Volume 6 | Issue 2 | 2026

ISSN | 2771-019X

Journal of
Clinical & Medical Images
Case Reports

Open Access |Case Series

Pentalogy of cantrell: A report of two incomplete cases

***Corresponding Author:** M Elmassi*

Email: marwa.elmassi@gmail.com

Elmassi[1]*; H Mghazli[2]; A Lalaoui[2]; K Abi El Aala[2]; F Benaoui[2]; N Slitine[2]; FMR Maoulainine

[1]Research Laboratory on Childhood, Health and Development, FMPM, Cadi Ayyad University, Marrakech, Morocco.

[2]Neonatal Intensive Care Unit, Mohammed VI University Teaching Hospital, Marrakech, Morocco.

Abstract

Background: Pentalogy of cantrell (POC) is an extremely rare congenital malformation with an estimated incidence of 5.5 to 7.9 per million live births. It is defined by five cardinal anomalies: a supraumbilical abdominal wall defect, an anterior diaphragmatic defect, a diaphragmatic pericardial defect, a lower sternal defect, and congenital cardiac malformations. Incomplete forms associating two or more components have been described. Prognosis is largely determined by the type and severity of associated cardiac defects.

Objective: To report two cases of incomplete pentalogy of cantrell in female premature neonates and discuss the diagnostic and management challenges in a resource-limited setting, along with a review of the current literature.

Cases: Two female premature neonates, born one month apart, were diagnosed postnatally with incomplete POC at the neonatal intensive care unit of mohammed vi university teaching hospital, marrakech, morocco. The first case (35+5 weeks of gestation) presented with four components of the pentalogy and was hospitalised for one week with deferred surgical correction. The second case (34 weeks), born to a mother with no antenatal follow-up, presented with two components and was lost to follow-up after initial postnatal evaluation.

Conclusion: Incomplete forms of POC represent a significant diagnostic challenge, particularly in the absence of prenatal surveillance. A multidisciplinary approach and early systematic cardiac evaluation are essential to guide prognostic counselling and surgical planning. Strengthening antenatal care remains the most effective strategy to enable earlier diagnosis and optimal perinatal management.

Received: Jun 06, 2026

Accepted: Jul 27, 2026

Published Online: Aug 03, 2026

Cite this article: Elmassi M, Mghazli H, Lalaoui A, El Aala KA, Benaoui F, et al. Pentalogy of cantrell: A report of two incomplete cases. J Clin Med Images Case Rep. 2026; 6(2): 1828.

Keywords: Pentalogy of cantrell; Congenital abdominal wall defect; Sternal cleft; Ectopia cordis; Incomplete form; Neonatal management; Rare congenital malformation.

Introduction

Pentalogy of Cantrell (POC) is a complex and extremely rare congenital malformation with an estimated incidence of 5.5 to 7.9 per million live births [1]. First described in 1958 by Cantrell and colleagues based on five index cases, it is defined by the combination of five cardinal anomalies: a midline supraumbilical abdominal wall defect, an anterior diaphragmatic defect, a diaphragmatic pericardial defect, a lower sternal defect, and congenital cardiac malformations [2]. These anomalies result from a disruption of the differentiation or migration of mesodermal and mesenchymal structures occurring between the third and fifth weeks of embryonic development.

Incomplete forms, associating at least two components of the pentalogy without fulfilling all five criteria, have been reported in the literature [1]. The Toyama classification (1972), still widely used, distinguishes three classes according to the number of components present [1]. The clinical presentation is highly variable, ranging from isolated sternal anomalies to complete forms with ectopia cordis, the latter carrying a mortality rate exceeding 80% [7]. Diagnosis may be established prenatally by first-trimester ultrasonography or fetal MRI, or postnatally on initial clinical examination.

We report two cases of incomplete pentalogy of Cantrell diagnosed in the neonatal period in two female premature neonates managed at the Neonatal Intensive Care Unit of Mohammed VI University Teaching Hospital, Marrakech, Morocco. These cases illustrate the diagnostic challenges associated with the absence of prenatal surveillance and the specific management considerations in a resource-limited setting.

Case reports

Case 1

A female premature neonate was born at 35 weeks and 5 days of gestation by instrumental vaginal delivery in the context of severe pre-eclampsia complicated by retroplacental haematoma, with breech presentation. The pregnancy was poorly monitored, with no systematic prenatal ultrasound examination performed. No family history of congenital malformation, consanguinity, or exposure to teratogenic agents was identified. The mother had a history of one unexplored spontaneous miscarriage.

On initial clinical examination, the neonate was in good general condition: pink, reactive, with normal spontaneous activity. Vital parameters were as follows: heart rate 158 beats per minute (bpm), respiratory rate 60 cycles per minute, temperature 36.5°C, oxygen saturation (SpO_2) 92% on room air, and Capillary Refill Time (CRT) under 3 seconds. Anthropometric measurements showed a birth weight of 1,400 g, length 37 cm (< 10th percentile), and head circumference 27 cm (<3rd percentile). Neurological examination revealed satisfactory axial and peripheral tone with present primitive reflexes.

