



Partial Trisomy 12p in a Preterm Infant with Dysmorphic Features, Recurrent Apnea, Feeding Dysfunction, Neuroimaging Abnormalities, and a Large Left Diaphragmatic Hernia: A Case Report

Yasser Mohamed El Gohary^{1*}, Nuha Al Zaabi¹, Ahmed Adnan¹, Luay Abdalwahab¹, Olfat Al Zaabi¹, Reem Mahmoud¹, Ashraf Abboud Elabatal², Mohamed Ali Kasem¹, Aly Rashwan², Anshad Ummerkhan¹, Nawal Nawabjan¹, Wasimah Amanullah¹, Mays Alrim¹, Somar Alhourany¹

¹Department of Pediatrics and Neonatology, Fujairah Hospital, Emirates Health Services, Fujairah, United Arab Emirates

²Salma Rehabilitation Hospital, Abu Dhabi and Al Ain, United Arab Emirates

Email: *dr_yaser44@yahoo.com

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Abstract

Partial trisomy 12p is a rare chromosomal disorder caused by duplication of the short arm of chromosome 12. Its clinical manifestations are variable and may include craniofacial dysmorphism, developmental delay, hypotonia, feeding difficulties, and congenital anomalies. Neonatal presentations, particularly in preterm infants, remain infrequently reported. We report a preterm female infant born at 31 weeks and 5 days of gestation, with a birth weight of 1.6 kg, to a mother with diabetes mellitus. The infant required positive-pressure ventilation at birth, continuous positive airway pressure, subsequent intubation, and one dose of surfactant for respiratory distress syndrome. During a prolonged neonatal intensive care unit stay, she developed hypotonia, recurrent apnea with oxygen desaturation and bradycardia, excessive airway secretions, impaired sucking and swallowing, and prolonged dependence on respiratory support. Chromosomal microarray analysis demonstrated a pathogenic copy-number gain involving 12p13.33-p11.1, consistent with partial trisomy 12p. Neuroimaging revealed mild ventriculomegaly, germinal matrix cystic changes interpreted as sequelae of prior intraventricular hemorrhage, and later CT abnormalities concerning for hypoxic-ischemic injury. At 4 months of age, persistent respiratory and feeding difficulties prompted further diagnostic evaluation, which identified a large left diaphragmatic hernia containing the stomach and spleen within the left hemithorax, associated with compressive atelec-

tatic changes in the left lower lobe and adjacent consolidation. Following multidisciplinary assessment and family counseling, pediatric surgical repair was recommended. This case expands the documented neonatal phenotype of partial trisomy 12p and highlights the importance of continued diagnostic reassessment when respiratory and feeding abnormalities persist despite an established genetic diagnosis. The diaphragmatic hernia may have contributed substantially to respiratory morbidity; however, the recurrent apnea was likely multifactorial, and a causal relationship between the 12p duplication and the diaphragmatic hernia cannot be established from a single case.

Subject Areas

Pediatrics

Keywords

Partial Trisomy 12p, Chromosome 12 Duplication, Prematurity, Neonatal Dysmorphism, Apnea, Feeding Dysfunction, Hypotonia, Diaphragmatic Hernia, Chromosomal Microarray

1. Introduction

Partial trisomy 12p is a rare chromosomal duplication syndrome caused by complete or partial duplication of the short arm of chromosome 12. The syndrome exhibits considerable phenotypic variability depending on the size and location of the duplicated segment. Common clinical manifestations include characteristic craniofacial features, developmental delay, intellectual disability, hypotonia, feeding difficulties, and variable congenital malformations.

Most reported patients are diagnosed during infancy or childhood after developmental concerns become evident. Reports describing the neonatal presentation are relatively uncommon. We describe a preterm neonate with partial trisomy 12p who presented with multiple dysmorphic features, prolonged respiratory support requirements, recurrent apnea, feeding dysfunction, and neuroimaging abnormalities.

2. Case Presentation

2.1. Antenatal and Perinatal History

A female infant was born at 31 weeks and 5 days of gestation at a hospital in the United Arab Emirates to a mother with diabetes mellitus. Birth weight was 1.6 kg, length 42.5 cm, and head circumference 29.5 cm. The available neonatal records did not document the mode of delivery or specific prenatal imaging findings.

