to 17.		Last Amended on	1	
vali	idity	10.12.2017 (0 17.	14.4.10			
SI.	Product / Material of Test	Specific Test Performed	Test Method	Range of Testing / Limits of Detection	CV%	
12.	FFPE / Genomic	IDH1 gene analysis	PCR and DNA	Qualitative within limits	NA	
13.	DNA Blood/	Alpha Thalassemia	sequencing PCR and DNA	of Sanger sequencing Qualitative within limits	NA	
13.	Genomic DNA	gene analysis (HBA1 & HBA2)	sequencing	of Sanger sequencing		
14.	Blood/	Beta Thalassemia	PCR and DNA	Qualitative within limits of Sanger sequencing	NA	
15.	Genomic DNA Blood/	(HBB) gene analysis TSC1 & TSC2 gene	PCR and DNA	Qualitative within limits	NA	
15.	Genomic DNA	analysis	sequencing	of Sanger sequencing		
16.	Blood/ Genomic DNA	NOTCH3 (CADASIL) gene analysis -2	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA	
17.	Blood/	exons (2 & 3) BRCA1 and BRCA2	PCR and DNA	Qualitative within limits	NA	
	Genomic DNA	gene analysis	sequencing	of Sanger sequencing		
18.	Blood/	DPYD mutation	PCR and DNA	Qualitative within limits of Sanger sequencing	NA	
19.	Genomic DNA Blood/ Genomic DNA	analysis (intron 14) Connexin- 26(GJB2)gene	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA	
20.	Blood/ Genomic	analysis (exon 2) MPN reflex panel	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA	
21.	Blood/ Genomic DNA	Sickle cell anemia ( <i>HBB</i> ) gene analysis (Exon 1)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA	
22.	Blood/ FFPE / Genomic DNA	RET oncogene - 8 exons(5.8,10,11,13,1 4,15 & 16)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA	
23.	Blood/ Genomic DNA	4,15 & 16) Cardiomyopathy predisposition - MYBPC3 (25bp deletion)	PCR and DNA Qualitative within limits N sequencing of Sanger sequencing			
24.	Blood/ Genomic DNA	Clopidogrel dosage:CYP2C19*2 and CYP2C19*3	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA	

lac	MRA S	(A Constituent E	Accreditation ad Calibration to and of Quality Co FACCREE	uncil of India)	
	oratory	Cochin, Kerala		SDF 17, 1st Floor, C-Block C	SEZ,
Acci	reditation Standar	d ISO 15189: 2012			
Cert	ficate Number	MC-2497		Page 2 of 7	
	414	18.12.2017 to 17.	12 2010	Last Amended on	1
Valie	dity	18.12.2017 to 17.	12.2019	Last Amended on	
SI.	Product / Material of Test	Specific Test Performed	Test Method	Range of Testing / Limits of Detection	CV%
2.	FFPE / Genomic DNA	IDH1 gene analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
3.	Blood/ Genomic DNA	Alpha Thalassemia gene analysis (HBA1 & HBA2)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
4.	Blood/ Genomic DNA	(HBA) & HDA2) Beta Thalassemia (HBB) gene analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
15.	Blood/ Genomic DNA	TSC1 & TSC2 gene analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
16.	Blood/ Genomic DNA	NOTCH3 (CADASIL) gene analysis -2 exons (2 & 3)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
17.	Blood/ Genomic DNA	BRCA1 and BRCA2 gene analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
18.	Blood/	DPYD mutation	PCR and DNA	Qualitative within limits of Sanger sequencing	NA
19.	Genomic DNA Blood/ Genomic DNA	analysis (intron 14) Connexin- 26(GJB2)gene	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
20.	Blood/ Genomic	analysis (exon 2) MPN reflex panel	PCR and DNA	Qualitative within limits of Sanger sequencing	NA
21.	DNA Blood/ Genomic DNA	Sickle cell anemia (HBB) gene analysis (Exon 1)	sequencing PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
22.	Blood/ FFPE / Genomic DNA	RET oncogene - 8 exons(5.8,10,11,13,1 4,15 & 16)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
23.	Blood/ Genomic DNA	Cardiomyopathy predisposition - MYBPC3 (25bp deletion)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
24.	Blood/ Genomic DNA	Clopidogrel dosage:CYP2C19*2 and CYP2C19*3	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA

