ISSN: 2578-7365

Case Report

Journal of Chemistry: Education Research and Practice

Patau Syndrome: Genetic Disorder Clinically Diagnosed Under Limited Resources at Singida Regional Referral Hospital

Amedeus L Mushi*, Patrick Ng'unda, Roza Majengo, Rukia Ibrahim, Leonidas Chano

Singida Regional Referral Hospital, Tanzania; Laboratory Department, Tanzania; Pediatric Department, Tanzania

***Corresponding author**
Amedeus L Mushi,Singida Regional Referral Hospital, Tanzania; Surgery Department, Tanzania; Radiology Department, Tanzania.

Submitted: 17 Jun 2022; **Accepted**: 29 Jun 2022; **Published**: 02 Nov 2022

Citations: *Mushi, A. L., Ng'unda, P., Majengo, R., Ibrahim, R., Chano, L. (2022). Patau Syndrome: Genetic Disorder Clinically Diagnosed Under Limited Resources at Singida Regional Referral Hospital. J Chem Edu Res Prac, 6(2), 410-412.*

Abstract

Introduction: *Patau syndrome or trisomy 13 is genetic disorder clinically characterized by the presence of numerous malformations with a limited survival rate for most cases. It is characterized by the cardinal triad of orofacial clefts, microphthalmia and postaxial polydactyly of the limbs and non-cyanotic heart defects. This article will create awareness of capturing all birth defect in country so as to know the magnitude of this problem.*

Case Description: *A 6 hours baby boy delivered at gestation age 32 weeks by 36 years woman was referred from a lower health facility with diagnosis of cleft lip has several congenital anomalies.*

Discussion: *Diagnosis of patau syndrome based on the clinical manifestation, phenotypes and the genetic. This case was managed to be diagnosed by clinical manifestation and phenotype. The presence of features provides more confidence of concluding the condition as patau syndrome. Parents deny referral due to poor prognosis although the medical counseling was provided which is not genetic counseling as developed countries.*

Conclusion: *Capacity building to health practitioners in identifying and diagnosing patau syndrome should be done to all health facilities levels*

Keywords: Patau Syndrome, Trisomy 13, Cutis Aplasia, Polydactyly

Introduction

Patau syndrome is common chromosomal anomalies clinically characterized by the presence of numerous malformations with a limited survival rate for most cases [1]. The incidence rate is about 1 out of 5,000 to 20,000 birth [2-4]. Individuals with this chromosomal syndrome have a short lifespan which is the third most common autosomal chromosome trisomy clinically severe condition [5, 6]. This syndrome is caused by presence of an extra copy of chromosome 13, some time there other changes in chromosome 13, such as translocation [7]. It is s a multiple malformation syndrome and includes anomalies of the central nervous system (CNS), cardiac, and circulatory system, and the urogenital system [5, 6]. Patau syndrome is a well-recognized, multiple congenital anomaly syndrome, characterized by the cardinal triad of orofacial clefts, microphthalmia and postaxial polydactyly of the limbs and non-cyanotic heart defects [5, 8]. Cleft lip and palate are present in eighty percent of this syndrome's carriers [6]. Eighty-five percent do not survive beyond one year of life and most die before completing six month [6, 9]. About 28% of children born with trisomy 13 die during the first week of life in which median life expectancy is about 2.5 days [1]. The most common cause of death is cardiopulmonary complications [6, 7]. The diagnosis of Patau syndrome can be confirmed antenatally or after delivery by cytogenetics [5]. There is no previous reported such case in a country, so this will create awareness to healthcare workers to categorize all identified birth defects. Also, this is among the rare patau syndrome case as it is presented with cutis aplasia. This article will create awareness of capturing all birth defect in country so as to know the magnitude of this problem with will also determine the frequency of several birth defects in which for the future will be used to determine the risk factors of congenital anomalies.

Case Description

A 6 hours baby boy was referred from a lower health facility with diagnosis of cleft lip for further management. The baby was reviewed at the neonatal care unit our referral hospital. He was born

by spontaneous vertex delivery, weighed 2.6 kg and had an APGAR SCORE of 9[1] & 10[5].

Baby had a bluish coloration of the whole body and ambubag ventilation was done prior referral.

Clinical History of Biological Parents

Mother visited antenatal clinic once at 31 weeks of gestation age, where she received malaria prophylaxis, Folic acid, mebendazole and tetanus vaccination. Laboratory investigation done during antenatal visit included HIV test, syphilis test and protein in urine test which all revealed negative results. She was blood group was A Rh (D) positive. All those management was done as per country regulation on the management of pregnant woman.

There was no history of other medication used during pregnancy, or history of any contraception prior pregnancy. No any imaging diagnostics like X-ray or ultrasound scan were done throughout mother's pregnancy period.

His parents were a non-consanguineous couple, and at the time mother was 36 years old and father was 44 years old and both had no history of smoking or alcohol use. He was a ninth born in the family where other older siblings were phenotypically normal.

