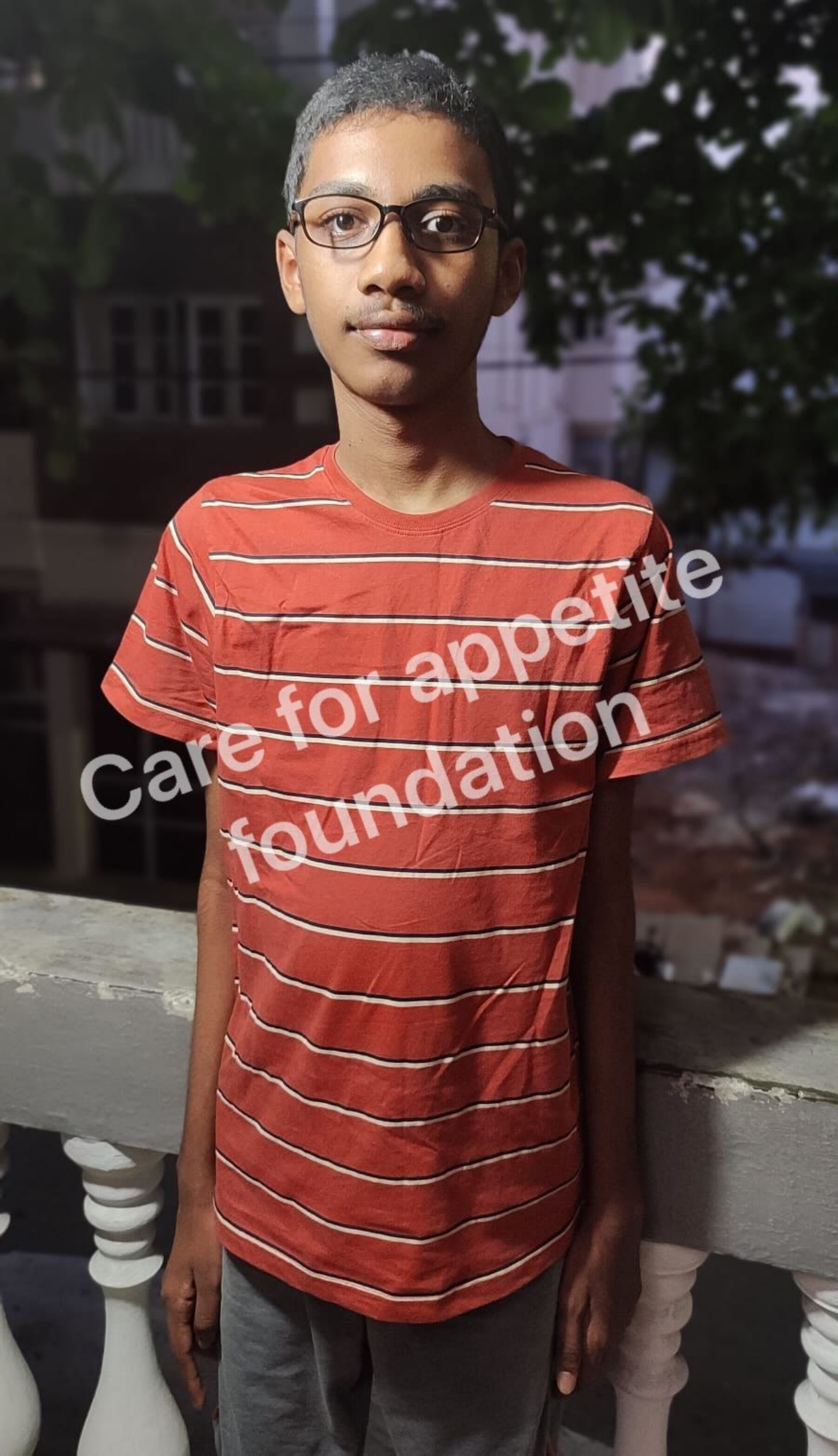
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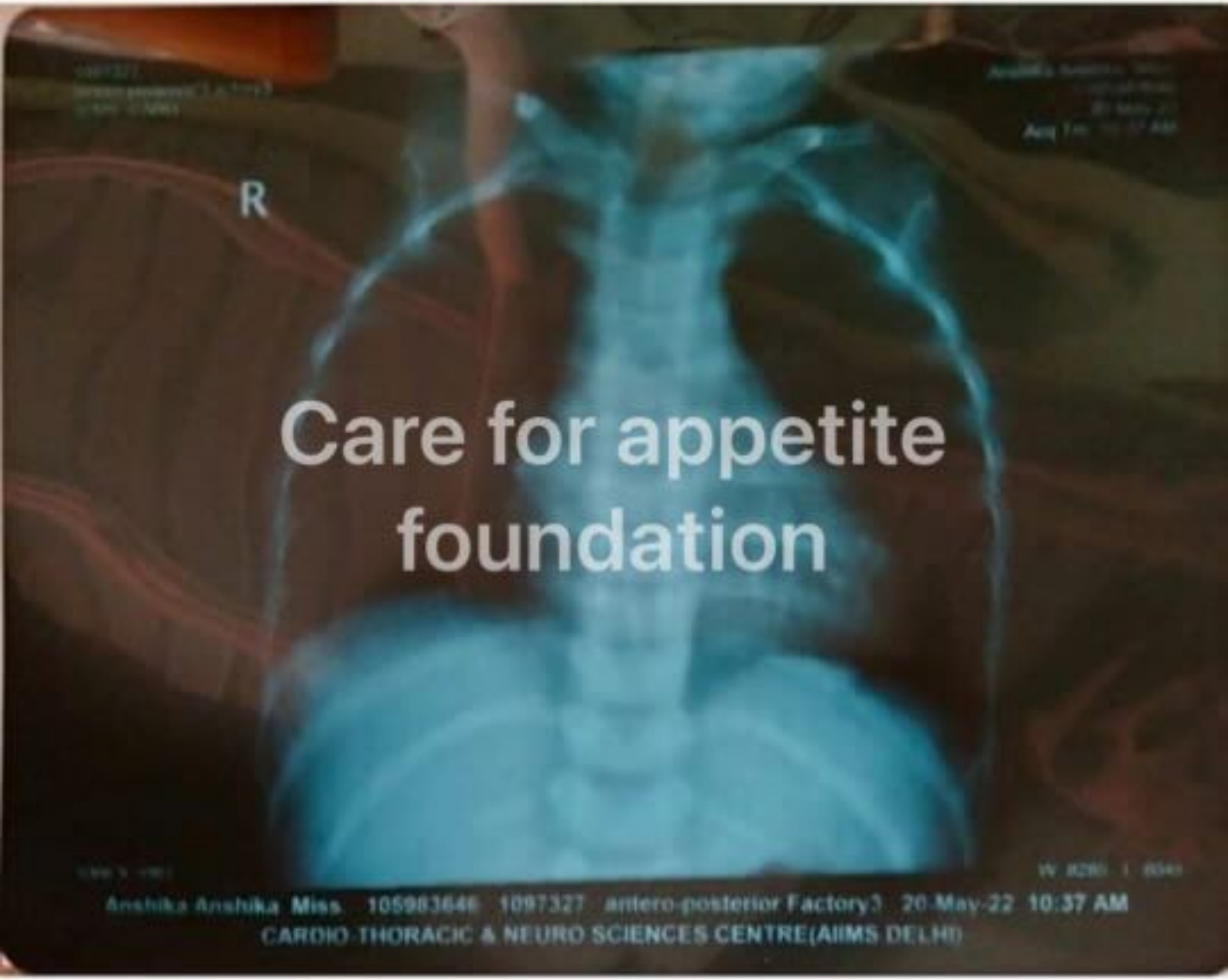
