From: "ROOT" <root@sctimst.ac.in>
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Date: 03/02/2025 03:45 PM
Subject: Invitation for CGR

Greetings from AIIMS, Rishikesh !!

The CGR will be held on the **Feb 4, 2025** in **CPD Hall**, AIIMS Rishikesh from **8:00 AM to 9:00 AM**. You can join online through the following link:

Meeting link:
https://aiimsrishikesh.webex.com/aiimsrishikesh/j.php?MTID=m0c5b23dd0cdab43b989938baa675f002
Tuesday, Feb 5, 2025, 8:00 AM | (UTC+05:30) Chennai, Kolkata, Mumbai, New Delhi

Meeting number: 2517 940 0402
Meeting password: 040225

Thanks & Regards
Regional Resource Centre
Dept of Telemedicine
AIIMS Rishikesh

CLINICAL GRAND ROUNDS

Department of Neonatology- 4th February 2025

Name: B/O S	Age/Sex: 6days/ male	Residence: Kashipur, UK
UHID: 20240165923		
Case Presenter: **Dr Gaurav Gautam** **(Academic SR)**		**Consultant in charge:** **Dr. Sriparna Basu** **(Professor and HOD)**

Chief complaints:

- Poor spontaneous respiratory efforts
- Failure to wean from ventilator support
- Recurrent apnea upon weaning

Brief History:

Outborn, term, AGA neonate referred to AIIMS on 6th day of life with issues of poor spontaneous efforts, multiple extubation failures and failure to wean from respiratory support.

Mother was a 37 years old multigravida and had an uneventful antenatal period. There was also history of similar issues in previous sibling since birth who expired in 3rd week of life.

During the course in AIIMS, baby was extubated on DOL- 10 but could not be weaned off from non-invasive respiratory support. There was h/o recurrent apnea each time upon weaning.

Examination

General Examination

- Baby was lethargic with poor spontaneous activity at presentation
- No pallor, Icterus, cyanosis
- No facial dysmorphism

Vitals

- T- Temperature- 36.6 deg C
- A- Airway- baby intubated, SpO2- 95%
- B- Breathing- minimal spontaneous efforts, RR- 30-35/min
- C- Circulation- baby pink, CRT- 2 sec, HR- 140 bpm, pulses- good, BP- 65/41(50) mmHg
- RBS- 109 mg/dL

Systemic examination

- Respiratory System – B/L air entry equal, no added sounds
- Per abdomen - Soft, non-distended, no organomegaly, bowel sounds+
- Cardiovascular System - S1 S2 heard, no murmur
- Central Nervous System – activity reduced, tone decreased

Neurological Examination

Higher mental functions	
Prechtl State	State 3-4
Inter state variability	**Less variability**
Defensive reaction	Not done
Habituation (Glabellar tap)	After 5 attempts (normal)
Auditory and visual orientation	Could not be done

Cranial Nerves

I	Not done
II	Pupillary reflex normal
III, IV, VI	Conjugate eye movements present
V	Rooting reflex present
VII	No features of facial nerve palsy
VIII	Not done
IX, X, XI, XII	**Pooling of secretion present**

Motor Examination	
Posture	Baby is nesting. **Arms and legs partially flexed**
Passive Tone	**Scarf sign- Elbow till midline** Square window- 30 degrees Popliteal angle- 110 degrees Adductor angle- 80 degrees Popliteal angle- 110 degrees **Heal to ear- till xiphisternum**
Active tone	**Arm recoil- partial** Rest could not be tested
DTRs	B/L knee jerk 2+
Primitive reflexes	Rooting, sucking and swallowing- normal **Plantar grasp- partial** **Palmar grasp- partial** Moro's- could not be tested

Differential Diagnosis

1. Perinatal asphyxia
2. Sepsis/ CNS infections
3. CNS malformations
4. Neuromuscular disorders
5. Congenital Myopathies
6. Neurometabolic disorders
7. Hypoventilation syndromes

Diagnostic Investigations:

Investigation	**Result**
CBC	Hb- 14.2, TLC- 11268, ANC- 4729, Platelet- 2.6 lac
Micro ESR/ CRP	4 mm/ 1mg/dL
KFT	Urea- 48, Creatinine- 0.35
Na/ K/ Ca/ PO4	132/ 4.4/ 8.7/ 2.4
LFT	TsB/DsB- 11.2/ 0.8, OT/PT- 92/ 25, ALP- 224, T. Prt/ Alb- 4.7/ 3.1
CSF analysis	Acellular, Sugar- 74, Protein- 120, C/S- sterile
Blood C/S	Sterile
aEEG	Continuous normal voltage pattern with sleep wake cycle
Neurosonogram	Normal
Creatine Kinase (CK)	47 U/L (normal)
TSH	1.704 mIU/L (normal)
MRI brain	Normal
WES	**PHOX2B gene mutation detected- s/o congenital central hypoventilation syndrome**

Management

Baby was kept on non-invasive positive pressure ventilation (NIPPV) via nasal mask during the course of NICU stay. WES was s/o Congenital Central Hypoventilation Syndrome (CCHS). Parents were given options of tracheostomy, home ventilation or diaphragmatic pacing but they declined. Baby was discharged on DOL- 64 on BiPAP support. Baby was tolerating full enteral feeds, was neurologically normal and was gaining adequate weight at time of discharge.

Summary:

Outborn/ singleton/ term/ AGA/ male baby born to a 37 years old multigravida female presented to AIIMS on DOL-6 with issues of poor respiratory efforts, failure to wean from ventilator support and recurrent apnea upon weaning. There was history of previous sibling death due to similar illness at 3 weeks of age.

Baby was extubated on DOL-10 but could not be weaned-off from NIV support. Relevant investigations were all normal. Baby had recurrent apnea upon weaning from NIV support. WES was s/o Congenital Central Hypoventilation Syndrome. Baby was kept on NIV support throughout the NICU and was discharged on DOL-64 on BiPAP support.