SSR Institute of International Journal of Life Sciences
ISSN (O): 2581-8740 | ISSN (P): 2581-8732
Sunita *et al*., 2026
doi: 10.21276/SSR-IIJLS.2026.12.6.6

Case Report

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Right Lower-limb Phocomelia with Anorectal Malformation and a Rudimentary Caudal Appendage in a Neonate without Thalidomide Exposure: Case Report

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Received: 25 Jul 2026/ Revised: 15 Aug 2026/ Accepted: 26 Oct 2026

ABSTRACT

Background: Phocomelia is a rare congenital limb-reduction defect. Its coexistence with anorectal malformation (ARM) and a caudal appendage is unusual. This report aims to highlight the clinical presentation, anaesthetic management, and importance of evaluating associated caudal anomalies in such neonates.

Methods: We report a case of a term neonate presenting with right lower-limb phocomelia, ARM, and a rudimentary caudal appendage. Clinical examination, available investigations, anaesthetic management, surgical intervention, and postoperative follow-up were reviewed.

Results: A one-day-old male neonate (2.4 kg), born at 38 weeks and 5 days of gestation, presented with absence of the right femur, tibia and fibula, with the foot arising from the hip. Examination revealed ARM, an absent anal opening and a 1-cm caudal appendage. No maternal thalidomide exposure was reported. Echocardiography was normal, but genetic testing and spinal imaging were unavailable. Emergency high divided sigmoid colostomy was performed under general anaesthesia with a caudal epidural block. Intubation, haemodynamic stability and extubation were uneventful. The neonate was discharged on postoperative day five with a functioning colostomy and established breastfeeding.

Conclusion: The coexistence of lower-limb phocomelia, ARM and a caudal appendage suggests a broader caudal developmental anomaly. Evaluation for caudal regression syndrome, VACTERL association and occult spinal dysraphism is warranted. Spinal imaging before neuraxial blockade, genetic assessment where available and multidisciplinary follow-up are important, while antenatal anomaly screening may facilitate early diagnosis and counselling.

Key-words: Phocomelia; Limb reduction defect; Anorectal malformation; Caudal appendage; Caudal regression syndrome; Spinal dysraphism; Neonatal anaesthesia

INTRODUCTION

Phocomelia (from the Greek *phōkē*, "seal", and *melos*, "limb") is a severe congenital limb-reduction defect characterised by absence or marked hypoplasia of the proximal and/or intermediate limb segments, so that the hand or foot is attached directly, or almost directly, to the trunk [1].

How to cite this article
Sunita, Arti, Kumari M, Sidharth. Right Lower-limb Phocomelia with Anorectal Malformation and a Rudimentary Caudal Appendage in a Neonate without Thalidomide Exposure: Case Report. SSR Inst Int J Life Sci., 2026; 12(6): 11125-11130.

Access this article online
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In a worldwide study of 22.7 million births from 19 birth-defect surveillance programmes, the total prevalence of phocomelia was 0.62 per 100,000 births; 53.2% of cases were isolated, 36.9% occurred with other major congenital anomalies and 9.9% were part of a recognised syndrome, with a slight male excess (male-to-female ratio 1.23) [1]. Traditionally classified as a transverse intercalary deficiency, upper-limb phocomelia has been reinterpreted as a severe longitudinal dysplasia, because a true intercalary defect with a normal distal segment is rarely demonstrable [2].

Although phocomelia occurs independently of drug exposure, it became widely recognised during the thalidomide epidemic of 1957–1961, when more than

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doi: 10.21276/SSR-IIJLS.2026.12.6.6

10,000 children were born with limb and other malformations [3]. Outside this context, phocomelia has been associated with single-gene disorders, vascular disruption and other teratogenic or environmental exposures, although many cases remain unexplained [1, 4]. Lower-limb phocomelia is the less frequent pattern: among single-limb cases in the worldwide series, 64.9% involved an upper limb [1], and recent case reports have predominantly described upper-limb disease [4,5].

We report a neonate with unilateral right lower-limb phocomelia associated with an ARM and a rudimentary caudal appendage, born to a mother with no thalidomide exposure. The case is reported for this unusual combination of caudal anomalies, which raises a specific differential diagnosis and has direct implications for spinal evaluation before caudal anaesthesia. This report follows the CARE guidelines [6].

