ECTRODACTYLY: A RARE LIMB DEFORMITY

BY

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Abstract

Ectrodactyly is a rare congenital limb malformation with median clefts of the hand and hypoplasia or aplasia of the metacarpals and phalanges. This case was an isolated anomaly, non familial sporadic form in unrelated parents with no previous personal or family history of limb or skeletal abnormalities. The index case presented at 7weeks of life with aplasia of the metacarpals and phalanges of the second, third and fourth digits of the left hand. The diagnosis, genetic counselling and management is presented.

Keywords: Ectrodactyly, Congenital, Aplasia, Hypoplasia, Sporadic, Malformation.

Introduction

Ectrodactyly is a rare limb malformation in which a central portion of the hand and digits are missing(Al Rawi et al., 2020; Rayan & Upton III, 2014; Shetty et al., 2014);ectrodactyly, split hand complex, median hypoplasia, lobster claw hand, crab claw hand, typical and atypical cleft hand have been used interchangeably for this limb abnormalities(Rayan & Upton III, 2014). The term ectrodactyly is derived from two greek word ektroma(abortion) and daktylos(finger)(Caicedo et al., 2022), and isoften associated with monodactyly, syndactyly, aplasia and or hypoplasia of the phalanges, metacarpals and metatarsals(Shetty et al., 2014).

The incidence of ectrodactyly is 1:90000(Pindaria et al., 2023) while the prevalence is 1:100000 births(Ruaud et al., 2020), and is usually inherited as an autosomal dominant trait with incomplete penetrance(Ruaud et al., 2020)andaffectation more common in the upper than the lower limbs(Caicedo et al., 2022).Two subtypes are recognized: typical form with absence of metacarpal bones and phalanges with a v-shaped defect and atypical form where the metacarpal bones assume a u-shaped pattern(Caicedo et al., 2022).

The diagnosis of ectrodactyly can be done in utero in the second trimester of pregnancy using 2D ultrasound(Bailess, 2022; Tambawalaa et al.) and with 3D ultrasound with improved image quality in the first trimester of pregnancy(Blitz & Rochelson, 2016). Plain radiograph was utilized in making the diagnosis in the index case at 7 weeks of life.

Case Report

Baby A.Y is a 7week old male infant who was brought to the children out patient department with atypical cleft of the left middle fingers. The infant was a product of parents with no family ties or personal history of skeletal or limb abnormalities.The parents are young adults, mother is a 25year old trader and father a 28year old artisan.

The mother visited the antenatal clinic thrice before delivery and had her routine antenatal drugs. There was no history of any febrile illness, ingestion of alcohol or herbal mixtures during pregnancy

Pregnancy was uneventful with parturition via spontaneous vaginal delivery at term. Birth weight was 3.14kg and physical examination of the infant was essentially normal. The left hand had a median cleft of the central digits (figures 1 and 2), with plain radiograph showing absence of the second to fourth metacarpal and associated phalanges (figure 3), while the right hand was normal (figure 4). Systemic evaluation was essentially normal.

The child was referred to the orthopaedic and plastic unit for consultation and further management. The orthopaedic unit reviewed with routine investigations and plain radiograph of the affected left hand, thereafter the burns and plastic unit reviewed for reconstruction of the structural anatomy.

We report a rare case of non-familial unilateral ectrodactyly of the left hand in a male infant.

Figure 1: Picture of the left hand palmar surface showing median cleft deformity

Figure 2: Picture of the lefthand (dorsal surface) showing lobster claw deformity

