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CASE REPORT

Newborn with Multiple Congenital Anomalies (MCAs)

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Abstract

Congenital anomalies are one of the main causes of physical disabilities, stillbirths and neonatal deaths. It Can contribute to long-term disability, which may have significant impacts on individuals, families, health-care systems and societies. Diagnosis of a child who presents with multiple congenital anomalies both prenatal and postnatal is still a complex issue. We present a complex case of a newborn with multiple congenital anomalies (MCAs) including hydrocephalus, Down syndrome, phocomelia, ectrodactyl and ambiguous genitalia. This is the first case to be reported in Tanzania and among the few cases in World's English literatures.

Keywords: Congenital, Hydrocephalus, Down syndrome, Phocomelia, Ectrodactyl, Ambiguous genitalia

1 | INTRODUCTION

Multiple congenital anomalies (MCAs) are defined as two or more unrelated major structural malformations that cannot be explained by an underlying syndrome or sequence[1]. Approximately 3 million fetuses and infants are born each year with major malformations[2].An estimated 303 000 newborns die within 4 weeks of birth every year, worldwide, due to congenital anomalies[3]. The exact etiology of MCA is unidentified but genetic, infectious, nutritional, and environmental causes are accused[2,3].

2 | CASE PRESENTATION

A 24 years old, Gravida 3 para 0, women not in labour presented to the Obstetrics and Gynecology department at $28+^{4}$ weeks of pregnancy. She has history of abortion in previous 1^{st} and 2^{nd} pregnancies due to cervical incompetence at 8 weeks and 12 weeks respectively. The cervical cerclage was done in the past 5 months, no history of medication use or illness, nor family history of congenital anomalies neither known exposure to teratogens.

During admission she was complaining of not feeling the fetal movement in the last 5 hours and PV leakage. In the prenatal check up after ultrasound

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scan the parents were informed that; non viable fetus has abnormalities (hydrocephalus and phocomelia). The cervical cerclage was removed and labour induction was done per intrauterine fetal death (IUFD) hospital protocol.

A Fresh still born baby (FSB) through assisted breech delivery was born without any complications. Postnatal examination of newborn revealed multiple congenital anomalies including hydrocephalus, Down syndrome, phocomelia, ectrodactyl, ambigous genitalia and some signs of Klipel-Feil syndrome. Further investigations of internal organs, genetic and chromosomal investigations were not done due to limitations of resources at the hospital and family financial constraints.