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Lab	oratory	Medgenome Lab Cochin, Kerala	s Private Limited, Si	DF 17, 1st Floor, C-Block C	SEZ,
Acc	reditation Standar	d ISO 15189: 2012			
	tificate Number	MC-2497	F	age 2 of 7	
				ast Amended on	
Vali	dity	18.12.2017 to 17	12.2019 L	ast Amended on	
SI.	Product / Material of Test	Specific Test Performed	Test Method	Range of Testing / Limits of Detection	CV%
12.	FFPE / Genomic	IDH1 gene analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
13.	Blood/ Genomic DNA	Alpha Thalassemia gene analysis	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
14.	Blood/	(HBA1 & HBA2) Beta Thalassemia	PCR and DNA	Qualitative within limits	NA
15.	Genomic DNA Blood/	(HBB) gene analysis TSC1 & TSC2 gene	sequencing PCR and DNA	of Sanger sequencing Qualitative within limits	NA
	Genomic DNA	analysis	sequencing	of Sanger sequencing Qualitative within limits	NA
16.	Blood/ Genomic DNA	NOTCH3 (CADASIL) gene analysis -2 exons (2 & 3)	PCR and DNA sequencing	of Sanger sequencing	144
17.	Blood/	BRCA1 and BRCA2	PCR and DNA	Qualitative within limits of Sanger sequencing	NA
18.	Genomic DNA Blood/	gene analysis DPYD mutation	sequencing PCR and DNA	Qualitative within limits	NA
10.	Genomic DNA	analysis (intron 14)	sequencing	of Sanger sequencing	
19.	Blood/ Genomic DNA	Connexin- 26(GJB2)gene analysis (exon 2)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
20.	Blood/ Genomic	MPN reflex panel	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
21.	Blood/ Genomic DNA	Sickle cell anemia (HBB) gene analysis (Exon 1)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
22.	Blood/ FFPE / Genomic DNA	RET oncogene - 8 exons(5.8,10,11,13,1 4,15 & 16)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
23.	Blood/ Genomic DNA	4,15 & 16) Cardiomyopathy predisposition - MYBPC3 (25bp deletion)	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA
24.	Blood/ Genomic DNA	Clopidogrel dosage:CYP2C19*2 and CYP2C19*3	PCR and DNA sequencing	Qualitative within limits of Sanger sequencing	NA

IBC=MRA



# National Accreditation Board for Testing and Calibration Laboratories



(A Constituent Board of Quality Council of India)