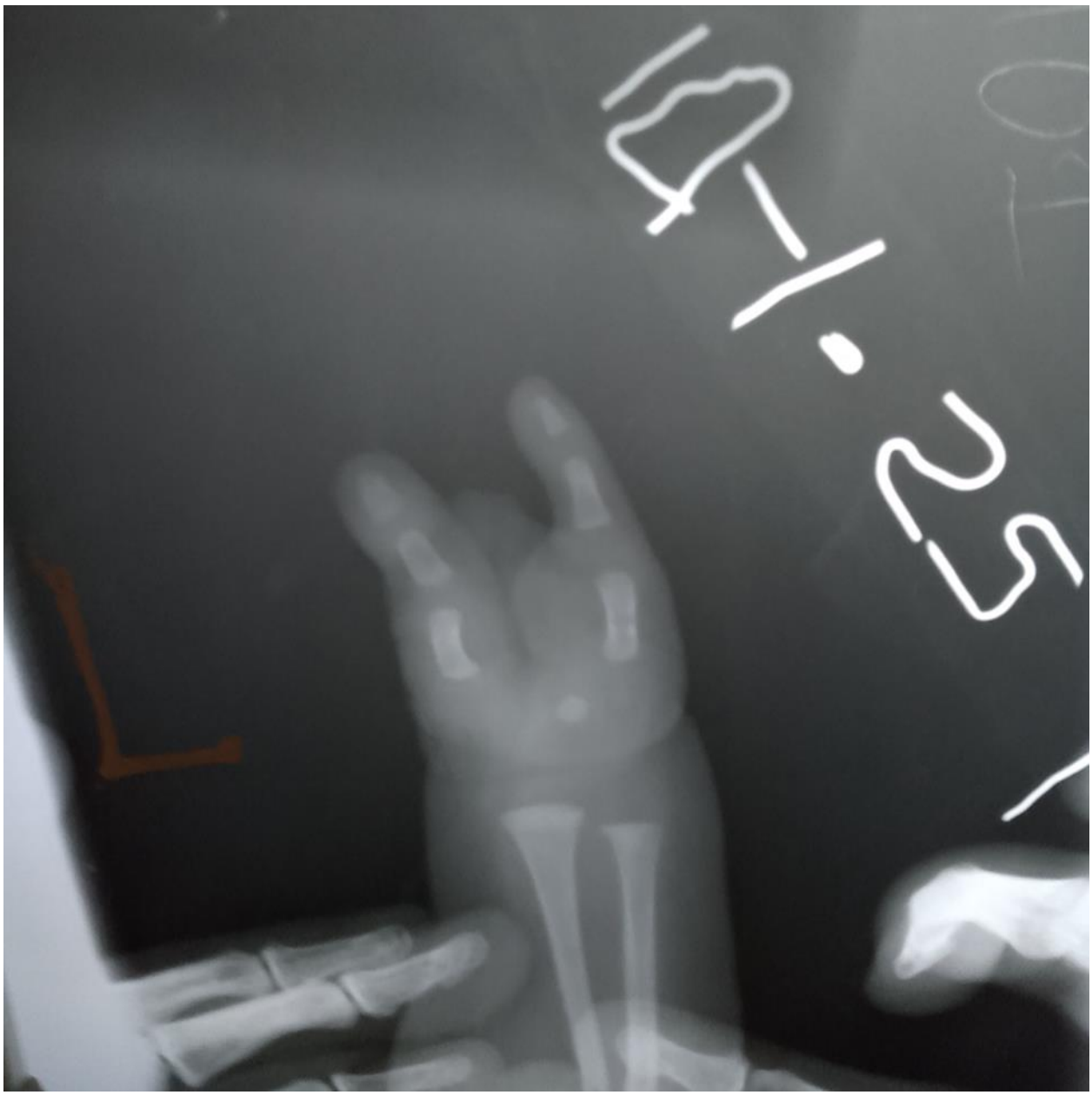

Figure 3: Plain radiograph of the left hand showing absence of the 2nd, 3rd, and 4th metacarpals and their associated phalanges