Physical Examination

Upon examination he had age appropriate growth measurement with multiple congenital anomalies including cutis aplasia congenita that measured 3-4cm, in diameter, with visible vessels pulsating on parietal - occipital junction, microtia, microphthalmia, a broad nasal tip, bilateral cleft lip and cleft palate, bilateral polydactyl, syndactly left ring finger as shown in figure A to C. Clinically he was dyspneic with central and peripheral cyanosis when off oxygen therapy and was unable to breastfeed or drink from a cup.

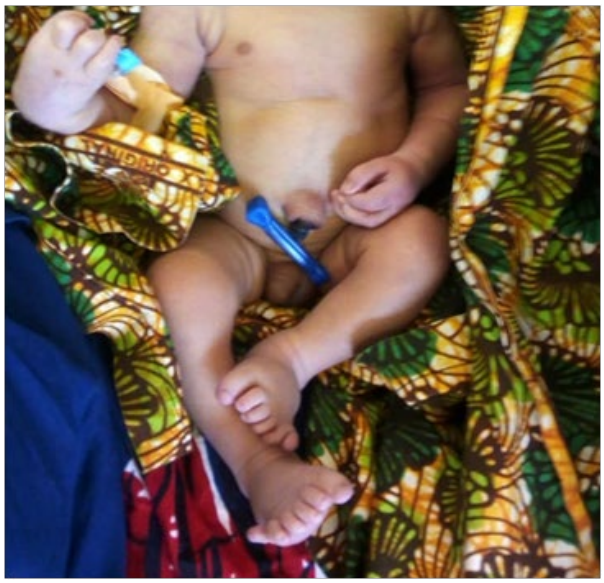

Figure A: This Shows Polydactyl

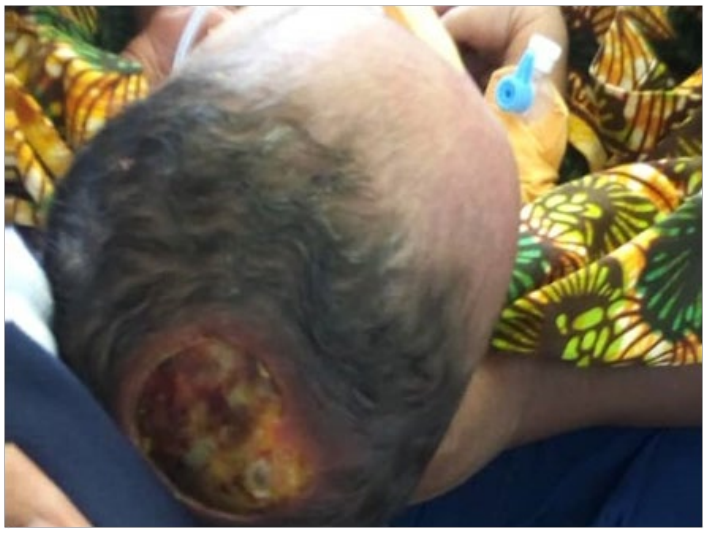

Figure B: Cutis Aplasia Congenita

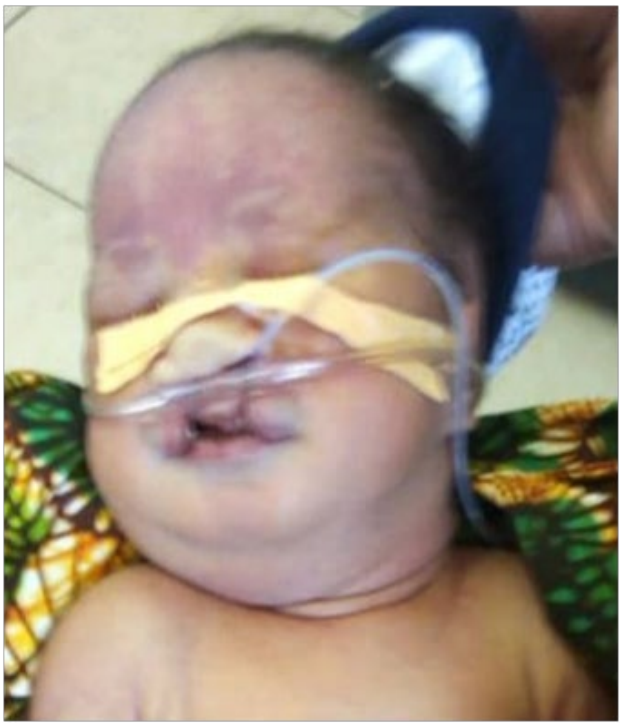

Figure C: Bilateral Cleft Lip

However, the sonographic studies were planned to rule out other internal congenital anomalies (cardiac, renal and further cranial studies) but could not be done. No genetic examinations were done due to unavailability of service in the hospital and region. The final clinical diagnosis was Patau syndrome.