# SCOPE OF ACCREDITATION

SI.       Product / Material of Test       Specific Test Performed       Test Method       Range of Testing / Limits of Detection         IS.       Blood/ Genomic DNA       Cystic fibrosis (CFTR) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         46.       Blood/ Genomic DNA       Androgen receptor (AR) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         47.       Blood/ Genomic DNA       Haemophilia (F8) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         48.       Blood/ Genomic DNA       Dravet syndrome (SCN1A) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         49.       Blood/ Genomic DNA       CGA1 deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         50.       Blood/ Genomic DNA       GLUT1 deficiency (SLC2A1) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         51.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         52.       Blood/ Genomic DNA       Merkes disease (ATP7A) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         53.       Blood/ Genomic DNA       Merkes disease (ATP7A) deletion/ duplication analysis       MLPA       Qualitati	Acci	reditation Standa	rd ISO 15189: 2012			
SI.       Product / Material of Test       Specific Test Performed       Test Method       Range of Testing / Limits of Detection         45.       Blood/ Genomic DNA       Cystic fibrosis (CFTR) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         46.       Blood/ Genomic DNA       Androgen receptor (AR) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         47.       Blood/ Genomic DNA       Haemophilia (F8) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         48.       Blood/ Genomic DNA       Dravet syndrome (SCN1A) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         49.       Blood/ Genomic DNA       Dravet syndrome (SAA) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         50.       Blood/ Genomic DNA       GLUT1 deficiency (SL 22A1) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         51.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         52.       Blood/ Genomic DNA       Merkes disease (ATP7A) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         53.       Blood/ Genomic DNA       Merkes disease (ATP7A) deletion/ duplication analysis       MLPA	Cert	ificate Number	MC-2497		Page 5 of 7	
Material of TestPerformedLimits of Detection45.Blood/ Genomic DNACystic fibrosis (CFTR) deletion/ duplication analysisMLPAQualitative within limits of MLPA46.Blood/ Genomic DNAAndrogen receptor (AR) deletion/ duplication analysisMLPAQualitative within limits of MLPA47.Blood/ Genomic DNAHaemophilia (F8) deletion/duplication analysisMLPAQualitative within limits of MLPA48.Blood/ Genomic DNADravet syndrome (SCN1A) deletion/duplication analysisMLPAQualitative within limits of MLPA49.Blood/ Genomic DNAPompe disease (GAA) deletion/ duplication analysisMLPAQualitative within limits of MLPA50.Blood/ Genomic DNAGLUT1 deficiency (SLC2A1) deletion/duplication analysisMLPAQualitative within limits of MLPA51.Blood/ Genomic DNAGlycine encephalopathy (GLDC) deletion/ duplication analysisMLPAQualitative within limits of MLPA52.Blood/ Genomic DNAGlycine (ALPA, D2HGDH, MLPA, D2HGDH, MLPA, D2HGDH, MLPAMLPAQualitative within limits of MLPA53.Blood/ Genomic DNAMenkes disease (AT7A) deletion/ duplication analysisMLPAQualitative within limits of MLPA53.Blood/ Genomic DNAMenkes disease (AT7A) deletion/ duplication analysisMLPAQualitative within limits of MLPA	Vali	dity	18.12.2017 to 17	.12.2019	Last Amended on	
Genomic DNA       (CFTR) deletion/ duplication analysis       of MLPA         46.       Blood/ Genomic DNA       Androgen receptor (AR) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         47.       Blood/ Genomic DNA       Haemophilia (F8) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         48.       Blood/ Genomic DNA       Dravet syndrome (SCN1A) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         49.       Blood/ Genomic DNA       Pompe disease (GAA) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         50.       Blood/ Genomic DNA       GLUT1 deficiency (SLC2A1) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         51.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         52.       Blood/ Genomic DNA       Neurometabolic disorder (MLC1, 2HGDH, D2HGDH, MLYCD) deletion duplication analysis       MLPA       Qualitative within limits of MLPA         53.       Blood/ Genomic DNA       Merkes disease (ATPTA) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA	SI.	Material of		Test Method	Range of Testing / Limits of Detection	CV%
Genomic DNA       (AR) deletion/ duplication analysis       of MLPA         47.       Blood/ Genomic DNA       Haemophilia (F8) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         48.       Blood/ Genomic DNA       Dravet syndrome (SCN1A) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         49.       Blood/ Genomic DNA       Pompe disease (GAA) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         50.       Blood/ Genomic DNA       CL22A1) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         51.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         52.       Blood/ Genomic DNA       Clycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         53.       Blood/ Genomic DNA       Menkes disease (Genomic DNA       Menkes disease (Graphic DIA (APT7A) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA	45.		(CFTR) deletion/	MLPA		NA
47.       Blood/ Genomic DNA       Haemophilia (F8) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         48.       Blood/ Genomic DNA       Dravet syndrome (SCN1A) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         49.       Blood/ Genomic DNA       Pompe disease (GAA) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         50.       Blood/ Genomic DNA       GLUT1 deficiency (SLC2A1) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         51.       Blood/ Genomic DNA       GLUT1 deficiency (SLC2A1) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         52.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         53.       Blood/ Genomic DNA       Menkes disease (ATP7A) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA	46.		(AR) deletion/	MLPA		NA
48.       Blood/ Genomic DNA       Dravet syndrome (SCN1A) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         49.       Blood/ Genomic DNA       Pompe disease (GAA) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         50.       Blood/ Genomic DNA       GLUT1 deficiency (SLC2A1) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         51.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         52.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         53.       Blood/ Genomic DNA       Menkes disease (ATP7A) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA	47.		Haemophilia (F8) deletion/duplication	MLPA		NA
49.       Blood/ Genomic DNA       Pompe disease (GAA) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         50.       Blood/ Genomic DNA       GLUT1 deficiency (SLC2A1) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         51.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         52.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         53.       Blood/ Genomic DNA       Menkes disease (ATP7A) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA	48.		Dravet syndrome (SCN1A) deletion/duplication	MLPA		NA
50.       Blood/ Genomic DNA       GLUT1 deficiency (SLC2A1) deletion/duplication analysis       MLPA       Qualitative within limits of MLPA         51.       Blood/ Genomic DNA       Glycine encephalopathy (GLDC) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA         52.       Blood/ Genomic DNA       Neurometabolic disorder (MLC1, L2HGDH, D2HGDH, MLYCD) deletion duplication analysis       MLPA       Qualitative within limits of MLPA         53.       Blood/ Genomic DNA       Menkes disease (ATP7A) deletion/ duplication analysis       MLPA       Qualitative within limits of MLPA	49.		Pompe disease (GAA) deletion/	MLPA		NA
51.     Blood/ Genomic DNA     Glycine encephalopathy (GLDC) deletion/ duplication analysis     MLPA     Qualitative within limits of MLPA       52.     Blood/ Genomic DNA     Neurometabolic disorder (MLC1, L2HGDH, D2HGDH, MLYCD) deletion duplication analysis     MLPA     Qualitative within limits of MLPA       53.     Blood/ Genomic DNA     Menkes disease (ATP7A) deletion/ duplication analysis     MLPA     Qualitative within limits of MLPA	50.		GLUT1 deficiency (SLC2A1) deletion/duplication	MLPA		NA
52.     Blood/ Genomic DNA     Neurometabolic disorder (MLC1, L2HGDH, D2HGDH, MLYCD) deletion duplication analysis     MLPA     Qualitative within limits of MLPA       53.     Blood/ Genomic DNA     Menkes disease (ATP7A) deletion/ duplication analysis     MLPA     Qualitative within limits of MLPA	51.		Glycine encephalopathy (GLDC) deletion/	MLPA		NA
53. Blood/ Menkes disease MLPA Qualitative within limits Genomic DNA (ATP7A) deletion/ duplication analysis	52.		Neurometabolic disorder (MLC1, L2HGDH, D2HGDH, MLYCD) deletion	MLPA	of MLPA	NA
	53.		Menkes disease (ATP7A) deletion/	MLPA		NA
54. Blood/ TSC1 and TSC2 MLPA Qualitative within limits Genomic DNA deletion/duplication analysis	54.	Blood/ Genomic DNA		MLPA	Qualitative within limits of MLPA	NA

