

MIKROMELIA IN NEWBORNS: MIDWIFERY MANAGEMENT AND CLINICAL IMPLICATIONS

¹Yosefin Nainggolan, ²Tina, ³Ernawati Nasution, ⁴Agusmawati Zega, ⁵Nazibah Saridatun Nahla

^{1,2,3,4,5}Sekolah Tinggi Ilmu Kesehatan Mitra Husada Medan

Email: 2419201515@mitrahusada.ac.id , 2519201504@mitrahusada.ac.id , 2519201182@mitrahusada.ac.id ,
2519201742@mitrahusada.ac.id , 2519201805@mitrahusada.ac.id

ABSTRACT

Background: Micromelia is a rare congenital skeletal dysplasia characterized by generalized shortening of the upper and lower extremities due to impaired endochondral ossification during fetal development. Early detection and appropriate management are essential to reduce complications and improve neonatal outcomes. **Objectives:** This study aimed to describe the concept of micromelia and analyze the implementation of midwifery care management in infants diagnosed with this congenital anomaly. **Methods:** This study employed a descriptive case study approach based on literature review and application of Varney's seven-step midwifery management process. Data were collected through assessment, physical examination, identification of problems and needs, planning, implementation, and evaluation of care in an infant with micromelia. **Results:** The case assessment revealed generalized shortening of the upper and lower extremities, mild respiratory distress, weak sucking reflex, and risk of growth and developmental problems. Midwifery management included monitoring vital signs, maintaining body temperature, supporting breastfeeding, observing respiratory function, providing parental education, and collaborating with pediatric specialists for further evaluation and referral. The infant's condition remained stable, and the family demonstrated improved understanding of the condition and agreed to the referral plan. **Conclusions:** Comprehensive and evidence-based midwifery care plays an important role in the early detection, monitoring, referral, and family support of infants with micromelia. Multidisciplinary collaboration is essential to optimize clinical outcomes and improve the quality of life of affected infants.

Keywords: Micromelia; Congenital Anomaly; Skeletal Dysplasia; Midwifery Care; Newborn

INTRODUCTION

Micromelia is a rare congenital musculoskeletal disorder characterized by generalized shortening of the upper and lower extremities resulting from impaired endochondral ossification during fetal development. This condition is commonly associated with severe skeletal dysplasia, including achondrogenesis and thanatophoric dysplasia, which may lead to significant morbidity and mortality in neonates. Congenital anomalies remain a major public health concern worldwide because they contribute substantially to infant disability, long-term health complications, and neonatal deaths.

According to the World Health Organization (WHO), approximately 240,000 newborns die within the first 28 days of life each year due to congenital anomalies. Skeletal dysplasias are estimated to occur in approximately 1 in 3,000–5,000 live births globally, with micromelia representing one of the most severe manifestations of limb shortening. In Southeast Asia, congenital anomalies continue to be an important contributor to neonatal morbidity, particularly in areas with limited access to prenatal diagnostic services. In Indonesia, congenital abnormalities remain among the leading causes of neonatal mortality, emphasizing the need for improved antenatal screening, early diagnosis, and comprehensive neonatal management.

Various interventions and policies have been implemented to reduce the burden of congenital anomalies, including routine antenatal care, prenatal ultrasonography, genetic counseling, and referral systems for high-risk pregnancies. The Indonesian Ministry of Health also promotes integrated antenatal care services to facilitate early detection of fetal abnormalities. Previous studies have demonstrated that prenatal ultrasound and genetic testing significantly improve the detection and classification of skeletal dysplasias, enabling better pregnancy planning and neonatal management. However, evidence regarding comprehensive midwifery care management for infants with micromelia remains limited, particularly in the Indonesian context.

Considering the rarity of micromelia and its potential complications, comprehensive and evidence-based midwifery care is essential to ensure early identification, appropriate referral, family education, and multidisciplinary collaboration. Therefore, this study is necessary to describe and analyze the implementation of midwifery care management in infants with micromelia.

Despite advances in prenatal diagnosis and neonatal care, evidence regarding comprehensive midwifery management for infants with micromelia remains limited, particularly in Indonesia. Therefore, this study aims to describe the implementation of comprehensive midwifery care in an infant diagnosed with micromelia using the Varney management approach. The research question addressed in this study is:

How is comprehensive midwifery care implemented in an infant with micromelia to support early detection, intervention, referral, and neonatal outcomes?

METHODS

This study employed a descriptive case study design to describe the implementation of comprehensive midwifery care for an infant diagnosed with micromelia. The study was conducted using the Varney seven-step midwifery management approach and SOAP documentation to assess, plan, implement, and evaluate care provided to the patient.

The participant in this study was a female newborn diagnosed with micromelia who was delivered at 38 weeks of gestation. The infant presented with generalized shortening of the upper and lower extremities, mild respiratory distress, and weak sucking ability. Because this

research was conducted as a single-case study, no sample size calculation or sampling technique was required. The participant was selected purposively based on the diagnosis of micromelia and the availability of complete clinical data.

