Case Report ISSN 2689-1069

Clinical Reviews & Cases

A Rare Case of Apert Syndrome in a Tanzanian Newborn: Diagnosis and Management Challenges in a National Tertiary Hospital

Raymond Leiya[1], Martine M Sosoma[1], Peter Msinde[1], Jackline zambi[1] and Mwanaidi A Msuya[2]

[1]*Department of ENT-Muhimbili National Hospital, Mloganzila.*

[2]*Department of Paediatrics and Child Health- Muhimbili National Hospital, Mloganzila.*

***Correspondence:**
Dr. Raymond Leiya, Department of ENT-Muhimbili National Hospital, Mloganzila

Received: 12 Mar 2025; **Accepted:** 10 Apr 2025; **Published:** 21 Apr 2025

Citation: Raymond Leiya, Martine M Sosoma, Peter Msinde, et al. A Rare Case of Apert Syndrome in a Tanzanian Newborn: Diagnosis and Management Challenges in a National Tertiary Hospital. Clin Rev Cases. 2025; 7(1): 1-4.

ABSTRACT

Introduction: *Apert syndrome is a rare congenital condition characterized by coronal craniosynostosis, ex-orbitism, midface hypoplasia as well as symmetric syndactyly of both feet and hands. This condition is also known as acrocephalosyndactyly type 1. Apert syndrome is linked by autosomal dominant inheritance to fibroblast growth factors receptor gene alteration.*

Case Report: *This is the case report of a 42 days old baby who presented with classical facial features of Apert syndrome. Syndactyly of both fingers and toes. He was born from non-consanguineous parents, his father being 44 years old when the baby was born. An X-ray of the skull revealed increase in anterior posterior diameter of the skull, while cranial ultra sound depicted no evidence of space occupying lesions, no evidence of extra axial collection with no signs of midline shift. X-ray of hands and feet revealed the pathognomonic syndactyly of the disease.*

Conclusion: *There is a paucity of data about Apert Syndrome in African countries. Which poses the emphasis on the advocacy of this condition in African countries for more cases to be reported to capacitate the healthcare workers to diagnose and treat this rare condition properly. Given the limitations of genetic testing and prenatal screening in the region, this case call for action for the need to increase awareness, early intervention and genetic counseling to guide treatment decisions and improve outcomes.*

Keywords

Apert syndrome, Syndactyly, Craniosynostosis, Acrocephalo-syndactyly.

Introduction

Apert syndrome was first introduced by scientists named Baumgartner in 1842 and then by Wheaton in 1894, thereafter in 1906 a French pediatrician called Eugene Apert published 9 cases of this disease [1,2]. The case report named "De l'acrocephalosyndactylie" this condition was later named Apert syndrome in honor of Eugene Charles Apert [3].

Apert syndrome is characterized by coronal craniosynostosis, ex-orbitism, midface hypoplasia as well as symmetric syndactyly of both feet and hands. Low hairlines, webbed neck and pectus excavatum are other features of this rare condition [4,5]. Apert syndrome is differentiated with other forms of acrocephalosyndactyly by presence of fusion of both fingers and toes [6]. Also Apert syndrome is also characterized by oral features ranges from pseudo cleft, high-arched palate, transverse and sagittal maxillary hypoplasia, dental crowding, delayed dentition, ectopic teeth to disorganized teeth [6,7]. Also, Apert syndrome rarely manifests with systemic symptoms, but the patient may present with frequent ear infection, hearing loss, obstructive sleep apnea, slow intellectual development, excessive sweating, hydrocephalus and ventriculomegaly [8]. Apert syndrome is linked by autosomal dominant inheritance to fibroblast growth factors receptor gene alterations [9]. This indicate that for Apert

syndrome to become prevalent, only one parent needs to pass the mutation [6]. The disease is attributed by Advanced paternal age of greater than 35 years and familial predispositions, though most cases occur de-novo. Apert syndrome occurs in 1 out of 65,000 to 88,000 live births, with no gender and geographical or ethnicity predilections [10].

