www.jmscr.igmpublication.org
Impact Factor 5.84
Index Copernicus Value: 83.27
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v5i6.187

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

Aplasia of Tibia –Ectrodactyly Syndrome – A Rare Disease

Authors
Dr N.L.N.Moorthy[1], Dr Padmaja .S[2], Dr M. Vitaleshwar Rao[3], Dr Harshini Reddy[4]
[1]Professor Radiology, [2,4]Senior Resident Radiology, [3]Assistant Professor Radiology
Department of Radiology, Apollo Institute Of Medical Science and Research Jubilee Hills, Hyderabad 500096 Telangana INDIA
Corresponding Author
Dr Padmaja .S
Department of Radiology, Apollo Institute of Medical Science and Research, Jubilee Hills, Hyderabad 500096 Telangana India
Email: *docpadmajareddy@yahoo.co.in*, Mobile: 9652965345

ABSTRACT
Also called as split hand/foot malformation with long bone deficiency, aplasia tibia and ectrodactyly is an extremely rare and isolated trait with degree of deficiency of tibia and fibula occurring at an incidence of 1: 1,000,000 live births. We report a case of a 9 year old boy who presented with unilateral lower limb malformation along with ectrodactyly.
Key Words: *Split Hand/Foot Malformation- Tibial Aplasia – Plain Radiographs.*

INTRODUCTION

Tibial aplasia with ectrodactyly belongs to a group of disorders under limb deficiencies which occur very sporadically. They are listed as " rare disease" by the office of Rare disease (ORD) of National Institute of Health and by Ophanet.

The approximate incidence is about 1 in 1,000,000 live births. The mode of inheritance is autosomal dominant with reduced penetrance. The gene maps to chromosome 1q42.2-q 43. This rare entity can occur as an isolated anomaly or a part of complex syndrome. The extent of limb deficiency is very variable extending from absent digits to absent or hypoplastic tibia , fibula or patella.

CASE REPORT

A 9 year old boy born to nonconsanguineous couple presented with multiple limb abnormalties involving both hands and right lower limb. The right hand had two fingers and the left hand showed three fingers, with the thumb absent in both.

The right lower limb was hypoplastic with flexion contracture deformities at knee and ankle joint. Left lower limb was normal. There was no evidence of any systemic disorder. Plain radiographs of right hand showed absent thumb and middle finger and the presence of two fingers only with a deep cleft in between

the two fingers. The left hand showed the absence of thumb and middle finger and the presence of three fingers with a deep cleft between the fingers. There was fusion of two metacarpals. 4-5 well corticated carpal centers appeared suggesting delayed skeletal maturation. Radiographs of right lower limb showed relativey hypoplastic femur

with normal hip joint. Tibia absent. Fibula showed marked bowing. Patella absent. Only talus, cuneiform and two metatarsal bones formed. Other tarsals and metatarsals absent. Left lower limb was normal. Routine lab investigations, chest radiograph, ECG, 2D echocardiography were normal.

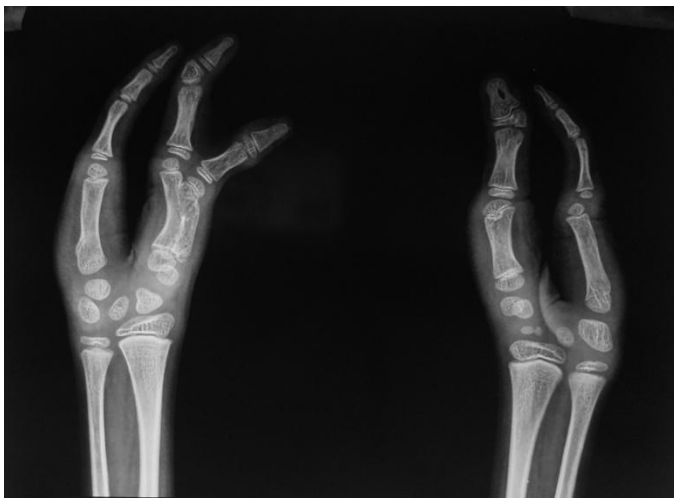

Fig 1 Radiographs of both hands show absent middle fingers and thumb with deep cleft between the fingers.