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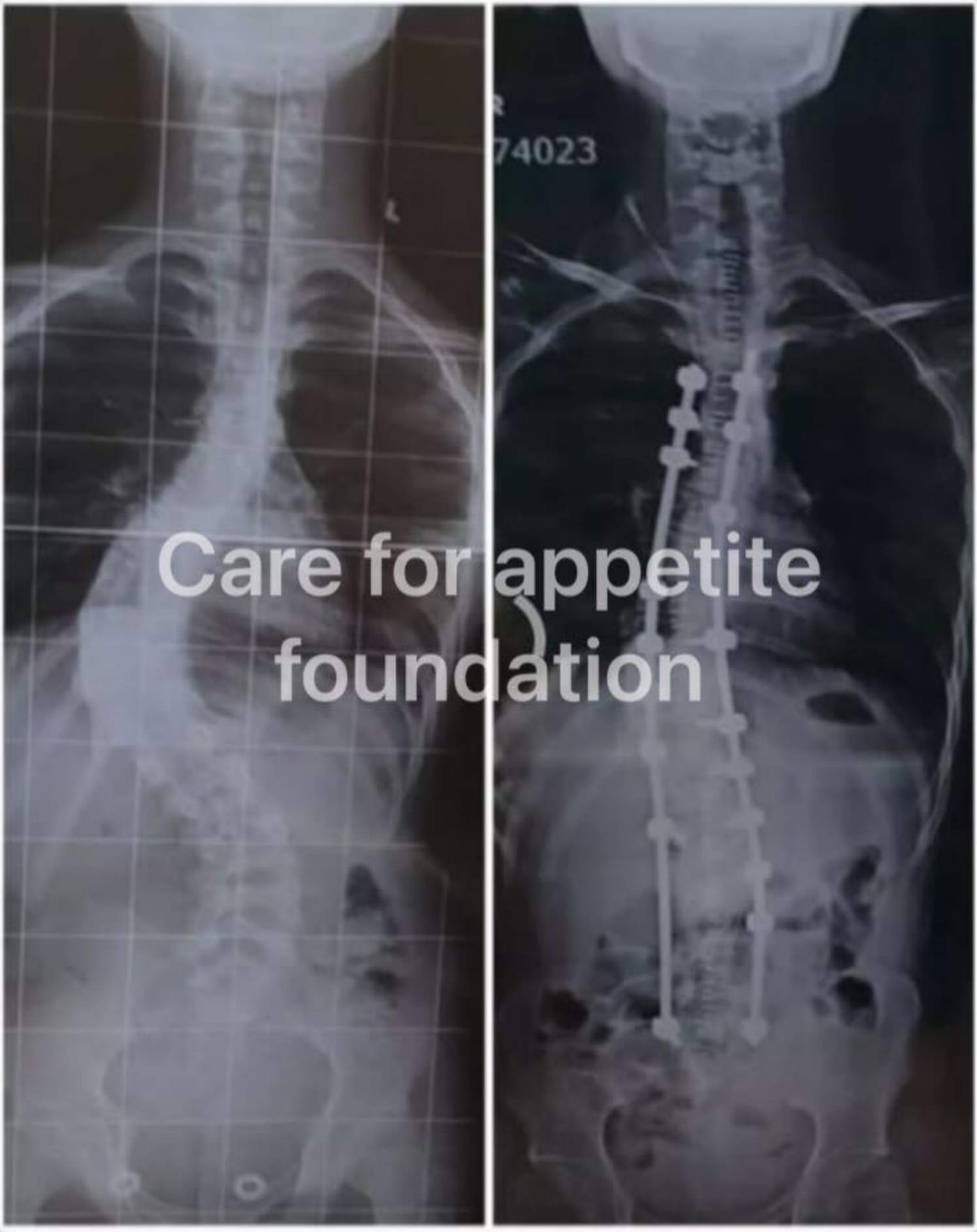
CARDIO-THORACIC & NEURO SCIENCES CENTRE(AIIMS DELHI)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MLPASure: SMA Test Report