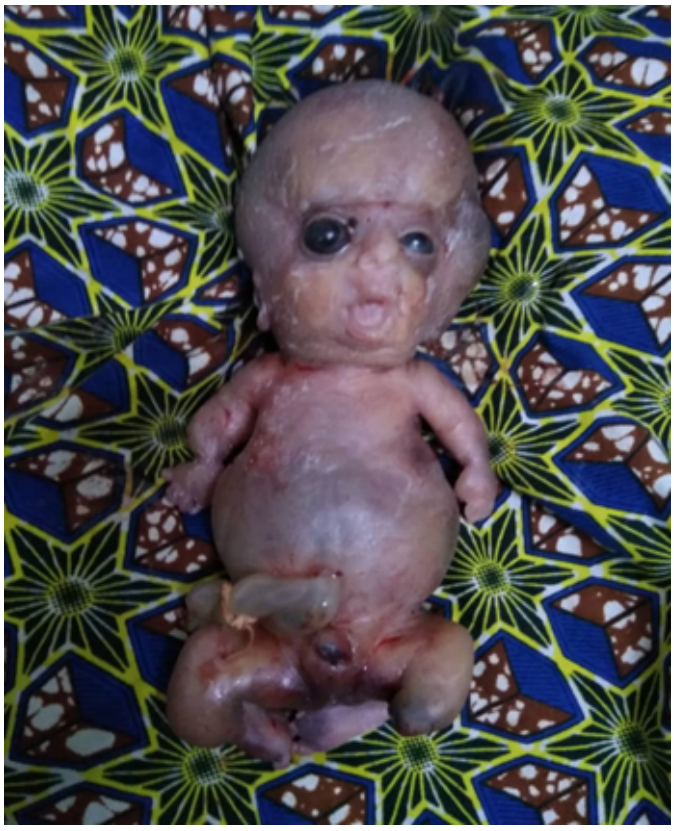

FIGURE 1: Clinical picture of a newborn front view

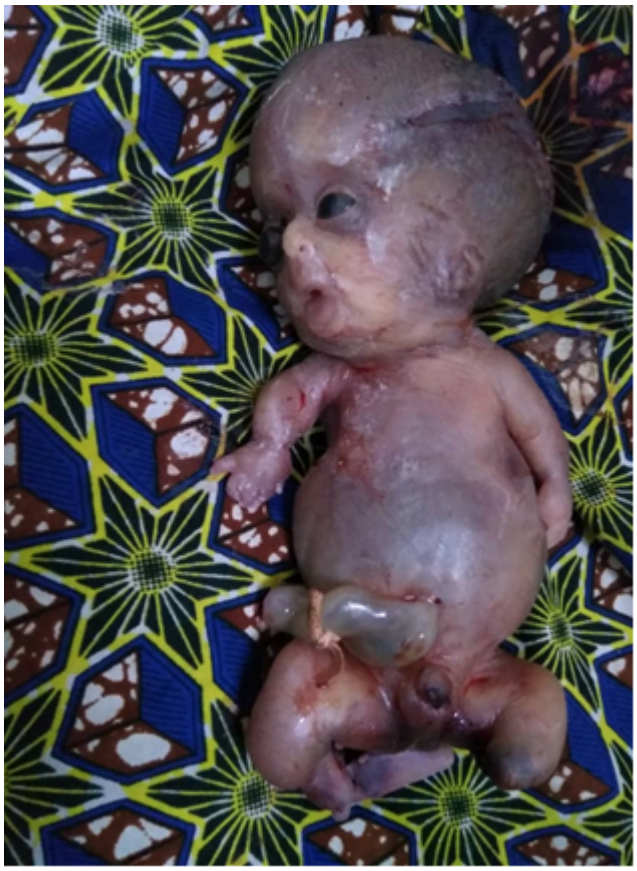

FIGURE 2: Clinical picture of a newborn side view

3 | DISCUSSION

The causes of congenital malformations are divided into four broad categories, genetics, environmental, multifactorial and unknown [2]. 50% of congenital malformation are linked to unknown causes, this case is inclusive [3].The birth of an infant with MCAs generates difficult multiple medical, surgical, ethical, psychosocial, and physical issues for patients and parents. Because MCA is rare and few cases has been reported, no much information available[10]. The management need supportive treatment with multidisplinary team of neonatologists, geneticist, cardiologists, radiologists, physical therapist and psychologist[10].

The world Health organization recommends introduction of surveillance program and teams for follow up of preventive public health measures work to decrease frequency of MCAs through elimination of risk factors or reinforcement of protective factors and adoption of preconception and periconception medical genetic screening and counseling[3].

TABLE 1: Physical description of diagnosed syndromes

Birth defect	Description and features
Hydro-cephalus	Is an abnormal buildup of cerebrospinal fluid (CSF) in the ventricles of the brain[4]. In infants the most obvious sign of hydrocephalus is usually an abnormally large head [4].
Down syn-drome	Down's syndrome is a genetic disorder caused when abnormal cell division results in extra genetic material from chromosome 21[5]. A flattened face, especially the bridge of the nose, Almond-shaped eyes that slant up, A short neck, Small ears, A tongue that tends to stick out of the mouth, Tiny white spots on the iris (colored part) of the eye, Small hands and feet, A single line across the palm of the hand (palmar crease),Small pinky fingers that sometimes curve toward the thumb, Poor muscle tone or loose joints [5].
Pho-comelia	Phocomelia syndrome (PS) is a rare birth defect that causes severe birth defects, especially of the upper limbs [6].The bones of the arms, and in some cases other appendages, may be extremely shortened and even absent. The fingers of the hands may be fused. An extreme case results in the absence of the upper bones of both the arms and legs so that the hands and feet appear attached directly to the body[6].This disorder, PS, may be genetically transmitted within families as an autosomal recessive trait or may be the result of spontaneous (sporadic) changes in the gene[6].
Ectro-dactyl	Involves the deficiency or absence of one or more central digits of the hand or foot and is also known as split hand/split foot malformation (SHFM) [7].The incidence of cleft hand varies from 1 in 90,000 to 1 in 10,000 births depending on the used classification. Cleft hand can appear unilateral or bilateral [8].
Ambigu-ous genitalia	Ambiguous genitalia is a rare condition in which an infant's external genitals don't appear to be clearly either male or female [9]. In a baby with ambiguous genitalia, the genitals may be incompletely developed or the baby may have characteristics of both sexes [9]. The external sex organs may not match the internal sex organs or genetic sex[9].

4 | CONCLUSION

This case is extremely rare and few information about management are available in the literature, psychological counseling to the parents was done successfully in the present case. However, Early and precise diagnosis of a child with multiple congenital anomalies is important for management, genetic counseling concerning etiology, recurrence risk, prenatal diagnosis, screening and recommendation for evaluation of other family members. (1–10)

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