CASE PRESENTATION

Patient information- A 1-day-old male neonate weighing 2.4 kg was referred from a primary health centre to the paediatric emergency department of our tertiary-care hospital with multiple congenital anomalies noted at birth. He was born at 38 weeks and 5 days of gestation by normal vaginal delivery at a primary health centre, with Apgar scores of 7 and 9 at 1 and 5 min, respectively. The mother, aged 26 years (gravida 1, para 1, living 1), had not received routine antenatal care, and no obstetric ultrasonography had been performed after the initial confirmation of pregnancy. She reported no intake of thalidomide or any other medication during pregnancy, and specific enquiry confirmed no exposure to alcohol, tobacco, recreational drugs or radiation. Pre-gestational and gestational diabetes mellitus were excluded on the basis of the maternal history and blood glucose measurements. The parents were not consanguineous, and there was no family history of limb or other congenital anomalies on either side.

Clinical findings- On examination, the right femur, tibia and fibula were clinically absent, and the right foot arose from a small soft-tissue mass at the level of the right hip (Fig. 1). The left lower limb and both upper limbs were normal. The neonate had an ARM, with an absent anal opening and no perineal or urinary fistula, and a rudimentary caudal appendage approximately 1 cm long in the midline coccygeal region, covered with intact skin and with no overlying dimple, hair tuft or haemangioma. The external genitalia were normal, and no craniofacial anomaly such as cleft lip or palate, micrognathia or upslanting palpebral fissures was present. No cardiac murmur was heard on auscultation.



Fig. 1: Clinical photograph of the neonate showing right lower-limb phocomelia

Diagnostic assessment- Transthoracic echocardiography was performed to screen for associated cardiac anomalies and was normal. No vertebral or spinal imaging was performed, so vertebral anomalies and occult spinal dysraphism could not be excluded. The platelet count was 280 × 10^9/L and the preoperative blood glucose was 92 mg/dL. Limb and sacral radiography, renal ultrasonography, and spinal ultrasonography or magnetic resonance imaging were not performed. Karyotyping and molecular genetic testing were not available at our centre. A clinical diagnosis of non-thalidomide right lower-limb phocomelia with multiple congenital anomalies (ARM and caudal appendage) was made; a syndromic association could not be excluded without genetic testing.

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Table 1: Differential diagnosis of lower-limb phocomelia with anorectal malformation

Condition	Key features	Relevance to this patient
Caudal regression syndrome (sacral agenesis)	Sacral or lumbosacral vertebral agenesis; lower-limb hypoplasia; anorectal and genitourinary anomalies; associated with maternal diabetes	Leading consideration; needs sacral radiograph and spinal ultrasonography or MRI
VACTERL association	At least three of vertebral, anal, cardiac, tracheo-oesophageal, renal and limb anomalies	ARM plus limb defect; screen with echocardiography, renal ultrasonography and vertebral imaging
Femoral hypoplasia–unusual facies syndrome	Uni- or bilateral femoral hypoplasia; upslanting palpebral fissures, short nose, long philtrum, micrognathia, cleft palate; maternal diabetes in about one-third	Requires facial examination and maternal glycaemic history
Proximal femoral focal deficiency	Deficiency of the proximal femur, usually with tibia present	Less likely, because the tibia and fibula were also absent
Roberts syndrome / SC phocomelia ($ESCO_2$)	Usually symmetrical tetraphocomelia; cleft lip or palate; growth deficiency; autosomal recessive	Unlikely (single, unilateral limb); cytogenetic or $ESCO_2$ testing if available
Thalidomide embryopathy	Predominantly upper-limb, often bilateral reduction defects; ear, eye, cardiac and renal anomalies	Excluded by history
Sirenomelia	Fused lower limbs with severe renal and anorectal anomalies; usually lethal	Excluded clinically (separate lower limbs)
TAR and Holt–Oram syndromes	Radial-ray (upper-limb) defects with thrombocytopenia (TAR) or congenital heart disease (Holt–Oram)	Unlikely with a lower-limb-only phenotype

ARM, anorectal malformation; ESCO2, establishment of sister chromatid cohesion N-acetyltransferase 2; MRI, magnetic resonance imaging; TAR, thrombocytopenia–absent radius; VACTERL, vertebral, anal, cardiac, tracheo-oesophageal, renal and limb anomalies