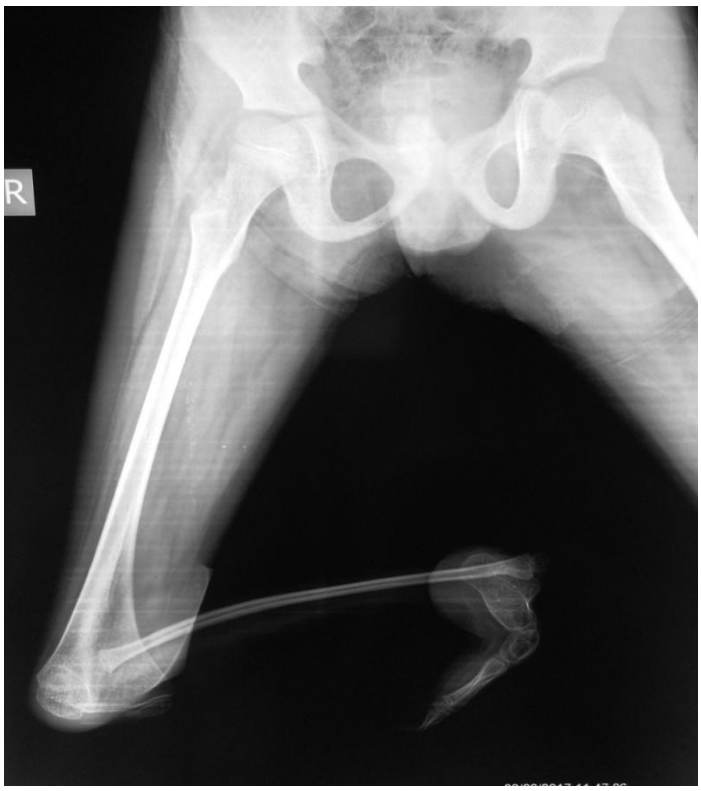

Fig 2 Radiograph of pelvis show absent tibia, patella and calcaneum

DISCUSSION

Aplasia of tibia with ectrodactyly also called as split hand/foot malformation with long bone deficiency (SHFLD) is an extremely rare disease with limb deficiency. The main clinical features of this condition include split hand, absence of multiple fingers, absence of toes, tarsals, metatarsals, tibia and fibula. Other associated symptoms include cupped ears, aplasia of humerus, ulna, radius, monodactaly, clubfoot, post axial polydactaly, metatarsus adductus, bifid femur and hypoplastic femur.[1,2] This disorder was reported as early as 1985 by Majewski and others [3] in six families involving 34 persons. The spectrum of involvement varied from bilateral aplasia of tibia, hypoplasia or bifurcation of femora, hypo or aplasia of ulna, hypoplastic big toes along with ectrodactyly. Lee ML and Chen M[4] reported this syndrome in a 9 month old female infant in association with coarctation of aortic arch and bronchial stenosis. By using segregate analysis and multipoint Lod score calculation Karina et al[5] identified the chromosomal region 17p13.1-17p13.3 as the novel mutated gene responsible for this disorder and suggested that there was incomplete penetrance and variable expression. Ultrasound diagnosis of this condition was done in the antenatal period also.[6,7] Prenatal ultrasound is advised in all pregnancies in the affected families to screen for the evidence of limb anomalies[8]. The differential diagnosis of this condition include Gollop Wolfgang complex which show bifid femur besides hand ectrodactyly. Neha and others[9] in their review on pediatric orthogenomics state that there is a potential role for genetics in many pediatric disorders including limb malformation.

CONCLUSION

Awareness of this condition along with the radiographic studies are important in suspecting and early and better management in the childhood.

REFERENCES

1. Stefan Mundlos and Denise Horn Chap; reduction anomalies: split hand foot malformation in Limb Malformations: an atlas of genetic disorders of limb development, Springer Verlag, Berlin 2014 page 229-231.
2. Kenneth Lyons Jones, Marilyn Crandall Jones, Miguel del Campo in : Smith's recognizable patterns of human malformations 7th edition Elsevier Canada
3. Majewski F, Kuster W, ter Haar B, Goecke T.Aplasia of tibia with split – hand /split –foot deformity. Report of six families with 35 cases and considerations about variability and penetrance Hum Genet 1985 ; 70(2): p 136-47
4. Lee ML, Chen M Tibial agenesis-ectrodactyly syndrome associated with novel cardiovascular and brochopul-monary malformations Clin Dysmorphol 2007 jan ; 16 (1): 47-9
5. Karina Lezirovitz, Sylvia Regina Pedrosa Maestrelli, Nelson Henderson Cotrim. Paulo A.Otto. Peter L.Pearson. Rehgina Celia Mingroni-Netto A novel locus for split-hand /foot malformation associated with tibial hemimelia(SHFLD syndrome) maps to chromosome region 17p13.1-17p13.3 Hum Genet (2008) 123: p625-631
6. http://www.fetalultrasound.com/online/text/30-104 .HTM
7. M.Dreyfus J,J.Baldauf,E.Rigaut J,M. Clavert,B.Gasser, J.Ritter Prenatal diagnosis of unilateral tibial hemimelia Ultrasound Obstet Gynecol 7 (1996) p 205-206.
8. Akhil M.Kulkarni,Bharath Madanlal Jain, Muralidhar Laxmanrao Kulkarni, Bhagyavathi Muralidhar Kulkarni Tibial aplasia with ectrodactyly- a rare case report. Journal of research in radiodiagnosis, teleradiology and imaging JRRTI 2016;vol2.issue 2.july-december 2016 ; page 18-19 .
9. Neha Sinha, Mark A. Seeley ,Daniel S.Horwitz, Andrea H.Seeley Pediatric orthgenomics: the latest trends and controversies AIMS medical science 2017 4(2): 192-216.