PATIENT INFORMATION

Name	: Kundan V	Sample Type	:	Blood
Date of Birth	: 13/10/2006	Collection Date	:	27/08/2019
Gender	: Male	Collection Time (Hrs.)	:	08:40
Age (years)	: 12 years 10 months	Receipt Date	:	28/08/2019
Patient ID	: -	Report Date	:	09/09/2019
Sample ID	: BECGI195777	Reporting Time (Hrs.)	:	08:00
Test Code	: MLP-SMA-ECGI	Clinician Name	:	Dr. A A Mathew
Test Method	: MLPA	Hospital Name	:	Sagar Hospital, Bengaluru

RESULTS

***SMN1*: Zero copies of exon 7 and exon 8 in *SMN1* gene identified.** Homozygous deletion on exon 7 and exon 8 in *SMN1* gene was observed. This indicates that individual is likely to be affected of SMA

***SMN2*: Greater than four copies of exon 7 and 8 in *SMN2* gene identified.** Ambiguous duplication in exon 7 and exon 8 of *SMN2* gene observed.

INTERPRETATION

A sample from this individual was referred to our laboratory for molecular testing for Spinal Muscular Atrophy (SMA). SMA is a group of autosomal recessive neuromuscular disorders characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy.

SMN Gene	Exons	Dosage quotient*	Copy Number Status	Deletions / Duplications
SMN1	Ex 7	0.0	0 copies	Homozygous deletion
	Ex 8	0.0		Homozygous deletion

SMN Gene	Exons	Dosage quotient*	Copy Number Status	Deletions / Duplications
SMN2	Ex 7	5.56	>4 copies	Ambiguous duplication
	Ex 8	3.34		Ambiguous duplication

*MLPA probe ratio-Dosage quotient (DQ) – Homozygous wild type: 0.85<DQ<1.20, Homozygous deletion: DQ=0.0, Heterozygous deletion: 0.40<DQ<0.65, Heterozygous duplication: 1.30<DQ<1.65, Homozygous duplication: 1.75<DQ<2.15, Ambiguous duplication: DQ>2.20

Comment: The above mentioned result must be interpreted in the context of the individual's clinical and biochemical profile. **Genetic counseling is advised.**

Note: The SALSA MLPA probemix P060-B2 SMA detects deletions/duplications in specific regions in *SMN1* and *SMN2* genes only. Smaller deletions, duplications and point mutations in these genes or elsewhere in the genome will not be detected by this technique.

METHODOLOGY

Mutational analysis by multiplex ligation probe dependent amplification (MLPA, MRC Holland) using SALSA MLPA probe mix P060-B2 SMA for *SMN1* and *SMN2* gene. Analysis was done by Coffalyser (designed by MRC-Holland).
Note: Despite all precautions, the error rate in molecular tests can be 1-2%. We strongly recommend that this report should be correlated with clinical information.

REFERENCE

Prior TW, Nagan N, Sugarman EA, Batish SD, Braastad C. Technical standards and guidelines for spinal muscular atrophy testing. Genet Med. 2011 Jul;13(7):686-94. doi: 10.1097/GIM.0b013e318220d523. PubMed PMID: 21673580.

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REPORT OF HISTOPATHOLOGICAL EXAMINATION

Name: Master Kundan Age:4yrs Sex:M

Neuropath No.:X-2579 /11
ID No.

Ref. By:Drs.Krishna Prasad, Suguna Hospital, B.lore

Nature of Specimen:
Received muscle biopsy (irregularly sectioned), appearing fibrous with intramuscular septum(1.5x1x0.8cm)

Histopathology report:
Paraffin and cryosections from skeletal muscle tissue shows preserved architecture, moderate perimysial fibrosis and adipose tissue infiltration. Fascicular architecture is partly effaced. Fascicles comprise hypertrophic and atrophic fibers in groups and an admixture of hypertrophic and atrophic fibers in a few. Enzyme stains reveal type I fiber hypertrophy and grouping and group atrophy of type II fibers.

Impression:
Spinal muscular atrophy-Type II (vastus lateralis)

Dr.N.Gayathri
Addl.Professor
16/08/11

Professor
16/08/11

Bangalore Baptist Hospital

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Bangalore Baptist Hospital

10/11/21
Bengaluru

Name: Kundan Veluru
DOB: 13/10/06
Age: 15 years and 1 month
Weight: 37.8 kgs

RISDIPLAM (EVRYSDI) 60 mgs in each vial- to be mixed with 79 ml of sterile water to make up to a volume of 80 ml. So the concentration is 0.75mg in each ml.

Dose: To take 5 mgs once daily = 6.6 ml once daily x 6 months and continue.

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Fellow Paediatric Epilepsy
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