

# WHEN PPHN REFUSES TO RESOLVE

An interactive diagnostic case

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# A term infant with early warning signs

**39+3**

Weeks

**2270 g**

**46 cm**

**31 cm**

Weight – Length – HC

**8 / 9 / 9**

Apgar scores

## Initial course

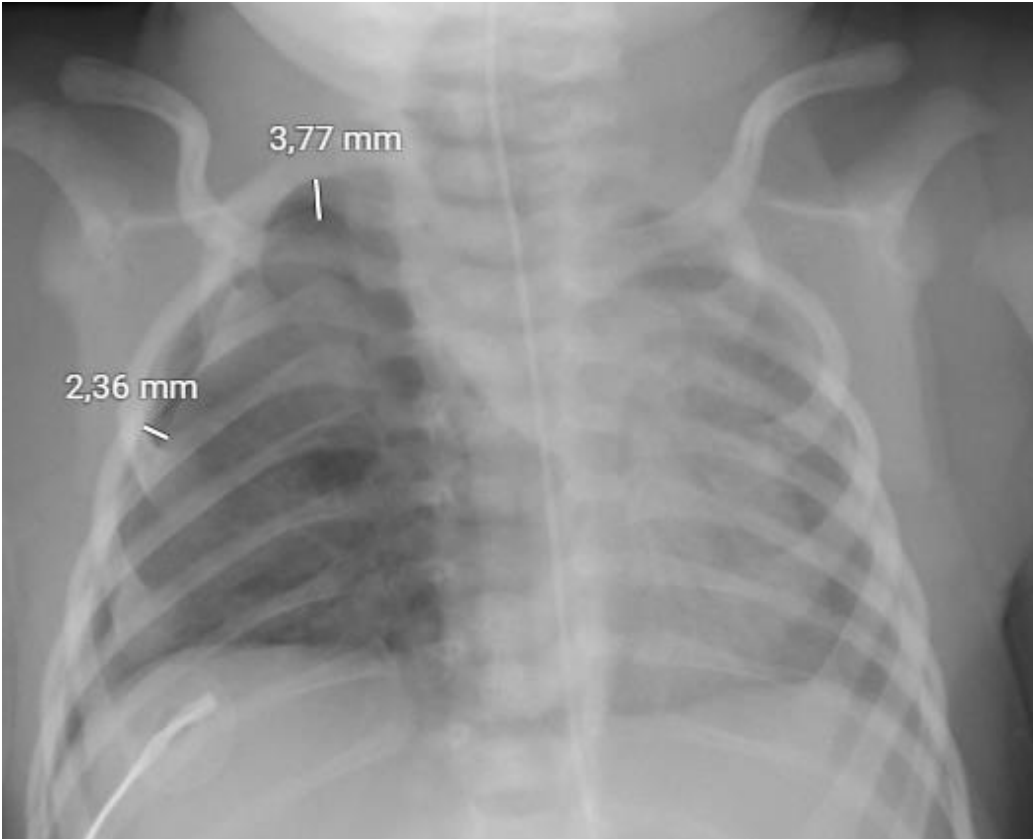
Uncomplicated pregnancy and vaginal delivery

No respiratory support immediately after birth

## Within hours

**Progressive hypoxaemia and  
respiratory distress**

# The first deterioration



Case radiograph | right pneumothorax

## 01 **Preductal 94% | postductal 85%**

Early evidence of right-to-left shunting

## 02 **FiO<sub>2</sub> increased to 1.0**

Saturations remained unstable

## 03 **Right pigtail inserted**

SpO<sub>2</sub> initially rose to 100%

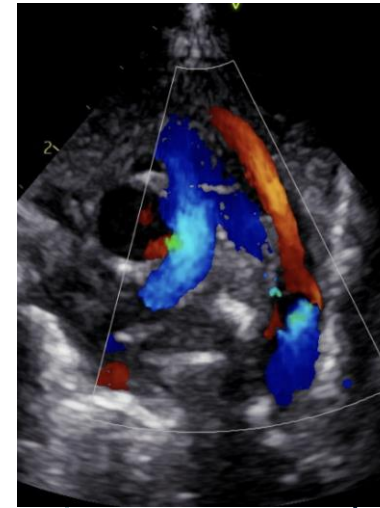
## 04 **During transfer**

Progressive desaturation to below 80%

# Echocardiography confirms the physiology

## Persistent pulmonary hypertension of the newborn

- **Structurally normal heart** No congenital cardiac explanation
- **Bidirectional PFO and ductal flow** Dynamic right-to-left shunting
- **D-shaped interventricular septum** Right ventricular pressure loading
- **TR gradient +/- 50 mmHg** Supports elevated pulmonary pressure
- **Normal pulmonary venous return** No macroscopic venous obstruction