At birth, the infant was pale with poor respiratory effort and a heart rate between 60 and 100 beats per minute. Positive-pressure ventilation was administered via T-piece resuscitator, with improvement in heart rate and color. Apgar scores were 5 and 7 at one and five minutes, respectively. These findings document perinatal cardiorespiratory compromise; however, the available records did not include an um-

bilical cord blood gas or a formal diagnosis of perinatal asphyxia.

The infant was placed on continuous positive airway pressure immediately after delivery. Due to worsening respiratory distress and tachypnea at three hours of age, she was intubated and received one dose of surfactant for respiratory distress syndrome. At seven hours of age, she was transferred to a tertiary neonatal intensive care unit.

Initial venous blood gas analysis showed pH 7.29, PCO₂ 37.4 mmHg, bicarbonate 17.7 mmol/L, base excess -8.1, and a reported venous PO₂ of 86 mmHg.

2.2. Physical Examination

Physical examination revealed multiple dysmorphic features including:

- Low-set ears
- Folding of the left ear lobule
- Hypertelorism
- Broad nasal bridge
- Smooth philtrum
- High-arched palate
- Persistent fisting of both hands
- Wide anterior fontanelle
- Open sagittal and posterior sutures
- Cranial asymmetry
- Sacral dimple

Neurological examination demonstrated reduced spontaneous activity, hypotonia, poor sucking reflex, weak palatal and pharyngeal reflexes, and impaired feeding readiness.

2.3. Genetic Evaluation

Because of the presence of multiple dysmorphic features and neurodevelopmental concerns, chromosomal microarray analysis was performed in the proband. It identified a pathogenic copy-number gain involving chromosome 12p13.33-p11.1, reported as 12p13.33p11.1(64,621_34,650,483)x3, consistent with partial trisomy 12p. The original case record reported normal parental conventional karyotyping and chromosomal microarray results, supporting a *de novo* origin. The father's available follow-up microarray report confirmed a normal male result with no clinically significant copy-number variant or significant absence of heterozygosity. That parental study used a SNP-based CytoScan HD Array, analyzed with Chromosome Analysis Suite (ChAS; Affymetrix), and interpreted according to ACMG technical standards (2020). The proband's reference genome build and whether additional clinically relevant copy-number variants were identified were not stated in the supplied proband documentation and therefore could not be verified.

2.4. Respiratory Course and Apnea

The infant experienced a prolonged respiratory course characterized by recurrent

episodes documented clinically as apnea accompanied by oxygen desaturation and, on some occasions, bradycardia. The chart described central/obstructive apnea and required transitions among invasive ventilation, nasal continuous positive airway pressure, nasal conventional mechanical ventilation, and high-flow nasal cannula therapy. Exact episode counts were not consistently recorded in the available notes. On 8 June 2026, at a corrected gestational age of 41 weeks and 3 days and a weight of 3.5 kg, she remained on CPAP with PEEP 5.5; the preceding day she had a brief episode of apnea with desaturation and bradycardia lasting a few seconds.

Many episodes were associated with excessive airway secretions and improved following suctioning, supporting an obstructive component superimposed on possible central respiratory dysregulation. Regular gentle suctioning was incorporated into care, and glycopyrrolate was used to reduce secretion burden; the transfer note documented a dose of 0.25 mg per dose with planned titration. The combination of improvement after suctioning and persistent need for positive airway pressure supported a mixed central and obstructive mechanism.

At corrected gestational age of 38 weeks, the infant had been clinically stable on high-flow nasal cannula support at 5 L/min in room air. By 8 June 2026, she again required CPAP because of recurrent apnea. Blood gas at that time was acceptable (pH 7.40, PCO₂ 37.2 mmHg, HCO₃ 22.7 mmol/L).

2.5. Neurological Findings

Serial cranial ultrasonography during the NICU admission demonstrated persistent mild dilatation of the left lateral ventricle, measuring approximately 5 mm. Subsequent ultrasonography showed bilateral germinal matrix cystic changes together with persistent ventricular prominence, greater on the left. These cystic germinal matrix changes were interpreted clinically as sequelae of prior intraventricular hemorrhage. The exact postnatal ages of the earlier ultrasound examinations were not available in the records reviewed for this report.