Data were collected from patient assessments, maternal interviews, physical examinations, medical records, and observation of the infant's clinical condition. The instruments used included neonatal assessment forms, a thermometer for body temperature measurement, a stethoscope for heart and respiratory rate assessment, anthropometric measurement tools for body weight and length evaluation, and SOAP documentation sheets. The dependent variable in this study was the infant's clinical condition, including respiratory status, body temperature stability, feeding ability, and adaptation to extrauterine life. The independent variable was the implementation of comprehensive midwifery care based on the Varney management process. Measurements were conducted through routine clinical observation, physical examination, and evaluation of neonatal outcomes during the care period.

Data analysis was performed descriptively by comparing assessment findings, interventions, and outcomes with current evidence-based literature and midwifery care standards. Ethical principles were maintained by protecting patient confidentiality and anonymizing all personal identifiers used in the case presentation.

RESULTS

The infant was born at 38 weeks of gestation with a birth weight of 2500 g and body length of 44 cm. Physical examination revealed generalized shortening of both upper and lower extremities, consistent with the diagnosis of micromelia. The infant also presented with tachypnea (62 breaths/minute), mild chest retraction, and a weak sucking reflex. These findings indicate the presence of skeletal abnormalities accompanied by mild respiratory and feeding difficulties during the neonatal adaptation period.

The clinical manifestations observed in this case are consistent with previous studies reporting that micromelia is characterized by severe shortening of the limbs resulting from abnormal fetal bone development. Krakow and Rimoin reported that infants with skeletal dysplasia frequently present with disproportionate body measurements and limb shortening evident at birth. Similar findings were identified in the present case, supporting the diagnosis of micromelia and emphasizing the importance of careful neonatal physical examination for early recognition of congenital skeletal disorders.

Respiratory assessment demonstrated an increased respiratory rate and mild chest retraction.

These findings may be associated with thoracic involvement, which has been described as a common complication in severe skeletal dysplasia. Rao et al. reported that thoracic hypoplasia may contribute to respiratory compromise due to restricted lung development. Although the infant in this study experienced only mild respiratory distress, continuous monitoring was necessary because respiratory complications remain one of the leading causes of morbidity and mortality among newborns with skeletal dysplasia.

The findings of this study are consistent with previous evidence emphasizing the importance of multidisciplinary management for infants with skeletal dysplasia. Collaboration between midwives, pediatricians, genetic specialists, and rehabilitation professionals has been shown to improve clinical outcomes and facilitate long-term care planning. However, unlike studies conducted in tertiary referral hospitals with access to advanced genetic testing, the present case was managed in a primary healthcare setting where diagnostic resources were limited. This difference highlights the crucial role of midwives in early detection, timely referral, and family-centered care.

Format for Table and Image:

Table 1. Clinical Characteristics of the Infant with Micromelia

Mother's Age	28 years
Gravida/Parity	G1P0A0
Gestation Age	38 weeks
Mode of Delivery	Spontaneous vaginal delivery
Sex of Newborn	Female
Birth Weight	2500 g
Body Length	44 cm
Heart Rate	145 beats/minutes
Respiratory Rate	62 breaths/minutes
Body Temperature	36,4°C
Clinical Findings	Generalized shortening of upper and lower extremities (micromelia) Mild chest wall
Respiratory Condition	retractions and tachypnea
Feeding ability	Weak sucking reflex and limited breastfeeding intake
Midwifery Diagnosis	Newborn with micromelia accompanied by mild respiratory distress
Management Provided	Airway maintenance, respiratory monitoring, thermoregulation, breastfeeding support, parental education, and referral preparation
Outcome	Stable condition, parents understood the referral plan and agreed to further management

CONCLUSIONS

This study demonstrated that comprehensive midwifery care can be effectively implemented in an infant diagnosed with micromelia through the application of the Varney midwifery management approach. The care process included systematic assessment, early identification of clinical problems, continuous monitoring of respiratory and physiological status, breastfeeding support, parental education, and timely referral for further evaluation. The implementation of these interventions contributed to maintaining the infant's clinical stability and improving family understanding of the condition.

The findings indicate that comprehensive and evidence-based midwifery management plays a crucial role in the early detection and management of congenital skeletal abnormalities, particularly in resource-limited healthcare settings. Multidisciplinary collaboration and an effective referral system are essential to optimize neonatal outcomes and ensure continuity of care. Future studies involving larger sample sizes, long-term follow-up, and advanced diagnostic investigations such as genetic testing are recommended to strengthen the evidence regarding the management and prognosis of infants with micromelia.

ACKNOWLEDGEMENT

The authors would like to express their sincere gratitude to the Midwifery Study Program, Faculty of Health Sciences, Sekolah Tinggi Ilmu Kesehatan Mitra Husada Medan, academic supervisors, healthcare professionals involved in patient care, and all parties who provided guidance, support, and valuable contributions throughout the preparation of this manuscript. Special appreciation is extended to the patient's family for their cooperation and willingness to participate in this case study.

