



LAPORAN KASUS—CASE REPORT

Baby Born with Multiple Congenital Anomalies Including Ruptured Omphalocele at a Primary Healthcare

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Abstract

A baby was born at a primary healthcare with multiple congenital anomalies, which included ruptured omphalocele, cloacal exstrophy, and congenital talipes equinovarus. Intra Venous line was obtained, and the baby was stabilized. The baby was referred to a tertiary health care and underwent surgery to close the defect, ileostomy, and mucous fistula. He survived the surgery but is currently in intensive care.

Keywords: multiple congenital anomalies, baby born , primary health care

INTRODUCTION

Around 20 to 30 percent of infants born with birth defects will also have multiple congenital anomalies.¹ These infants have significantly poor prognosis and quality of life, and so far there are no fixed criteria on when a treatment is called futile.² The mortality itself is relatively high, comprising of 18.9% of all infant deaths.³ A study in Denmark found the prevalence of 19.7 per 10,000 births. The prevalence is shown to be increased as more screening is done, reaching 21.6 per 10,000 births after the implementation of nationwide prenatal screening.⁴ Therefore, it is extremely important for clinicians, especially in primary healthcare, to correctly diagnose and promptly refer any infant with multiple congenital anomalies to a higher care unit.

CASE REPORT

A baby was born prematurely at 35 weeks from a multigravida mother. The mother was a 35-year-old woman from Lombok, Indonesia. This was her third pregnancy and her previous pregnancies were uneventful. She went to a

primary healthcare at her village and was treated by a doctor. The delivery was spontaneous but upon inspection, it was found that the baby's intestinal organs were protruding outside from the abdomen, signifying a condition called gastroschisis. The baby's feet were also flexed abnormally in a congenital talipes equinovarus fashion. The baby had no anal opening and there was no genital organ, which signifies cloacal exstrophy. The baby weighed 2,400 grams at 48 centimeters height. After obtaining an IV line and stabilizing the baby's general condition, they were referred to West Nusa Tenggara General Hospital.

| Test | Date | | | | |
|-----------|----------------|----------------|----------------|----------------|----------------|
| | 02/24 /2022 | 02/25 /2022 | 02/26 /2022 | 03/02 /2022 | 03/12 /2022 |
| Blood Gas | | | | | |
| pH | | 7.38 | 7.28 | | |
| pCO2 | | 28.0 | 34.0 | | |
| pO2 | | 55.0 | 80.0 | | |
| HCO3 | | 16.3 | 16.1 | | |
| BE | | -7.8 | -9.7 | | |
| SatO2 | | 88 | 95.0 | | |
| Na | 126 | 137 | 135 | | |

| | | | | | |
|------------------------|---------|--------|------|---------|---------|
| K | 5.4 | 5.2 | 3.5 | | |
| Ca | | 0.50 | 0.23 | | |
| Hematology | | | | | |
| Hemoglobin | 13.2 | 8.6 | | 16.7 | 13.5 |
| Hematocrit | 41 | 25 | | 48 | 39 |
| Leukocyte | 13120 | 4510 | | 12860 | 11780 |
| Thrombocyte | 257,000 | 98,000 | | 261,000 | 227,000 |
| CRP | <5 | 6 | | <5 | <5 |
| Blood Culture | | | | (-) | |
| Blood Chemistry | | | | | |
| Total bilirubin | | | | 2.4 | 1.4 |
| Indirect bilirubin | | | | 0.50 | 0.40 |
| Albumin | 3.5 | | | | |
| Blood Glucose | 310 | | | | |



Upon arrival, the patient's condition worsened. There were cyanosis around the lips and nose. There was also use of accessory muscles when breathing. It was suspected that the patient had respiratory distress in relation to early onset sepsis. The patient was given Continuous Positive Airway Pressure (CPAP) and orogastric tube (OGT). Blood tests were drawn, and antibiotics were administered. The patient was prepared for emergency surgery. After consulting with several departments, the patient underwent ileostomy, closing of defect, and mucous fistula. The surgery was performed uneventfully, and the patient was brought to a recovery room.