A follow-up CT brain was still pending in the acute-care note dated 8 June 2026. A subsequent CT demonstrated the following abnormalities (**Figure 1** and **Figure 2**):

- Left frontal ping-pong skull deformity
- Hyperdensities involving both thalami
- Periventricular hypodense changes adjacent to the occipital horns

The bilateral thalamic hyperdensities and periventricular hypodense changes were considered radiologically concerning for neonatal hypoxic-ischemic injury. Because the available records did not document the exact date/postnatal age of the CT or definitive clinical criteria for hypoxic-ischemic encephalopathy, the imaging findings are described as concerning for hypoxic-ischemic injury rather than as proof of a specific perinatal hypoxic event.

Neurosurgical assessment concluded that the skull deformity was likely related to the underlying congenital abnormality or cranial developmental disorder. No

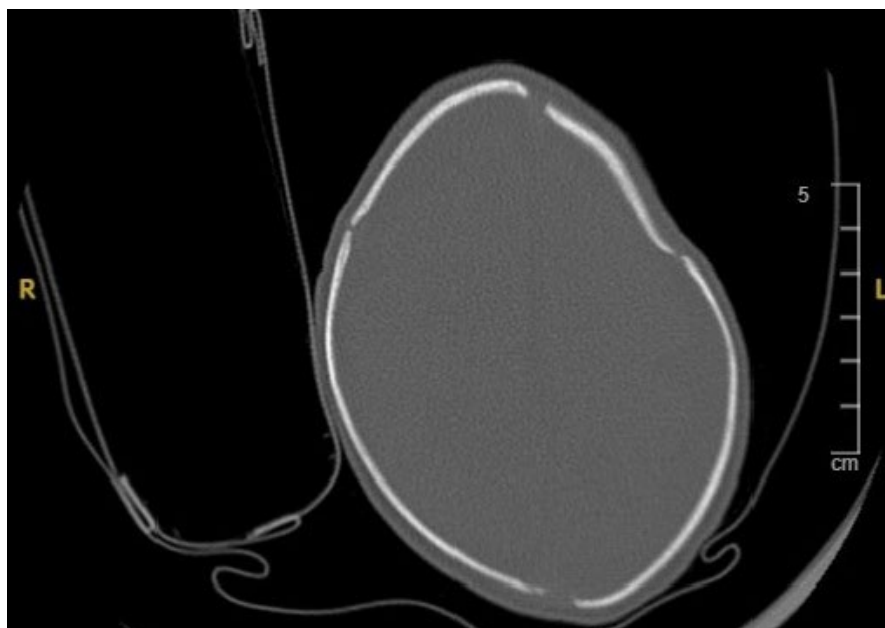


Figure 1. Brain CT showing bilateral thalamic hyperdensities and periventricular hypodense changes adjacent to the occipital horns. The corresponding image was not embedded in the source manuscript file supplied for this revision.

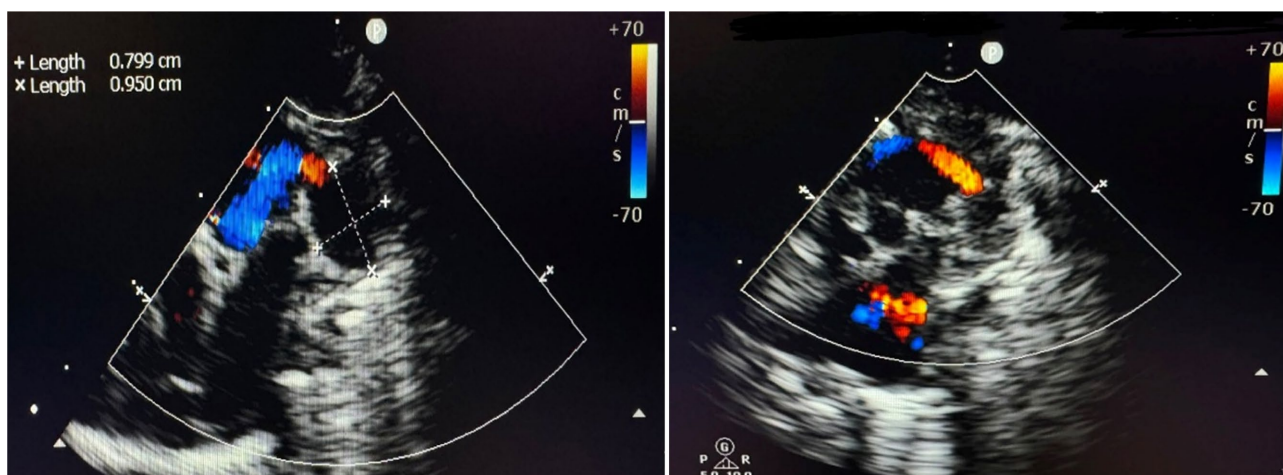


Figure 2. Skull CT showing a left frontal ping-pong skull deformity.

hydrocephalus or subdural collection was identified, and surgical intervention was not recommended.