CONFLICT OF INTEREST AND FUNDING DISCLOSURE

The authors declare that there is no conflict of interest related to the publication of this article. This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

AUTHOR CONTRIBUTIONS

YN (Yosefin Nainggolan): conceptualization, data curation, investigation, methodology, project administration, formal analysis, validation, visualization, writing original draft, and writing review & editing, T(Tina): supervision, methodology, EN (Ernawati Nasution): validation, resources, writing review & editing, AZ (Agusmawati Zega): supervision, methodology, validation, NSN (Nazibah Saridatun Nahla): formal analysis, writing review & editing.

REFERENCES

- American Academy of Pediatrics. *Guidelines for Neonatal Assessment and Management of Congenital Anomalies* (2024). *Pediatrics*, 154, e20240678.
- American College of Obstetricians and Gynecologists. *Screening and Diagnostic Testing for Genetic Disorders During Pregnancy* (2024). *Obstetrics & Gynecology*, 143, e1–e18.
- Crane, H. M., Schindewolf, E., Burrill, N., et al. *The Reliability of Ultrasound Markers in Identifying Fetuses With a Life-Limiting Skeletal Dysplasia* (2024). *Prenatal Diagnosis*, 44, 1318–1326. <https://doi.org/10.1002/pd.6638>
- Indonesian Midwives Association. *Evidence-Based Midwifery Practice in Maternal and Neonatal Care* (2023). Jakarta: IBI Press.
- International Federation of Gynecology and Obstetrics. *FIGO Good Clinical Practice Advice on Prenatal Diagnosis of Congenital Anomalies* (2024). *International Journal of Gynecology and Obstetrics*, 166, 15–27.
- International Society of Ultrasound in Obstetrics and Gynecology (ISUOG). *Practice Guidelines: Performance of the Routine Mid-Trimester Fetal Ultrasound Scan* (2024). *Ultrasound in Obstetrics & Gynecology*, 63, 111–128.

- Kementerian Kesehatan Republik Indonesia. *Pedoman Pelayanan Antenatal Terpadu Edisi Terbaru* (2023). Jakarta: Kementerian Kesehatan RI.
- Kementerian Kesehatan Republik Indonesia. *Pedoman Pelayanan Neonatal Esensial* (2023). Jakarta: Kementerian Kesehatan RI.
- Kementerian Kesehatan Republik Indonesia. *Profil Kesehatan Indonesia Tahun 2024* (2025). Jakarta: Kementerian Kesehatan RI.
- Kline-Fath, B. M. *Fetal Skeletal Dysplasia*. (2024). *Magnetic Resonance Imaging Clinics of North America*, 32, 497–511. <https://doi.org/10.1016/j.mric.2024.02.009>
- Lahel, R. S., Kumar, S., & Mishra, R. K. *Thanatophoric Dysplasia: Rare Fatal Skeletal Dysplasia Detected on Prenatal Ultrasound* (2024). *Journal of Medical Ultrasound*, 32, 341–344. https://doi.org/10.4103/jmu.jmu_20_23
- Li, L., Jin, X., Liu, S., & Fan, H. *Prenatal Ultrasound Findings and Prenatal Diagnosis of Fetal Skeletal Dysplasia* (2024). *Journal of Clinical Ultrasound*, 52, 575–587. <https://doi.org/10.1002/jcu.23673>
- Liu, W., Cao, J., Shi, X., et al. *Genetic Testing and Diagnostic Strategies of Fetal Skeletal Dysplasia: A Preliminary Study in Wuhan, China* (2023). *Orphanet Journal of Rare Diseases*, 18, 336. <https://doi.org/10.1186/s13023-023-02955-4>
- Nishimura, G., Handa, A., Miyazaki, O., et al. *Prenatal Diagnosis of Bone Dysplasias* (2023). *British Journal of Radiology*, 96, 20221025. <https://doi.org/10.1259/bjr.20221025>
- Prawirohardjo, S. *Ilmu Kebidanan* (Edisi ke-5) (2024). Jakarta: PT Bina Pustaka Sarwono Prawirohardjo.
- Royal College of Obstetricians and Gynaecologists. *Prenatal Diagnosis and Management of Fetal Abnormalities* (2024). London: RCOG Green-top Guideline.
- Varney, H., Kriebs, J. M., & Geger, C. L. *Varney's Midwifery* (7th ed.) (2023). Burlington: Jones & Bartlett Learning.
- World Health Organization. *Birth Defects* (2024). Geneva: World Health Organization.
- World Health Organization. *Standards for Improving Quality of Care for Small and Sick Newborns in Health Facilities* (2024). Geneva: World Health Organization.
- Xue, H., Yu, A., Zhao, W., et al. *Prenatal Diagnosis of Fetal Skeletal Anomalies via Whole-Exome Sequencing in a Tertiary Referral Center* (2024). *Scientific Reports*, 14, 27371. <https://doi.org/10.1038/s41598-024-75738-x>