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PUBLIC HEALTH

PHECOMELIA

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ABSTRACT

Purpose: Phocomelia is a congenital disorder characterized by the abnormal development of the limbs, specifically the upper limbs, during fetal development. In this condition, the upper limbs are either shortened or completely absent, and the hands may be attached to the shoulders or chest. Similarly, the lower limbs may be shortened or malformed, with the feet attached directly to the thigh bones. This study presents an epidemiologic analysis of phocomelia case in Kenya.

Problem: Phocomelia can be caused by a number of factors, including genetic mutations, chromosomal abnormalities, and exposure to certain teratogenic agents such as thalidomide. The disorder is considered to be rare, with only a few cases reported worldwide.

Research methodology; The study used a case report from Kericho county, Kenya.

Findings and discussions: The disease vary in degree of severity. It manifests with near normal hand or foot, hands or feet directly attached to trunk, absent or underdeveloped bones of hands and legs e.g humerus/femur/radius/tibia/ulnar fibula. The prevalence of these findings was lower than in previous studies, which is likely due to the study's careful selection of only true phocomelia and not amelia or other limb truncation. Due to its infrequency, lack of public awareness, and lack of adequate technology to assist in managing this condition, this condition receives less attention than other disorders such as cerebral palsy or Down syndrome.

Conclusion: Phocomelia is a rare disease for which no cause has been identified or documented. The cause is thought to be the use of thalidomide, a drug used to treat anxiety and Leprosy.

Recommendations: The most important management of a child born with Phocomelia is to assess for other associated abnormalities, especially of the heart and intestines, and address them promptly and appropriately. Pediatric cardiologists and gastroenterologists should examine the child and recommend a treatment plan for any abnormalities encountered.

Keywords: *Phocomelia, Case report, Etiology, Epidemiology, Pathophysiology*

INTRODUCTION

Phocomelia is a congenital disorder characterized by the abnormal development of the limbs, specifically the upper limbs, during fetal development. In this condition, the upper limbs are either shortened or completely absent, and the hands may be attached to the shoulders or chest. Similarly, the lower limbs may be shortened or malformed, with the feet attached directly to the thigh bones. Additionally, Phocomelia also associated with other malformations such as abnormalities in the spine and ribcage, and some visceral malformations. Phocomelia can be caused by a number of factors, including genetic mutations, chromosomal abnormalities, and exposure to certain teratogenic agents such as thalidomide. The disorder is considered to be rare, with only a few cases reported worldwide. There is currently no cure for Phocomelia, but the treatment options may include prosthetics, physical therapy, and surgery to improve mobility and function. The incidence rate is estimated 4.2per 1 million births. Phecomelia is a condition famously associated with a teratogenic side effects of thalidomide, a drug used in management of anxiety and morning sickness.

Case report

A 34-year-old female Para 5+3 Gravida 8 a resident of Tumda village in Kapkatet, Kericho County, Kenya was admitted to the hospital on 22/9/2022 at 1 PM, with complaints of lower abdominal pains radiating to the back and associated with rupture of membranes 1 hour before admission through drainage of liquor was noted, the liquor was clear. The pain was on and off and lasted for almost two minutes. On examination the fundal height was 36 weeks with a positive fetal height of 133 beats per minute, the cervix was 8 cm dilated and delivered within one hour The patient was a Para 4+5 gravida 8, Ante Natal Clinic (ANC) registered in Kapkatet Sub –

County Hospital. She attended 3 consecutive ANC visits. She was on regular Iron and Ferrous sulphate supplements.

She never did a prenatal obstetric abdominal Ultrasound, since there was no indication of it. Her (ANC) profile indicated a Hemoglobin of 14.7 g/dl, blood group B, and rhesus positive, both VDRL and HIV test was Non-reactive. Her urinalysis test was normal, no protein nor glucose traces were found in the urine.

At around July 2022, she had white discharge, itching and pains in the vagina area that saw her put on P.O Cefixime 400mg BD for seven days, as well as P.o Paracetamol 1g TDS and 200mg vaginal pessaries note for 3 days. The patient did not give a history of sore throat, fever, cough or Covid-19 positive contact. Despite a history of drug use, there was no record of admission to the hospital or exposure to any radiation or viral infection in the patient's medical history. The first caesarean section was performed on the patient in 2007 because of breech presentation, and the second one was performed in 2018 because of a previous scar. The patient has undergone two caesarian sections.