Case Report

A 42-day-old male infant was referred to Muhimbili National Hospital- Mloganzila from other facility for further evaluation of craniofacial abnormalities and limb deformities. The infant was born via emergency cesarean section due to breech presentation, with no significant antenatal complications. He was the fifth child of non-consanguineous parents, with the mother aged 39 and the father 44 years old.

The infant presented with a turribrachycephalic skull shape, bulging eyes, flat face, low-set ears. Further physical examination revealed the fusion of all digits, but on palpation a faint margin of phalangeal bones was noticed which signified a faint demarcation between the fingers. The infant's developmental milestones were within normal limits except for fine motor skills, which were impaired due to severe hand deformities.

The child was born through Caesarean section due to breech presentation. Antenatal and birth histories were un-eventful except postnatal in first week of life the baby suffered from Early Onset Neonatal Sepsis (EONS) where by the baby was de-saturating on room air and had labored breathing with use of accessory muscles, which the baby was investigated and kept on oxygen and antibiotics where the patient improved.

The immunization history was as per age requirements of the national immunization and vaccine development; On the dietary history the baby was kept on expressed breast milk with top up formula milk because the mother could not produce enough milk. No history of trauma or surgery on his fingers or toes prior to this condition. The mother denied use of un-prescribed medication during pregnancy. There is no history of radiation exposure or any complication during this pregnancy. The infant has no history of ear infection. There is no history of excessive sweating history was reported. The infant can see and hear properly. There was no any other congenital malformation reported in this patient.

A diagnosis of Craniosynostosis syndromes with symmetrical upper and lower limbs syndactyly (Apert syndrome) was established clinically based on history and physical findings with differential diagnosis of Pfeiffer syndrome and Crouzon's syndromes. Imaging studies, including skull X-ray and cranial ultrasound, confirmed the diagnosis of craniosynostosis with increased anterior- posterior skull diameter and complex syndactyly.

A full blood picture, renal and liver function test were not informative. Skull x- ray showed increased anterior posterior diameter of the skull. Also, the x-ray of upper limbs showed fusion of the phalanges suggestive of complex syndactyly. Cranial ultra sound revealed no focal mass and space occupying lesions, no extra axial collections, however- MRI of the brain revealed features of chronic focal encephalitis, mega cisterna magna and bilateral proptosis.

Discussion

Apert syndrome has an autosomal dominant inheritance pattern and the disease pathogenesis at molecular level is explained as product of mutagenesis of fibroblast growth factor receptor-2 gene specific at 10q26 [3,10]. Our country does not routinely perform specific genetic mutation analysis or prenatal ultrasound screening for congenital malformation in all levels of health care [11]. According to literature search there is no genetically confirmed case of Apert syndrome reported from Tanzania. This highlights the need for increased awareness and research on this rare condition within the country and Africa at large.

Cases with paternal mutation have been reported but most cases are said to be sporadic, where there is de-novo mutation of FGFR-2 gene [12,13]. In this case the father was 44 years old when the child was born, which aligns with majority of literatures that put advanced paternal age as one of the risk factor for Apert syndrome [10,14].

In this case the patient has most facial classic features of apert syndrome. The patient has turribrachycephalic head (tower like head), hypertelorism, flat nasal bridge, midface hypoplasia, down slanting palpebral fissure, ex-orbitism and exophthalmos. There were no any dental features in this patient.

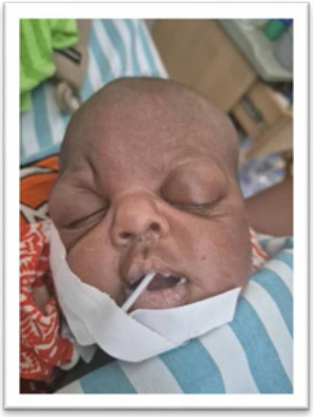

Figure 1: Frontal view of the patient's head showing hypertelorism, downward slanting outer canthus of eyes, and depressed nasal bridge.