2.6. Cardiovascular Findings

Echocardiography performed during the neonatal period demonstrated a patent ductus arteriosus with unusual saccular morphology. The narrowest diameter measured approximately 2 mm at the pulmonary end and showed restrictive left-to-right shunting (**Figure 3**).

Follow-up echocardiography demonstrated spontaneous closure of the ductus with otherwise normal intracardiac anatomy and function. The acute-care record dated 8 June 2026 also documented no pulmonary hypertension.

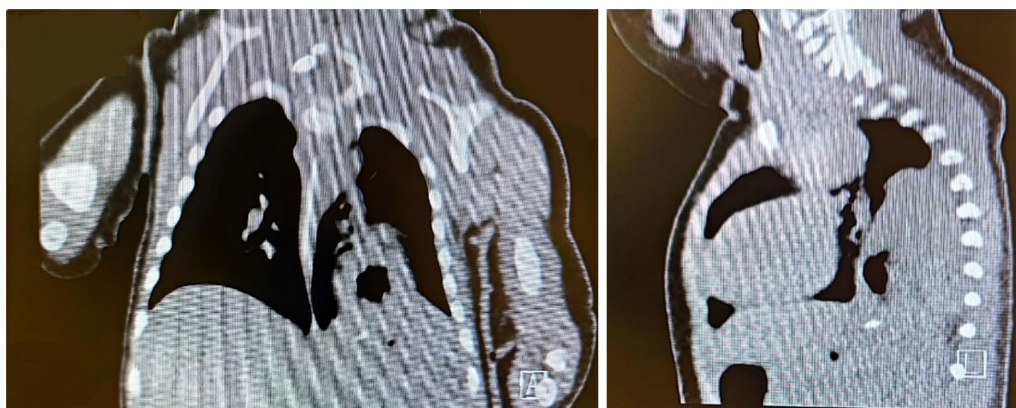


Figure 3. Echocardiographic patent ductus arteriosus views showing unusual saccular morphology. The narrowest diameter was approximately 2 mm at the pulmonary end, with restrictive left-to-right shunting.

2.7. Feeding, Growth, and Other Clinical Findings

Feeding difficulties became evident early in the clinical course. Speech and feeding assessments demonstrated:

- Moderate to severe oral secretions
- Weak suck response
- Absent response to lateral oral stimulation
- Poor palatal reflexes
- Poor pharyngeal reflexes
- Impaired coordination of oral feeding

The infant received oral stimulation therapy and non-nutritive sucking exercises. Enteral nutrition consisted initially of expressed breast milk fortified with human milk fortifier. By 8 June 2026, she was receiving 60 mL every 3 hours by orogastric tube with expressed breast milk and/or Aptamil Advance because human milk fortifier was unavailable.

Despite feeding challenges, growth was satisfactory, with weight increasing from 1.6 kg at birth to 2.77 kg at corrected gestational age of 38 weeks and to 3.5 kg at corrected gestational age of 41 weeks and 3 days.

The infant experienced indirect hyperbilirubinemia requiring phototherapy during the early neonatal period.

Laboratory evaluation documented an elevated thyroid-stimulating hormone concentration of 10.2 mIU/L, with thyroxine described as normal in the original case record. Subsequent thyroid testing on 18 May 2026 documented T4 27.29 and T3 6.68; units were not provided in the available note. The supplied records did not document a repeat TSH value, specific thyroid treatment, or a final thyroid-management decision.

The clinical course was further complicated by recurrent conjunctival infections. Eye cultures grew *Pseudomonas aeruginosa* and subsequently *Serratia* species, both of which responded to targeted antimicrobial therapy. Intermittent leukopenia and anemia of prematurity were also observed. Multiple blood cultures

remained negative.