She had menarche when she was 12 years old, and it was a regular, moderate, and painless cycle. She uses at least 2-3 pads per day. Her cycle lasts 28 days and she has no menstrual bleeding. She has lost three children to death shortly after birth in three consecutive years, in 2017, 2018, and 2019. All five other children, including the one born with Phecomelia, are alive and well. She has used traditional herbal medicine for birth control since her first pregnancy and has never changed. She does not use any of the hospital's family planning services. She has no history of thalidomide use, nor does she have Leprosy or any other condition that could have required her to use the thalidomide drug.

Phocomelia

Def: Phocomelia is a rare congenital defect characterized by absence of intermediate segment of extremity with only occasionally the hands or feet being directly attached to the trunk.

Etiology

It is a rare defect however, it was largely associated with drug Thalidomide used to treat anxiety and morning sickness in 1960. Thalidomide -induced phocomelia may arise in underdeveloped countries where it is still used to treat leprosy. Apart from causing phocomelia, thalidomide may also cause facial problems -irregular teeth number/spacing, small jaws, cleft palate/cleft lipid/small noses (Sethi S, Satia M, Ram S, kashyap P (2021)).

Symptoms

The symptoms may vary in degree of severity. It manifests with near normal hand or foot, hands or feet directly attached to trunk, absent or underdeveloped bones of hands and legs e.g humerus/femur/radius/tibia/ulnar fibula.

Types

Based on the absent portion of limb relative to the trunk, phocomelia can be classified into 3 categories

(1) complete phocomelia with absence of all limb bones proximal to the hand, (2) absence or extreme hypoplasia of proximal limb bones with forearm and hand attached to the trunk, and (3) hand attached directly to the humerus (Patel, Kreuzer *et al*. 2021).

Causes of phocomelia

The cause of phocomelia is uncertain. Little is known about the epidemiology and the causes of most cases of phocomelia today are still to be determined (Bermejo-Sánchez, 2011). However, it has been documented to have a direct link with thalidomide use because it prevents formation of new blood vessels in the limb, induction of cell death, encouraging free radicals in limb tissue, reduce or block growth factors in limb growth and also cause anomalous origin of subclavian artery resulting in disrupted vascular supply to other associated conditions. Genetics and Sporadic -Recessive trait/spontaneous mutation has been associated with development of phocomelia

Epidemiology

According to Bermejo-Sanchez et all (2011), the incidence and presentation of phocomelia were examined in 22,740,933 live births in an effort to assess the prevalence of true phocomelia. Reports indicate that true phocomelia occurs in 0.62 live births per every 100,000 patients. Approximately half (53.2%) of the cases displayed isolated phocomelia, while 36.9% had additional major congenital abnormalities, and 9.9% of cases correlated with a clinical syndrome. The data shows that cases involving only one limb is at 55.9%, while those involving two limbs are at 40.2%. Only four cases of 141 had involvement of all four.

When comparing single upper extremities deficiencies, the left side was more commonly affected (64.9%) than the right. When two limbs were involved, the upper limbs were involved 58.5% of the time compared to the lower limbs. The incidence of these findings was lower than what prior studies had reported, which is likely due to the care taken by this study, only to consider true phocomelia and not amelia or other limb truncation. This condition receives less attention than other disorders such as cerebral palsy or down syndrome due to its infrequent occurrence, lack of public awareness, and a lack of adequate technology to assist in managing this condition

Pathophysiology

In extremity development, the apical ectodermal ridge forms at the most distal end of the limb bud. This apical ectodermal ridge through its interaction with the underlying progress zone mesoderm subsequently determines appropriate longitudinal growth of the extremity, as well as differentiation of distal and proximal structures of the limb bud. Cell death due to apoptosis from any cause such as vascular insufficiency or drug toxicity that interrupts this relationship between the apical ectodermal ridge and the progress zone can result in phocomelia: (Sethi, et al., 2021).

Treatment / management

The most important management of a child born with Phocomelia is to assess for other associated abnormalities, especially of the heart and intestines, and address them promptly and appropriately. Pediatric cardiologists and gastroenterologists should examine the child and recommend a treatment plan for any abnormalities encountered.

From the perspective of the hyperplastic limb, prosthetics may be of some use, but many without concomitant pathology can function quite well. Families of children who have severely hyperplastic extremities should work with therapists to help them understand how to meet the unique needs of their children. And finally, caring for a disabled child can be traumatic for some people. Therefore, physicians should have a low threshold to recommend mental counselling services to assist in coping mechanisms that may be needed by both the parents and the child. Psychotherapy for parents Genetic Counselling Surgery; Stabilizing joints, bone lengthening, improving thumb apposition.