Morphological examination revealed a midline cleft measuring 60 mm in length, extending along the linea alba from the inferior hemisternum — equidistant from both nipples — to the umbilical cord (Figures 1, 2 & 3). The overlying skin was whitish and thinned, with a soft supra-sternal pouch whose volume increased with inspiration, consistent with agenesis of the sternal head and hypoplasia of the sternal body. No additional morphological anomaly was identified on segmental examination.

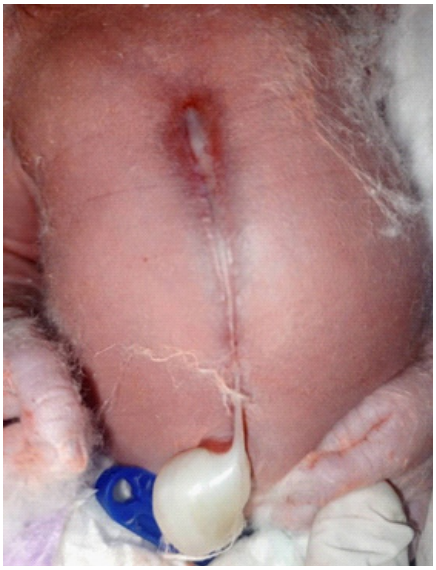

Figure 1: Case 1 — Overview of the premature neonate at birth, showing the anterior midline cleft.

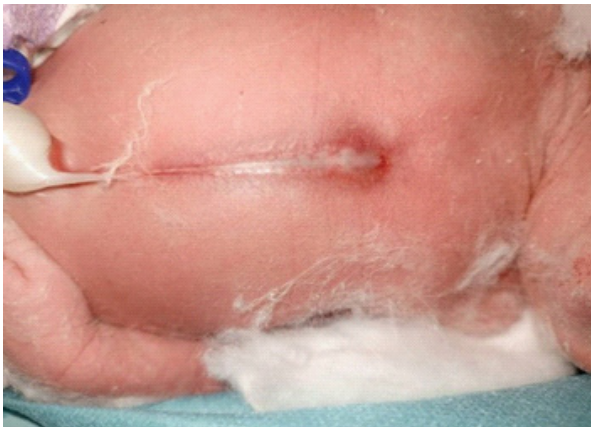

Figure 2: Case 1 — Close-up view of the lower sternal cleft: whitish, thinned skin with well-defined margins.

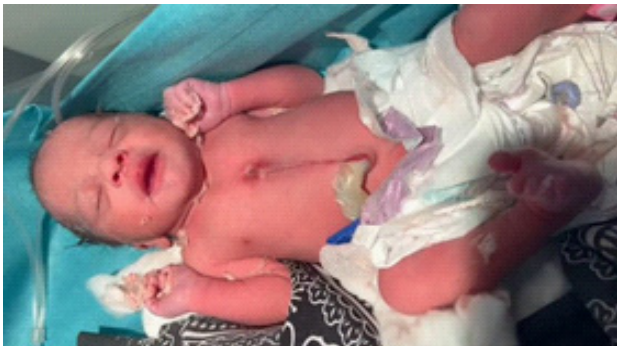

Figure 3: Case 1 — Sternal cleft extending vertically to the umbilical cord, with the soft supra-sternal pouch.

Investigations — Case 1

Transthoracic Echocardiography (TTE) performed on day 2 of life demonstrated a 2 mm Ventricular Septal Defect (VSD), a minimal Patent Ductus Arteriosus (PDA), and a small-to-moderate Patent Foramen Ovale (PFO). Abdominal ultrasonography revealed a semi-filled gallbladder with two intraluminal hyperechoic foci, with reassessment recommended after strict fasting.

Thoracoabdominal CT angiography confirmed the following findings: dextrocardia, an inferior sternal cleft measuring 43 mm, an anterior diaphragmatic defect, and a supraumbilical linea alba hernia. Post-contrast images demonstrated significant right ventricular dilatation with an increased RV/LV ratio, dilatation of the main pulmonary artery trunk, a ventricular diverticulum measuring 36×25 mm, and a supraumbilical hernia consistent with clinical findings.

Outcome — Case 1

The neonate was admitted to the neonatal intensive care unit for seven days. The immediate clinical course was favourable,

with no respiratory distress or haemodynamic decompensation. In view of the prematurity, low birth weight, and complexity of the anatomical anomalies, surgical correction was deferred pending adequate weight gain and multidisciplinary team review involving paediatric surgery, paediatric cardiology, and neonatology. The family received detailed counselling regarding the diagnosis, prognosis, and planned management strategy.