ilad	SIMRA S	(A Constituent	Accreditation nd Calibration Board of Quality Con DF ACCRED	uncil of India)	
Lab	oratory	Medgenome Lab Cochin, Kerala	es Private Limited,	SDF 17, 1st Floor, C-Block C	SEZ,
Acc	reditation Standa	rd ISO 15189: 2012			
Cer	tificate Number	MC-2497		Page 6 of 7	
Vali	dity	18.12.2017 to 17	12 2019	Last Amended on	1
van	uny	10.12.2011 10 11			
SI.	Product / Material of Test	Specific Test Performed	Test Method	Range of Testing / Limits of Detection	CV%
55.	Blood/ Genomic DNA	Krabbe disease (GALC) deletion/ duplication analysis	MLPA	Qualitative within limits of MLPA	NA
56.	Blood/ Genomic DNA	Pantothenate kinase- associated neurodegeneration (PANK2) deletion/ duplication analysis	MLPA	Qualitative within limits of MLPA	NA
57.	Blood/ Genomic DNA	Charcot-Marie-Tooth PMP22 deletion/duplication analysis	MLPA	Qualitative within limits of MLPA	NA
58.	Blood/ Genomic DNA	Dysferinopathy/LGM D2A (CAPN3) deletion/duplication analysis	MLPA	Qualitative within limits of MLPA	NA
59.	Blood/ Genomic DNA	Duchenne Muscular Dystrophy (DMD) deletion/duplication analysis	MLPA	Qualitative within limits of MLPA	NA
60.	Blood/ Genomic DNA	Congenital Muscular Dystrophy (LAMA2) deletion/duplication analysis	MLPA	Qualitative within limits of MLPA	NA
61.	Blood/ Genomic DNA	Dysferlinopathy/LGM D2B (DYSF) deletion/duplication analysis	MLPA	Qualitative within limits of MLPA	NA
62.	Blood/	RB1	MLPA	Qualitative within limits	NA
	Genomic DNA	deletion/duplication analysis		of MLPA	
	Blood/	NF1 and NF2 deletion/duplication	MLPA	Qualitative within limits of MLPA	NA