Other associated conditions

Roberts Syndrome: A rare autosomal recessive disorder caused by a mutation in the *ESCO2* gene that manifests with severe limb malformations and craniofacial defects

Thrombocytopenia with radial aplasia (TAR): A rare autosomal recessive disorder related to the *RBM8A* gene that presents with thrombocytopenia and limb radial deficiency.

Syndrome of severe limb defects, vertebral hyper segmentation, and mirror polydactyly: An autosomal recessive disorder resulting in severe limb hypoplasia with polydactyly and hyper segmentation of the spine.

The other associated conditions categorized Phocomelia into; Sporadic Phocomelia, Holt-Hormam Syndrome, DK Phocomelia Syndrome, Schnitzel-Type Phocomelia, and CHILD Syndrome (Aboud, Kadhim *et al*. 2022

Complications

Children with phocomelia have a high stillbirth rate compared to the general population. Additionally, children with severe limb hypoplasia may or will face problems with activities of daily life and mobility. For this reason, health care teams must be integrated to provide for more than just the medical challenges that present with phocomelia.

Deterrence and patient education

Increased drug regulatory practices across the world have significantly decreased the incidence of thalidomide-induced limb hypoplasia. Thalidomide has utility in the treatment of insomnia, anxiety, and even in the treatment of leprosy, but understanding the risks of thalidomide exposure during pregnancy is critical for any woman taking the medication. Patients and prescribes alike should understand the deleterious effects of thalidomide when taken in early pregnancy.

Enhancing Health outcomes

The dispute surrounding phocomelia and thalidomide was an important milestone in the progression of the regulatory procedures of pharmacology. The avoidance of prescribing dangerous drugs that might induce limb truncation is the most essential obligation that falls on the shoulders of the medical community about phocomelia. This can be accomplished by cautious

prescription procedures and comprehensive patient education. A multidisciplinary team of doctors is needed to assess infants born with limb hypoplasia, such as is observed in phocomelia, to check for accompanying abnormalities that may be deadly. This is because 50% of the children with phocomelia and amelia have associated problems.

Because many of the associated abnormalities of the heart, vertebrae, or other essential organs may pose a danger to the patient's life, these anomalies should be treated as soon as possible by contacting specialists. If surgical intervention is necessary, a dedicated operating room team, consisting of a nurse, scrub technician, and surgical assistants, must be well-versed in any operational plan and operate effectively as a team to maximize positive results for patients. A child who is growing but has difficulties may finally operate at a higher level with the assistance of a prosthetist and a therapist. Families that are responsible for the care of disabled children sometimes face significant financial and emotional burdens. Therefore, adequate mental health therapy and social work may be required to provide care not just for the patient but also for the patient's family members.

CONCLUSION

Phocomelia is a rare disease whose cause has not been either documented or determined. The cause is attributed to the use of thalidomide, the drug that is used to treat anxiety and Leprosy. It is hoped that this paper will contribute to the knowledge.

RECOMMENDATIONS

The most important management of a child born with Phocomelia is to assess for other associated abnormalities, especially of the heart and intestines, and address them promptly and appropriately. Pediatric cardiologists and gastroenterologists should examine the child and recommend a treatment plan for any abnormalities encountered. Families of children who have severely hyperplastic extremities should work with therapists to help them understand how to meet the unique needs of their children. Physicians should have a low threshold to recommend mental counselling services to assist in coping mechanisms that may be needed by both the parents and the child.

REFERENCES

Aboud, M. J., Kadhim, S. M., Abudi, N. M., Joudi, H. M., & Joudi, Z. M. (2022). Congenital limbs deficiency versus phocomelia the hard work up with the subtleties and malformations accompanying the syndromes images: Case series. *International Journal of Medical Reviews and Case Reports*, *6*(13), 65-65.

Bermejo-Sánchez, E., Cuevas, L., Amar, E., Bianca, S., Bianchi, F., Botto, L. D., ... & Martínez-Frías, M. L. (2011, November). Phocomelia: a worldwide descriptive epidemiologic study in a large series of cases from the International Clearinghouse for Birth Defects Surveillance and Research, and overview of the literature. In *American Journal of Medical Genetics Part C: Seminars in Medical Genetics* (Vol. 157, No. 4, pp. 305-320). Hoboken: Wiley Subscription Services, Inc., A Wiley Company.

Patel, A. H., Kreuzer, S. W., & Sherman, W. F. (2021). Bilateral Total Hip Arthroplasty in the Setting of Developmental Dysplasia of the Hip and Extreme Hip Flexion Requirements due to Phocomelia. *Arthroplasty today*, *8*, 262-267.

Sethi S, Satia M, Ram S, kashyap P. (2021). Medical termination of pregnancy with phocomelia. Southeast Asian J Health Prof 2021;4(1):15-18.