Earlier abdominal and spinal ultrasonography were reported as normal. A repeat abdominal ultrasound on 8 June 2026 showed a mildly prominent left renal pelvis measuring 3.8 mm, with otherwise unremarkable abdominal findings.

2.8. Follow-Up and Left Diaphragmatic Hernia

At 4 months of age, the patient remained clinically stable in a chronic care facility. She continued to require nasal CPAP for recurrent apnea, tolerated enteral feeding, and maintained adequate urine and stool output. An upper gastrointestinal contrast study performed because of persistent respiratory and feeding concerns showed the gastric fundus and part of the gastric body projecting into the left hemithorax, with mild gastroesophageal reflux. Subsequent chest CT confirmed a large left diaphragmatic hernia containing the stomach and spleen within the left hemithorax, associated with compressive atelectatic change of the left lower lobe and adjacent consolidation. The available imaging reports did not definitively classify the defect as Bochdalek, hiatal, or paraesophageal; therefore, the term "left diaphragmatic hernia" is used consistently in this report. Cardiology review found no contraindication to general anesthesia. Pediatric surgery recommended operative repair, with possible fundoplication, after family discussion and consent. At the time of manuscript preparation, operative findings and postoperative outcomes were not available.

3. Discussion

Partial trisomy 12p is a rare chromosomal duplication syndrome with a recognizable but variable phenotype. Characteristic craniofacial features may include hypertelorism, abnormal ears, a broad nasal bridge, a high-arched palate, and generalized facial dysmorphism, many of which were present in this patient. These findings are consistent with published descriptions of trisomy 12p and disorders involving increased 12p dosage [1]-[5].

Hypotonia and developmental impairment are among the most consistently reported manifestations of partial trisomy 12p. In this infant, poor feeding, weak oral reflexes, excessive secretions, recurrent apnea, and reduced spontaneous activity indicated substantial neurological and bulbar dysfunction from the neonatal period. Similar combinations of hypotonia, feeding difficulty, and respiratory instability have been described in patients with trisomy 12p or related 12p dosage abnormalities [3]-[5].

The coexistence of recurrent apnea, severe oral-motor dysfunction, ventriculomegaly, germinal matrix cystic changes, and an unusual cranial contour contributed to a complex neonatal course. Reports comparing trisomy 12p with Pallister-Killian syndrome demonstrate phenotypic overlap but also emphasize that mosaic tetrasomy 12p is a distinct cytogenetic disorder and should not be considered equivalent to isolated partial trisomy 12p [3] [4] [6].

The prolonged need for respiratory support was likely multifactorial, reflecting

prematurity, respiratory distress syndrome, hypotonia, impaired airway clearance, excessive secretions, and possible central respiratory dysregulation. Improvement after suctioning suggests that secretion-related upper-airway obstruction also played an important role. These observations support a mixed central and obstructive mechanism rather than a single cause for the apnea.

Neuroimaging demonstrated mild ventriculomegaly, evidence of previous germinal matrix injury, and findings concerning for hypoxic-ischemic injury. These abnormalities may reflect a combination of prematurity-related germinal matrix injury, perinatal cardiorespiratory compromise, and potentially abnormal underlying neurodevelopment. The relative contribution of the 12p duplication cannot be determined from this single case. Preterm infants are independently susceptible to germinal matrix hemorrhage and hypoxic-ischemic brain injury [7].

The spontaneous closure of the unusual saccular patent ductus arteriosus was reassuring. Congenital cardiac abnormalities have been described in disorders involving increased 12p dosage, but cardiac involvement is variable, and this infant ultimately had normal intracardiac anatomy and function [2]-[5].

A clinically important additional finding was the large left diaphragmatic hernia containing the stomach and spleen. Late-presenting congenital diaphragmatic hernia can produce nonspecific respiratory or gastrointestinal manifestations and may be overlooked when symptoms are attributed to prematurity, neurological impairment, gastroesophageal reflux, or recurrent infection [8]. In this infant, intrathoracic displacement of the stomach and spleen with left lower-lobe compression provides a plausible additional mechanism for persistent respiratory-support needs and feeding difficulty. Nevertheless, the apnea was probably multifactorial, and the available evidence does not establish that the diaphragmatic defect was caused by the 12p duplication. Although diaphragmatic hernia has been recognized in Pallister-Killian syndrome and other disorders involving increased 12p dosage, its relationship with apparently isolated partial trisomy 12p remains poorly defined [3] [4].