Therapeutic intervention and perioperative management- An emergency high divided sigmoid colostomy under general anaesthesia was planned by a multidisciplinary team, and written informed consent was obtained from the parents. Standard American Society of Anesthesiologists (ASA) monitoring was applied, comprising electrocardiography, pulse oximetry, a precordial stethoscope, non-invasive blood pressure measurement with an appropriately sized neonatal cuff and a skin temperature probe. An orogastric tube was passed and the stomach decompressed before induction. Two 26-gauge peripheral intravenous cannulae were secured on the left hand. Anaesthesia was induced intravenously with fentanyl 5 µg (approximately 2 µg/kg), propofol 5 mg (approximately 2 mg/kg) and atracurium 1.2 mg (0.5 mg/kg). Laryngoscopy and tracheal intubation with a 3.0-mm internal diameter uncuffed tracheal tube were accomplished without difficulty at the first attempt, and the tube was fixed at 9 cm at the angle of the mouth.

After induction, a single-shot caudal epidural block was performed under aseptic precautions with 1.2 mL of 0.25% plain bupivacaine (0.5 mL/kg; 1.25 mg/kg). Spinal ultrasonography was not performed before the block, which was placed by the landmark technique with a 26-gauge needle. Anaesthesia was maintained with sevoflurane in a 50:50 mixture of oxygen and air at an age-adjusted minimum alveolar concentration of 0.8–1.0, with supplemental atracurium as required. The neonate was positioned supine with all pressure points padded, and normothermia was maintained with a forced-air warming device. Haemodynamic parameters remained stable throughout. Rectal paracetamol 24 mg (10 mg/kg) was administered at the end of surgery. The duration of surgery and anaesthesia was 1.5 h, during which the neonate received 50 mL of intravenous fluid and the blood glucose was 98 mg/dL.

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doi: 10.21276/SSR-IIJLS.2026.12.6.6

After reversal of neuromuscular blockade with neostigmine 1.2 mg and glycopyrrolate 0.48 mg, the trachea was extubated, and the neonate was observed in the recovery room for 45 min before transfer to the ward.

Follow-up and outcomes- The parents were counselled about the diagnosis and its uncertain aetiology. The postoperative course was uneventful: the colostomy functioned normally, breastfeeding was established and the neonate was discharged on the fifth postoperative day in a stable condition. Definitive repair of the ARM was not undertaken during this admission. At the most recent follow-up visit the infant was active and the colostomy was working well. The timeline of care is summarised in Table 2.

Table 2: Timeline of clinical events

Time point	Event
Antenatal period	No routine antenatal care; single early ultrasound for confirmation of pregnancy only; no reported thalidomide or other drug intake.
Day 0 (birth)	Normal vaginal delivery at a primary health centre. Right lower-limb phocomelia, ARM and caudal appendage noted at birth; referred from a primary health centre.
Day 1	Presented to the paediatric emergency department; clinical examination and echocardiography (normal); decision for emergency colostomy.
Day 1	General anaesthesia with caudal block; high divided sigmoid colostomy; tracheal extubation; 45 min in recovery room; transfer to the ward.
Postoperative day 5	Discharged in stable condition; colostomy functioning normally and breastfeeding established.
Age	Follow-up visit: infant active with a normally functioning colostomy; definitive repair of the ARM not yet performed; written consent for publication obtained from the parents.

ARM, anorectal malformation; NICU, neonatal intensive care unit

DISCUSSION

This case combines three caudal anomalies — unilateral lower-limb phocomelia, ARM and a rudimentary caudal appendage—in a neonate without thalidomide exposure. Lower-limb phocomelia is the less common pattern: in the International Clearinghouse for Birth Defects Surveillance and Research series, upper limbs were involved in 64.9% of single-limb cases, and among two-limb cases both lower limbs were affected in only 26.8% [1]. The same series found additional major anomalies in 36.9% of cases, and all four cases with ambiguous genitalia had lower-limb involvement [1], an observation consistent with a shared caudal developmental field. Our patient fits this pattern, and the anomalies are best regarded as a caudal field defect rather than an isolated limb malformation. The link between thalidomide and phocomelia shaped modern teratology.