Management and Outcome

He was kept on oxygen therapy, IV antibiotics, fed expressed breast milk via nasogastric tube in NICU, dressing and covering of cutis aplasia with Vaseline gauze and routine care for newborn

including counseling of parents with regards to Patau Syndrome. Due to poor prognosis of the baby referral was planned to a higher health facility for a more specialized neonatal care management but the parents did not consent and decided to continue staying in the hospital where baby died on the sixth day. The post counseling was done to the parents in which deemed to be successful.

Discussion

This genetic disorder affects a variety of organ systems, including the central nervous system, cardiopulmonary system, urogenital system, and musculoskeletal system, among others [4]. Diagnosis of patau syndrome based on the clinical manifestation, phenotypes and the genetic. This case was managed to be diagnosed by clinical manifestation and phenotype. The presence of features provides more confidence of concluding the condition as patau syndrome. As observed above in figure A to C shown some features of patau syndrome. The infant had a bilateral cleft lip and palate (figure C), became a karyotype that resulted with an extra copy of chromosome 13 [3]. The patient was informed in detail, as well as receiving appropriate advice and recommendation[3]. Ultrasound screening can reveal an enlarged nuchal translucency at 12–14 weeks of gestation, and a wide spectrum of major anomalies that can be associated with trisomy 13 such as; CNS anomalies although in this case it was not done [3]. Skeletal malformation such as postaxial polydactyly and clenched hands are also some characteristics of Patau syndrome as observed on figure A which shows six toes and clenched hand [3]. Also the Cutis aplasia congenita is one among the features of the patau syndrome as illustrated in literature, which such a feature are seen in the figure B [3]. Microcephaly, Low-set ears, Cleft palate, Cleft lip, Congenital heart disease, Polydactyly, are the common clinical features of full patau syndrome once are presentin which all were present in present case and some can be observed in figures A-Cabove [9]. New born with full patau syndrome died on the first week of life where by the survival median is about 2.5 days in which life expectancy of the present case died at age of 6 days.Risk factors include advanced maternal age [1, 10]. Counseling parents of a fetus with patau sydrome remains difficult because of the phenotypic variability associated with the condition; some patients exhibit the typical phenotype of full patau syndrome with very poor prognosis while others have few dysmorphic features and prolonged survival, in which in this case the parents deny referral due to poor prognosis although the medical counseling was provided which is not genetic counseling as developed countries [4].

Conclusion

To diagnose patau syndrome require skills of identifying multiple congenital anomalies with increased neonatal and infant mortality. Although care of the affected infant and support for the family can and should be offered by all healthcare practitioners. Capacity building to health practitioners in recognizing and diagnosing patau syndrome should be done to all health facilities levels also this case is rare cases of patau syndrome because few presented with cutis aplasis the birth defect should be included in the national electronic integrated diseases surveillance response (eIDSR) to determine the magnitude of all birth defects.

References

1. Čulić, V., Polić, B., Mišković, S., Dragišić Ivulić, S., Žitko, V., Šipalo, T., & Pavelić, J. (2016). Patau syndrome. Paediatria Croatica, 60(1), 27-29.
2. Mustaki, U., & Jackson, S. (2015, January). A patient with Trisomy 13 mosaicism: review and case report. In BMC Proceedings (Vol. 9, No. 1, pp. 1-1). BioMed Central.
3. Gashi, A. M. Patau Syndrome-A case later diagnosed.
4. Padilla, C. D., Padilla, P. J. D., Tanchanco, L. B. S., Myrian, R., & Salonga, E. G. (2020). A Case Study of Mosaic Trisomy 13 in a 2-year-old Filipino Child. Acta Medica Philippina, 54(4).
5. Doco-Fenzy, M., Mauran, P., Marie Lebrun, J., Bock, S., Bednarek, N., Struski, S., ... & Goossens, M. (2006). Pure direct duplication (12)(q24. 1→ q24. 2) in a child with Marcus Gunn phenomenon and multiple congenital anomalies. American Journal of Medical Genetics Part A, 140(3), 212-221.
6. Duarte, A. C., Menezes, A. I. C., Devens, E. S., Roth, J. M., Garcias, G. L., & Martino-Roth, M. G. (2004). Patau syndrome with a long survival. A case report. Genet Mol Res, 3(2), 288-92.
7. Sendow. (2007). Letter. J Neuropsychiatry Clin Neurosci, 3, 201-2.
8. Feben, C., Kromberg, J., & Krause, A. (2015). An unusual case of Trisomy 13. South African Journal of Child Health, 9(2), 61-62.
9. Choi, K. C., Shin, H. S., Park, Y. E., Seo, J. L., Lee, S. W., Ro, E. S., & Kim, Y. P. (2002). Partial Trisomy 13 (Patau Syndrome): An Autopsy Report. The Korean Journal of Pathology, 36(5), 338-340.
10. Staso, P., Paitl, S., & Patel, D. R. (2018). An 8-week-old infant with trisomy 13: dilemmas for medical decision making. AME case reports, 2.