Furthermore, the infant had history of systemic illnesses like Early onset neonatal sepsis, pneumonia and meningitis, but there was no history of recurrent ear infections. Infants with congenital disorders such as Apert's Syndrome may be at increased risk of sepsis due to factors such as impaired immune response or anatomical abnormalities [15,16].

There was bilateral symmetrical syndactyly with deformed digits. The feet of both legs and fingers of both hands have a full fusion of all digits. There have been no plan of yet to perform surgical

intervention/s due to patient's unstable clinical condition.

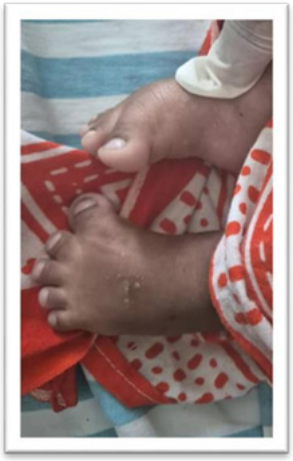

Figure 2: View of feet showing syndactyly of the toes.

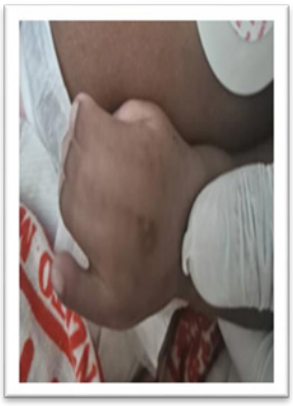

Figure 3: View of hands showing syndactyly of the fingers.

Surgery for Apert syndrome is performed to meet the child's developmental needs. Early interventions, such as the release of craniosynostosis, occur in the first months to help the development of the brain [17]. At 6 to 12 months, as orbital advancement and posterior expansion are performed to reshape the skull and protect the eyes [18]. Syndactyly release occurs at 13 months to improve hand function [19].

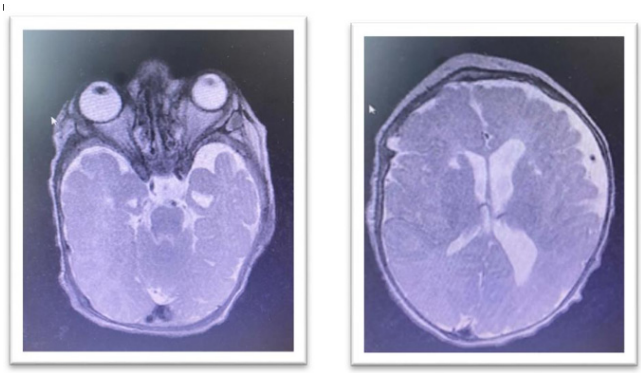

Figure 4: The MRI of the patient showing Mega cisterna magna and bilateral proptosis and chronic focal encephalitis.

Mid-face advancement and other facial corrections are performed between ages 4 and 12, and subsequent surgeries are performed during adolescence to correct facial growth and other adjustment [20]. In this case, the infant had no any features suggestive of increase intracranial pressure, therefore there have been no plans of yet to try to do surgical intervention recommended per age.

Conclusion

Apert syndrome is rarely reported in Africa, especially in the sub-Saharan region. Few cases of Apert disease have been reported in Africa. It is a devastating physical defect that can be treated with good outcome if detected early. Therefore, a multidisciplinary approach to the treatment of Apert syndrome is very important. While the infant didn't show no signs of increased intracranial pressure, which would necessitate urgent cranial surgery, timely planning for future interventions, such as craniosynostosis release and syndactyly correction, is crucial to support normal brain development and hand function.

Given the limitations of genetic testing and prenatal screening in the region, this case call for action for the need for increased awareness, early intervention and genetic counseling to guide treatment decisions and improve outcomes. Also, a serious consideration to health insurance coverage schemes should be done to patients with these rare conditions in order to improve the standard of care that can be afforded to these patients to help improve the outcomes and quality of life to these patients. Socioeconomic and expertise challenges tend to limit the quality of care provided. Hence, a multidisciplinary approach to the management of Apert Syndrome is of highly recommended.

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