Case 2

A female premature neonate was born at 34 weeks of gestation by spontaneous eutocic vaginal delivery without obstetric incident. Preterm labour was spontaneous with no identified aetiology. The pregnancy was poorly monitored, with no antenatal ultrasound or other prenatal investigations performed. No family history of congenital malformation or exposure to teratogenic agents was noted.

On initial clinical examination, the neonate was in good general condition: pink, reactive, with normal spontaneous activity. Vital parameters were: heart rate 143 bpm, respiratory rate 64 cycles per minute, temperature 36.2°C, SpO_2 94% on room air, and CRT under 3 seconds. Anthropometric measurements showed a birth weight of 2,200 g, length 39 cm (<10th percentile), and head circumference 28 cm (<3rd percentile). Neurological examination was unremarkable.

Morphological examination revealed a sternal cleft extending from the inferior hemisternum — equidistant from both nipples — to the diaphragm (Figures 4 & 5), with macroscopic features identical to those observed in Case 1: whitish, thinned overlying skin with a soft supra-sternal pouch showing respiratory variation, together with a supraumbilical abdominal wall defect.

Figure 4: Case 2 — Thoracoabdominal view showing the soft supra-sternal pouch and the supraumbilical abdominal wall defect.

Investigations — Case 2

Chest radiograph confirmed the absence of the sternum. Infectious workup (complete blood count, C-reactive protein, blood culture) was negative. Transthoracic echocardiography could not be performed prior to discharge.

Outcome — Case 2

In the absence of vital distress, inpatient admission was not retained. Surgical correction was deferred and a multidisciplinary outpatient follow-up appointment was scheduled. The family was subsequently lost to follow-up after the initial evaluation, precluding any further clinical monitoring.

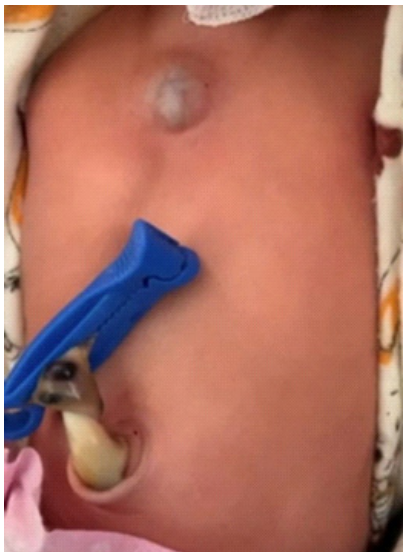

Figure 5: Case 2 — Close-up view of the lower sternal cleft with soft pouch showing respiratory variation.

Discussion

Pentalogy of Cantrell is a rare nosological entity first described in 1958 [2], with an estimated incidence of 5.5 to 7.9 per million live births [1]. Most published series report a male predominance, with an M/F ratio of approximately 2.7:1 [6]. The occurrence of two consecutive cases in female neonates, managed in the same unit one month apart, represents a notable epidemiological finding that contrasts with available literature data.

Both cases correspond to incomplete forms of the pentalogy, consistent with published reports acknowledging that combinations of at least two components constitute accepted expressions of the syndrome spectrum [1]. Table 1 summarises the components identified in each patient in comparison with the complete form.

Table 1: Components identified in each patient in comparison with the complete form.

Component	Complete form	Case 1	Case 2
Supraumbilical abdominal wall defect	√	√	√
Anterior diaphragmatic defect	√	√	—
Diaphragmatic pericardial defect	√	—	—
Lower sternal defect	√	√	√
Congenital cardiac malformation	√	√	Not assessed*

*Transthoracic echocardiography was not performed prior to discharge (Case 2).

From an embryological standpoint, Cantrell and colleagues proposed a two-category pathogenic framework: the first encompasses defects resulting from disruption of normal mesodermal development, leading to diaphragmatic, pericardial, and intracardiac anomalies; the second relates to

defects arising from perturbation of primordial mesenchymal structures, causing sternal and abdominal wall anomalies [2]. One or both categories may be simultaneously affected, accounting for the phenotypic variability of the syndrome.

Complementary genetic aetiologies have been reported. Associations with chromosomal abnormalities including trisomy 18, trisomy 21, trisomy 13, and Turner syndrome have been described [3]. Goltz-Gorlin syndrome — a rare multisystemic disorder of ecto-mesodermal origin linked to mutations in the X-linked PORCN gene — has also been reported in coexistence with POC [5]. In both cases presented here, no chromosomal investigation was carried out, which constitutes a notable limitation of the aetiological workup.