The coexistence of these findings raises the possibility that diaphragmatic abnormalities may represent an uncommon component of the phenotypic spectrum associated with 12p duplication; however, given the rarity of the condition and the limited number of comparable cases, a direct causal relationship cannot be established from a single observation. Further reports of patients with overlapping 12p duplications and similar diaphragmatic abnormalities are required to determine whether this represents a true genotype-phenotype association or a coincidental finding. Surgical repair is generally considered for symptomatic pediatric diaphragmatic hernia [8]. When fundoplication is contemplated because of associated reflux or anatomy, the decision should be individualized according to reflux burden, neurological status, and operative findings [9]-[11]. **Table 1** compares the present case with previously reported partial trisomy 12p cases and Pallister-Killian syndrome [3]-[6].

This case contributes to the limited literature on the neonatal manifestations of

partial trisomy 12p by highlighting severe oral-motor dysfunction, secretion-related airway compromise, recurrent apnea, neuroimaging abnormalities, and a large left diaphragmatic hernia diagnosed during follow-up. It also demonstrates the value of longitudinal reassessment: persistent or changing symptoms should not automatically be attributed solely to prematurity or to an established chromosomal diagnosis [3]-[6] [8].

Table 1. Comparison of the present case, previously reported partial trisomy 12p cases, and Pallister-Killian syndrome.

Feature	Present case	Previous partial trisomy 12p cases	Pallister-Killian syndrome
Facial dysmorphism	+	Common	Common
Hypotonia	+	Common	Common
Feeding dysfunction	Severe	Reported	Common
Apnea/respiratory dysfunction	+	Variable	Reported
Brain abnormalities	+	Variable	Common
Cardiac abnormality	PDA	Variable	Reported
Diaphragmatic hernia	+	Rarely reported	Recognized
Developmental impairment	Too young to fully assess	Common	Common

References for **Table 1:** [3]-[6].

4. Conclusions

We report a preterm infant with partial trisomy 12p who presented with multiple dysmorphic features, hypotonia, severe feeding dysfunction, recurrent apnea and bradycardia, prolonged respiratory-support requirements, and neuroimaging abnormalities. Chromosomal microarray analysis established the diagnosis after recognition of a syndromic phenotype. A large left diaphragmatic hernia containing the stomach and spleen was subsequently identified during evaluation of persistent respiratory and feeding concerns.

This case emphasizes the importance of considering chromosomal disorders in neonates with dysmorphic features and unexplained neurological or respiratory manifestations. It also illustrates the need for continued diagnostic reassessment when clinical abnormalities persist despite an established diagnosis. Early genetic diagnosis supports coordinated multidisciplinary care, anticipatory guidance, developmental surveillance, and family counseling.

Declarations

Ethics Approval

A verified institutional ethics determination or approval/reference number was not available in the source materials supplied for this revision. This statement

must be confirmed with the responsible institution and target journal before submission.

Consent for Publication

Documentation confirming written informed consent for publication of the clinical information and anonymized images was not available in the source materials supplied for this revision. Written consent status must be verified before submission.

Author Contributions

All listed authors contributed to patient management and/or data collection, manuscript preparation, critical revision, and approval of the final manuscript, as stated in the original manuscript. The corresponding author should confirm that each listed author satisfies the target journal's authorship criteria before submission.

Conflicts of Interest

The authors declare no conflicts of interest.

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Abbreviations

BE	Base Excess
BW	Birth Weight
CMA	Chromosomal Microarray Analysis
CPAP	Continuous Positive Airway Pressure
CT	Computed Tomography
EBM	Expressed Breast Milk
EHS	Emirates Health Services
GA	Gestational Age
HIE	Hypoxic-Ischemic Encephalopathy/injury, as applicable
HMF	Human Milk Fortifier
HR	Heart Rate
IVH	Intraventricular Hemorrhage
NICU	Neonatal Intensive Care Unit
PDA	Patent Ductus Arteriosus
PCO ₂	Partial Pressure of Carbon Dioxide
PO ₂	Partial Pressure of Oxygen
PPV	Positive Pressure Ventilation
RDS	Respiratory Distress Syndrome
TSH	Thyroid-Stimulating Hormone