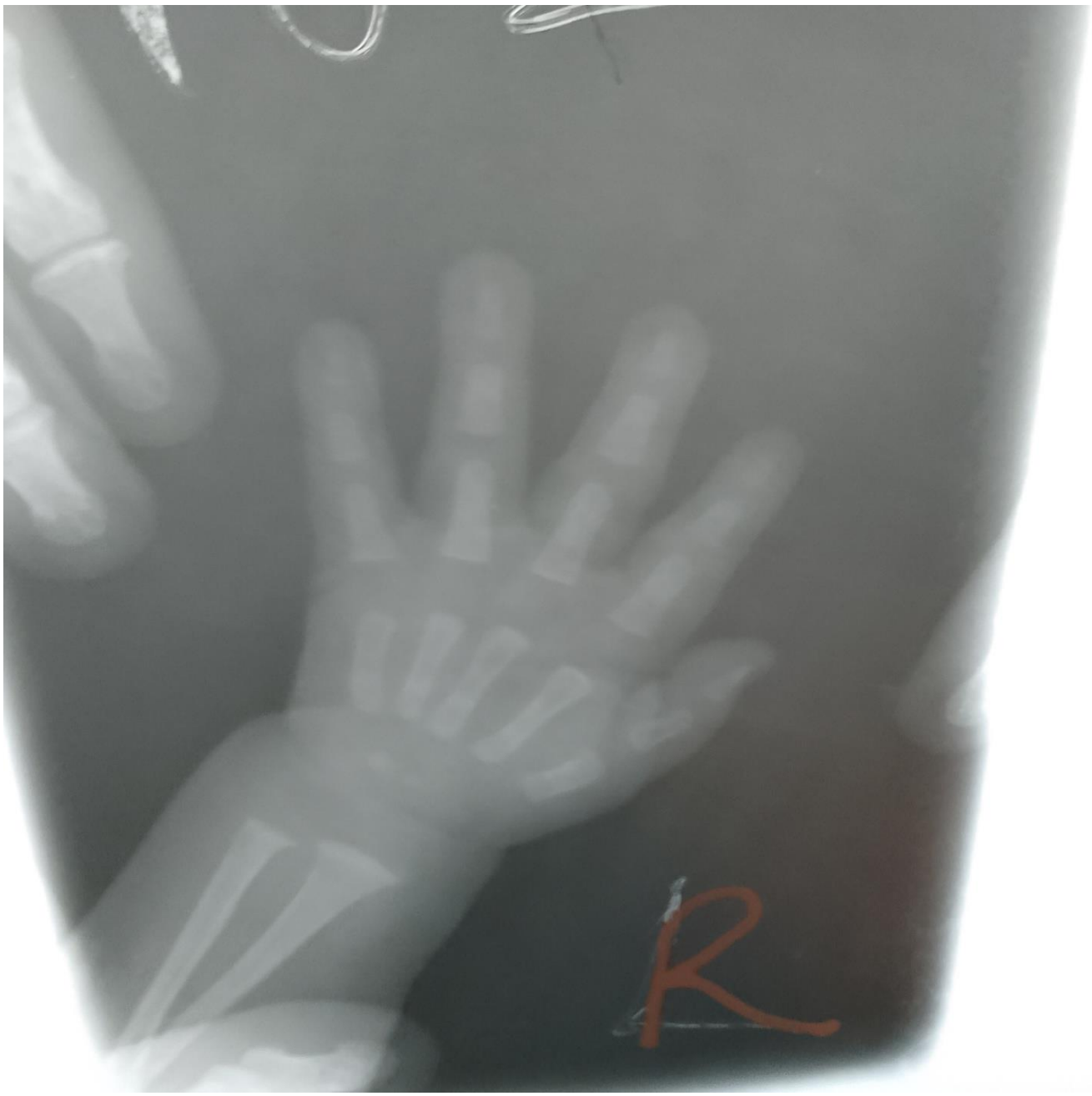

Figure 4: Normal right hand

Discussion

Ectrodactyly result from aberrant development of the hand or foot plates during the seventh week of gestation such that the central rays which form the second, third and fourth digits is defective while the apical ectodermal ridge that leads the differentiation of each ray ceases to function normally(Bailess, 2022; Blitz & Rochelson, 2016). It can be inherited as autosomal dominant, autosomal recessive or X linked depending on the specific genetic mutation pattern(Pindaria et al., 2023; Tambawalaa et al.) and sporadically with no family history(Tambawalaa et al.).

Ectrodactyly occurs in two forms, isolated or as a component of a syndrome(Radu et al., 2023), and any of these two forms can be sporadic or familial with sporadic cases being more common(Blitz & Rochelson, 2016; Durowaye et al., 2011; Kiran et al., 2021).

Chromosomal deletion and duplication are responsible for some cases with anomalies such as tibia aplasia, cranio facial defects and genitourinary abnormalities(Durowaye et al., 2011) and some syndromes such as Carpenter's syndrome, Miller syndrome, Goltz syndrome and Delange syndrome(Durowaye et al., 2011).

The diagnosis of Ectrodactyly can be made in utero (antenatally) or after birth (post natal period). An anomaly ultrasonography in the first trimester with 3Dimensional imaging can detect the cleft digit(s) while a 2Dimensionsal ultrasound with a diligent and patient operator can detect the anomaly in the second trimester.

The diagnosis of the limb anomaly after birth can be detected using a plain radiograph of the affected limb.

The index case presented at seven week of life with left limb anomaly affecting the central digits. The age of the child is evident with the ossification of only one carpal bone, the capitate which is seen in the first three months of life(Hacking, 20 October 2020).

The management of ectrodactly is multidisciplinary involving the radiologist, orthopaedic surgeon, plastic surgeon, physiotherapist, prosthetist and orthotics.

Orthotics help improve the maneuver of the hand with existing fingers whilst a full hand prosthesis is a mechanical solution that improves the range of movement in the upper limb such as wrist and or forearm(Caicedo et al., 2022).

The index case was seen by orthopaedic team, parents were counselled on the management options and given a 6month appointment.

Considerations

Child with ectrodactyly should be evaluated for other congenital anomalies, chest radiograph, plain radiograph, abdominopelvic ultrasound, electrocardiography, 2D echocardiography, genetic analysis should be included in their routine work up examinations. Management involves a multidisciplinary approach for correction of functional disability or improvement and for cosmetic reasons.

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