Prenatal diagnosis relies primarily on first-trimester ultrasonography, which may identify anterior wall anomalies, cardiac malformations, and diaphragmatic defects [8]. Fetal MRI serves as a valuable complement for detailed anatomical characterisation and surgical planning [8]. In both reported cases, diagnosis was exclusively postnatal due to the complete absence of antenatal surveillance — a situation not uncommon in resource-limited settings that underscores the importance of strengthening antenatal care programmes.

Congenital cardiac malformations represent the principal prognostic determinant in POC [6]. Ventricular septal defect is identified in approximately 72% of cases, and tetralogy of Fallot in approximately 20% based on original data from Cantrell et al. [2]. In Case 1, the combination of a 2 mm VSD, minimal PDA, patent foramen ovale, and a 36 × 25 mm ventricular diverticulum is consistent with the cardiac components described in the literature [6]. The survival rate of the complete form without surgery is below 20% [7]; following surgical correction, rates of up to 37% have been reported in selected series [9]. For incomplete forms such as those presented here, the prognosis is generally more favourable, primarily conditioned by the severity of residual cardiac anomalies.

The decision to defer surgical correction in both patients was justified by prematurity, low birth weight in Case 1 (1,400 g at 35+5 weeks), and clinical stability in the absence of vital distress. This approach is consistent with current recommendations advocating deferred correction in haemodynamically stable patients, with initial conservative management [3,9]. Recent studies have reported normal growth and development up to six years of age in children who underwent successful early surgical repair [7].

The loss to follow-up of Case 2 after initial evaluation represents a major limitation and reflects the difficulties of sustained clinical monitoring in socioeconomically precarious settings. It highlights the need for active social support and structured follow-up mechanisms for families of children with rare congenital malformations.

Conclusion

Incomplete forms of pentalogy of Cantrell present a significant diagnostic and therapeutic challenge, particularly in the absence of prenatal surveillance. The two cases reported here illustrate the phenotypic diversity of the syndrome and the specific management considerations encountered in a resource-limited neonatal context. The female predominance observed in our series contrasts with the male preponderance reported in the literature and warrants further documentation in larger series.

A multidisciplinary approach involving neonatology, paediatric cardiology, paediatric surgery, and medical genetics is essential from the time of birth. Systematic cardiac evaluation — including CT angiography when transthoracic echocardiography is insufficient — is central to prognostic assessment and surgical planning. Strengthening antenatal care remains the most effective strategy to enable prenatal diagnosis, appropriate genetic counselling, and optimal preparation for perinatal management.

Declarations

Conflicts of interest: The authors declare no conflicts of interest.

Funding: This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

Ethical approval: As a case report involving standard clinical care, formal ethics committee review was not required at the authors' institution. All procedures were conducted in accordance with the ethical standards of the Declaration of Helsinki.

Parental consent: Written informed consent was obtained from the parents of both patients for the publication of clinical data and photographic material in this manuscript.

Data availability: All relevant data are contained within the manuscript. No additional datasets were generated or analysed.

Acknowledgements: The authors wish to thank the nursing and medical staff of the Neonatal Intensive Care Unit of Mohammed VI University Teaching Hospital for their contribution to patient care.

References

1. Toyama WM. Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium, and heart: a case report and review of the syndrome. Pediatrics. 1972; 50: 778–792.

2. Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. Surg Gynecol Obstet. 1958; 107: 602–614.

3. Mallula KK, Sosnowski C, Awad S. Spectrum of Cantrell's pentalogy: case series from a single tertiary care center and review of the literature. Pediatr Cardiol. 2013; 34: 1703–1710.

4. Morales JM, Patel SG, Duff JA, Villareal RL, Simpson JW. Ectopia cordis and other midline defects. Ann Thorac Surg. 2000; 70: 111–114.

5. Smigiel R, Jakubiak A, Lombardi MP, Jaworski W, Slezak R, Patkowski D, Hennekam RC. Co-occurrence of severe Goltz-Gorlin syndrome and pentalogy of Cantrell: case report and review of the literature. Am J Med Genet A. 2011; 155A: 1102–1105.

6. Mallula KK, Sosnowski C, Awad S. Spectrum of Cantrell's pentalogy: case series from a single tertiary care center and review of the literature. Pediatr Cardiol. 2013; 34: 1703–1710.

7. Morales JM, Patel SG, Duff JA, Villareal RL, Simpson JW. Ectopia cordis and other midline defects. Ann Thorac Surg. 2000; 70: 111–114.

8. McMahon CJ, Taylor MD, Cassady CI, Olutoye OO, Bezold LI. Diagnosis of pentalogy of Cantrell in the fetus using magnetic resonance imaging and ultrasound. Pediatr Cardiol. 2007; 28: 172–175.

9. Vazquez-Jimenez JF, Muehler EG, Daebritz S, Keutel J, Nishigaki K, Huegel W, Messmer BJ. Cantrell's syndrome: a challenge to the surgeon. Ann Thorac Surg. 1998; 65: 1178–1185.