DISCUSSION

In a 2014 study in Europe, the prevalence of multiple congenital anomalies had reached 15.8 per 10,000 live births.⁵ Another study conducted in Denmark in 2020 found a higher prevalence of 21.6 per 10,000 births. This might be due to the implementation of nationwide prenatal screening.⁴ A similar case to ours was a patient with OEIS complex, which is a series of several birth defects



comprising Omphalocele, Exstrophy of cloaca, Imperforate anus, and Spinal defect. Other similar characteristic is the absence of previous congenital defect in their siblings. This patient needed multiple surgeries as he grew up and even then his quality of life is very low. No etiology is currently known for this type of defect. 6 Another similar case was also found with OEIS complex but was accompanied by body stalk anomaly. This patient died within 1 hour postnatal. 7 The following criteria have been described in the literature for prenatal diagnosis of cloacal exstrophy: (a) Major criteria – non-visualization of the fetal bladder, infra-umbilical anterior abdominal wall defect, omphalocele, and myelomeningocele. Lower extremity deformities (club foot), renal anomalies, ascites, expanded pubic arches, thin thorax, kyphoscoliosis, hydrocephalous, and single umbilical artery are among the minor requirements. Despite these criteria, the entire amount of abnormalities is not always detectable prenatally. 8

4. Toxværd ME, Garne E. Epidemiology of Multiple Congenital Anomalies Before and After Implementation of a Nationwide Prenatal Screening Program in Denmark. *Front Pediatr.* 2021;9:614864.
5. Calzolari E, Barisic I, Loane M, Morris J, Wellesley D, Dolk H, et al. Epidemiology of multiple congenital anomalies in Europe: a EUROCAT population-based registry study. *Birth Defects Res A Clin Mol Teratol.* 2014 Apr;100(4):270–6.
6. Neel N, Tarabay MS. Omphalocele, exstrophy of cloaca, imperforate anus, and spinal defect complex, multiple major reconstructive surgeries needed. *Urol Ann.* 2018 Mar;10(1):118–21.
7. Mandrekar SRS, Amoncar S, Banaulikar S, Sawant V, Pinto RGW. Omphalocele, exstrophy of cloaca, imperforate anus and spinal defect (OEIS Complex) with overlapping features of body stalk anomaly (limb body wall complex). *Indian J Hum Genet.* 2014 Apr;20(2):195–8.
8. Keppler-Noreuil K, Gorton S, Foo F, Yankowitz J, Keegan C. Prenatal ascertainment of OEIS complex/cloacal exstrophy - 15 new cases and literature review. *Am J Med Genet A.* 2007 Sep 15;143A(18):2122–8.

LEARNING POINTS

- Around 20% to 30% of babies born with birth defects will have multiple congenital anomalies.

- It is critical for doctors, particularly in primary care, to appropriately diagnose and refer any newborn with multiple congenital abnormalities to a higher care unit as soon as possible.

REFERENCES

1. Agopian AJ, Evans JA, Lupo PJ. Analytic Methods for Evaluating Patterns of Multiple Congenital Anomalies in Birth Defect Registries: Analysis of Multiple Congenital Anomaly Patterns. *Birth Defects Research.* 2018 Jan 15;110(1):5–11.
2. Pastura PSVC, Land MGP. CHILDREN WITH MULTIPLE CONGENITAL DEFECTS: WHAT ARE THE LIMITS BETWEEN THERAPEUTIC OBSTINACY AND THE TREATMENT OF UNCERTAIN BENEFIT? *Rev Paul Pediatr.* 2017 Mar;35(1):110–4.
3. Kim SB, Jang MJ, Song YH, Jung SY, Oh JS, Lim JW. Trends and Characteristics of Mortality Associated with Congenital Anomalies in Korean Children under 5 Years of Age. *Neonatal Med.* 2021 Aug 31;28(3):99–107.