Thalidomide was marketed in West Germany from 1957 as an over-the-counter sedative and was used for anxiety, insomnia and nausea in pregnancy; it was withdrawn in late 1961 after Lenz in Germany and McBride in Australia independently linked it to the epidemic of limb defects [3,7]. Exposure between approximately 20 and 36 days after fertilisation produces predominantly upper-limb reduction defects together with anomalies of the ears, eyes, heart, gastrointestinal and urinary tracts [3,7]. Its teratogenicity is mediated by binding to cereblon, a component of an E3 ubiquitin ligase complex [8]. Because thalidomide remains in clinical use for erythema nodosum leprosum and multiple myeloma [3], a specific drug history remains essential, and it was negative in our patient.

Non-thalidomide phocomelia has been attributed to genetic causes — most notably Roberts syndrome and SC phocomelia, autosomal recessive disorders caused by

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ESCO2 variants [9] — as well as to vascular disruption and other teratogenic exposures [1,4]. Maternal diabetes is an important risk factor for caudal and femoral developmental defects: about one-third of the reported infants with femoral hypoplasia–unusual facies syndrome were born to diabetic mothers [10], and maternal diabetes is the best-recognised association of caudal regression syndrome [11]. In our patient, maternal glycaemic status was normal, diabetes having been excluded by history and blood glucose testing. Because no antenatal anomaly scan was performed, the anomalies were first recognised at birth, which precluded prenatal counselling and planned delivery at a tertiary centre.

The combination of a lower-limb deficiency, ARM and a caudal appendage places caudal regression syndrome first; its confirmation requires sacral and spinal imaging [11]. VACTERL association must be considered whenever an ARM coexists with a limb defect, and requires echocardiography, renal ultrasonography, vertebral imaging and confirmation of oesophageal patency [12]. Femoral hypoplasia–unusual facies syndrome is distinguished by its characteristic facial features [10], whereas Roberts syndrome typically produces symmetrical tetraphocomelia with craniofacial anomalies [9]. Thrombocytopenia–absent radius and Holt–Oram syndromes predominantly affect the upper limbs and are unlikely in a lower-limb-only phenotype.

The most important practical lesson concerns the spine. Occult spinal dysraphism, particularly tethered spinal cord, is common in children with ARM: in a cohort of 110 neonates with ARM, tethered cord confirmed by magnetic resonance imaging was found in about 9%, and neonatal spinal ultrasonography showed 80% sensitivity and a 97% negative predictive value as a screening test [13]. Human tails are themselves markers of spinal pathology; in a review of 59 reported cases, spinal dysraphism was present in 49% and tethered cord in 20% [14]. Caudal block provides effective, opioid-sparing analgesia in neonates, but in a neonate with both an ARM and a caudal appendage we recommend spinal ultrasonography before any caudal or other neuraxial technique. Pre-block spinal imaging was not performed in our patient, which we acknowledge as a limitation.

In patients with phocomelia, absent or malformed limbs can make peripheral venous cannulation, non-invasive blood pressure measurement and arterial access difficult, and syndromic forms may be accompanied by airway abnormalities [15]. In our patient both upper limbs were normal, so venous access and blood pressure monitoring were possible on the upper limbs, with both cannulae secured on the left hand and an appropriately sized neonatal cuff used for blood pressure measurement, and laryngoscopy was straightforward. Because an underlying syndrome had not been excluded, a difficult-airway cart was kept ready. The ARM added neonatal-specific concerns—abdominal distension, impaired mask ventilation and a risk of regurgitation—which were managed by orogastric decompression before induction and by securing the airway rapidly with succinylcholine. The weight-based local anaesthetic dose (bupivacaine 1.25 mg/kg) was within recommended limits for paediatric regional anaesthesia [16].

LIMITATIONS

Genetic testing was not available, so a syndromic cause could not be confirmed or excluded; the absence of antenatal ultrasonography meant that prenatal diagnosis was not possible; spinal ultrasonography was not performed before the caudal block, so occult spinal dysraphism could not be excluded.

CONCLUSIONS

Lower-limb phocomelia accompanied by an ARM and a caudal appendage should be regarded as a marker of a broader caudal developmental defect rather than an isolated limb anomaly. Such neonates warrant systematic evaluation for caudal regression syndrome, VACTERL association and occult spinal dysraphism — including spinal ultrasonography before caudal or other neuraxial block — together with a detailed maternal drug and glycaemic history, genetic assessment where available, and coordinated follow-up by paediatric surgery, orthopaedics, rehabilitation and genetic counselling services. Wider access to antenatal anomaly ultrasonography would allow such anomalies to be detected before birth and delivery to be planned.

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doi: 10.21276/SSR-IIJLS.2026.12.6.6

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