	He	IBL	OF ACCRED		201	Jed floor, Norgana Nethalaja Buldeng, Negrava Arebit Core, 2340, Bennmasandro, Negrava Roads, Bangdiore, 560 099 India VIII (180 6075 4000 Vindia www.inedgenome.com
La	boratory	Medgenome Li	abs Private Limited.	SDF 17, 1st Floor, C-Block	CSE7	Date: 27 <sup>th</sup> November 2018
Ce	creditation Stand rtificate Number lidity	dard ISO 15189: 201 MC-2497	1	Page 7 of 7		To Whom So Ever It May Concern
	Product / Material of Test	18.12.2017 to 1 Specific Test Performed	Test Method	Range of Testing / Limits of Detection	CV%	This is to certify that MedGenome Labs Ltd. is giving an undertaking that the prices quoted in this quotation are not higher than what we had quoted to other Government hospitals across the country in the Financial year 2018-2019.
64.	Blood/ Genomic DNA	Spinal Muscular Atrophy (SMN1/SMN2) deletion/duplication	MLPA	Qualitative within limits of MLPA	NA	If you need any further information, please feel free to contact us. Thanking you,
65.	Blood/ Genomic DNA	analysis Canavan disease (ASPA) deletion/ duplication analysis	MLPA	Qualitative within limits of MLPA	NA	For MedGenome Labs Ltd.
66.	Blood/ Genomic DNA	Tay-Sachs disease (HEXA) deletion/ duplication analysis	MLPA	Qualitative within limits of MLPA	NA	Authorized Signatory
67.	Blood/ Genomic DNA	Rett Syndrome (MECP2) deletion/ duplication analysis	MLPA	Qualitative within limits of MLPA	NA	·
68.	Blood/ Genomic DNA	X-linked spastic paraplegia-2 (PLP1) deletion/duplication analysis	MLPA	Qualitative within limits of MLPA	NA	
69.	Blood/ Genomic DNA	NDP Gene Deletion/ Duplication Analysis	MLPA	Qualitative within limits of MLPA	NA	
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Hosur Road, Bangalore - 560 099, India T: +91(0) 80 6715 4901 www.medgenome.com		ы	MEDGE
	Rate Quotation	Date: 27 <sup>th</sup> N	ovember 2018
To, Dr. Neerja Gupta [MD (Pediatrics), DM (Medical Genetics)], As: All India Institute of Medical Sciences & Rese Ansari Nagar, New Delhi, Delhi 110029		artment of Gen	etics
	Sequencing & Analys		
Description	Unit Price per Sample (INR)	Quantity	Price (INR)
<ul> <li>Whole Exome panel.</li> <li>Sample type: Peripheral Blood 3-4 ml</li> <li>or &gt; 100µL for DNA (EDTA Lavender top or DNA in sealed Eppendort tube)</li> <li>OC analysis of samples and Data</li> <li>NGS Sequencing using 2×150bp reads ig 100x on Illumina Hiseq or XTen platforms.</li> <li>Bioinformatics analysis with Illumina HiseqXen</li> <li>Clinical Report of 70 patients</li> <li>Dired and In Sast format</li> <li>Turn Around Time 5-8 Week</li> <li>Sanger confirmation for NNE VARIANT If identified will be part of the project.</li> <li>Kit Used: Sequencing libraries will be prepared using <u>Agilent-Sare Select VS</u></li> <li>21,522 genes covered (coding regions and splice junctions)</li> <li>Targeted exots – 3,57,999</li> </ul>			
Amount in words: Eighteen Lacs	and Ninety Thousa	and only	18,90,000.00

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