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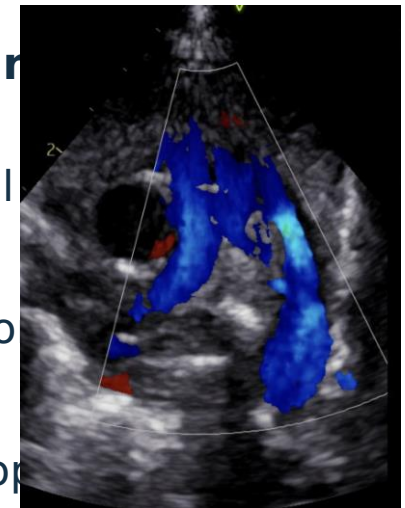
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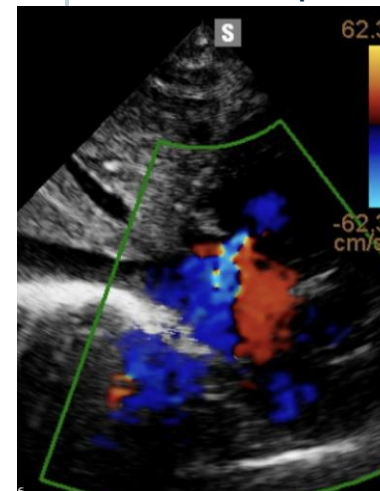
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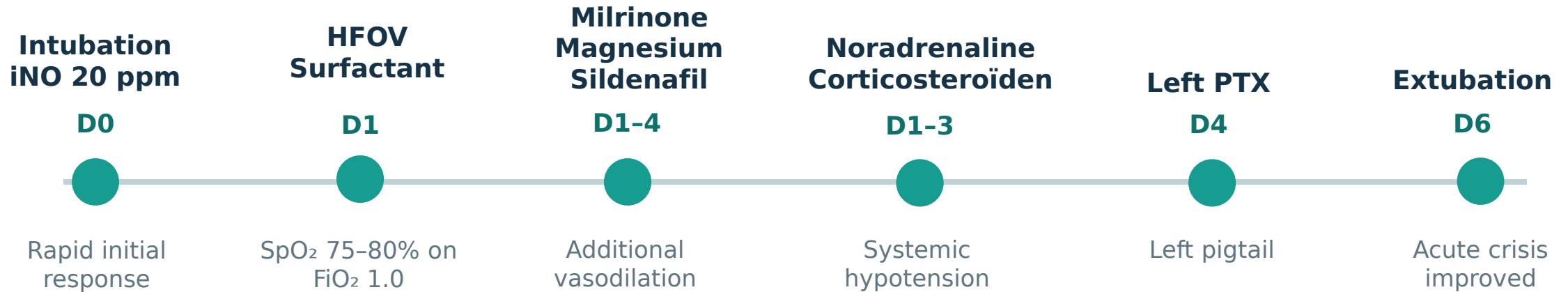
pulmonary vasculature



onary venous or  
tural heart disease



# The physiology becomes disproportionate



**Ventilation was adequate. Oxygenation repeatedly failed.**

# The recovery remains atypical

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**D6** Extubated without support

**D9** High-flow oxygen restarted

**D10** Both drains removed

**CT** No cysts, consolidation or interstitial pattern



Chest CT

# Refractory PPHN requires a broader differential

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## Lung development

Acinar dysplasia  
Congenital alveolar dysplasia  
Pulmonary hypoplasia

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## Capillary and venous disease

ACDMPV  
Pulmonary veno-occlusive disease  
Pulmonary capillary haemangiomatosis

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## Parenchymal disease

Surfactant dysfunction  
Rare diffuse lung disease

# Diagnosis

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Wu et al. *BMC Pediatrics* (2026) 26:491  
<https://doi.org/10.1186/s12887-026-06839-x>

BMC Pediatrics

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**CASE REPORT**

**Open Access**

## Chromosome 17q23.1-q23.2 deletion syndrome with severe pulmonary hypertension in neonates: two case reports and literature review



**BRIP  
1**

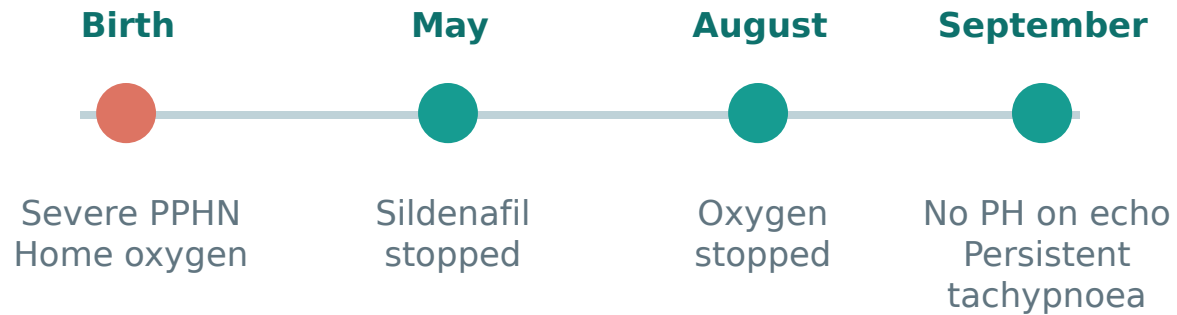
**TBX4**

The phe

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# Initial recovery does not end the risk

## This patient



## Take-home messages

- 01** PPHN that remains therapy-dependent into the second week warrants investigation for developmental causes
- 02** TBX4 variants and 17q23 deletions can cause neonatal PPHN, air leak and abnormal distal lung development.
- 03** Typically biphasic and need for lifetime follow-up

# THANK YOU FOR YOUR ATTENTION

Questions or comments?



# References

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