Case Report—Unusual Cases of Aplasia of Thumb

M.Y. Dofe[1*], Savgaonkar[2], M.M.Meshram[3]

[1]Assistant Professor, Dept. of Anatomy, Government Medical College Nagpur (MS) INDIA.

[2]Retried H.O.D, Dept. of Anatomy, B. J. Medical College, Pune (MS) INDIA.

[3]Prof & H.O.D., Dept. of Anatomy, Government Medical College, Nagpur (MS) INDIA.

[*]Corresponding Addresses:

madhuridofe@gmail.com

Case Report

Abstract: Aplasia of thumb is rare congenital anomaly of hand. It may occur isolated or with syndromes. Along with syndrome it serve as useful marker (along with clinical and radiological finding) helpful to establish correct diagnosis, leading to appropriate management and genetic counseling.

Keyword: Aplasia, congenital, thumb, anomaly, hand, syndrome.

Introduction:

Congenital absence of thumb and first metacarpal is rare anomaly of hand. It may occur as isolated defect or in association with other abnormalities of hand like radial club hand in which hand deviated to radial side or Ulnar club hand in which hand deviated to ulnar side with short forearm or it may occur as component of malformation syndromes like the cardiac, genitourinary, skeletal and haematopoietic system [1]This Aplasia of thumb serve as usual marker (along with clinical and radiological finding) helpful to establish correct diagnosis ,leading to appropriate management and genetic counseling. An early diagnosis has a better functional outcome after reconstructive surgery. The evolution of primate hand is characterized by specialization of the first digit as an opposable unit capable of a pincer like action with the other four digits this process has resulted in progressive shorting in the first ray. Further evolution adaption allow the first ray to be abducted 90 or more respect to 2nd ray[2].If there is absence of thumb there will be loss of pincer action and apposition and hamper the routine activity.

In the present study there are two unusual cases of aplasia of thumb. Case -1, 14years old child showing unilateral aplasia of thumb with hypoplasia little and ring finger. Case-2, showing unilateral aplasia of thumb with aplasia of index finger. All these cases were brought to the OPD at Sasoon Medical and Hospital, Pune with complaints of difficulties in forming routine activities.

Case-1- (figure-1) 14 year old female child deformed of upper limb in the form of a short, curved, right forearm and an absent thumb on one side since birth, she was a product of a non-onsanguineous marriage .There was no history of blood transfusion. There was no family history of a similar deformity in the past two generations. She was developmentally normal for his age and was studying in standard 9th. She was anthropometrically within normal limits. Physical examination revealed unilateral absence of thumb .with short forearm .She had movements of the right elbow flexion and extensions were limited. All the distal movements including the rotator movements of the forearm and the wrist and the fine finger movements were within limit. The systemic examination was normal. Evaluation Chest X-ray, echocardiogram, haemogram the platelet count, and ultrasound of the abdomen were normal.

Fig 1: photograph showing aplasia of thumb with hypoplasia of little and ring finger of right hand.

Case-2(Figure-2) 32 years male having deformed right upper limb. On The physical examination of the right hand show unilateral absence of thumb with aplasia of index finger .No family history of deformed hand..On systemic examination was normal. Evaluation Chest X-ray, echocardiogram, haemogram the platelet count, and ultrasound of the abdomen were normal.

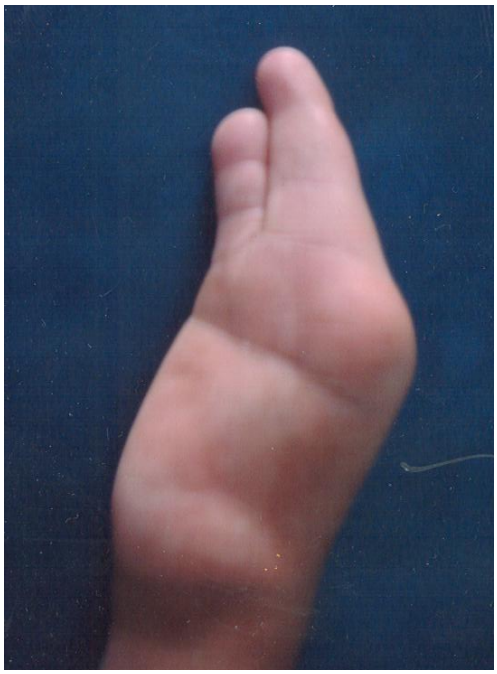

Fig 2- photograph showing aplasia of thumb and index finger of right hand

Discussion:

Absence of thumb is rare congenital anomaly. It is a longitudinal deficiency along the preaxial or the radial aspect of the upper extremity. The frequency of this anomaly is between 1:30000 to 1:100000 live births. Several theories were postulated, like maternal drug exposure, compression of the uterus and vascular injury, but the current theory relates the etiology to the Apical Ectodermal Ridge (AER). AER is a thickened layer of ectoderm that directs the differentiation of the underlying mesenchymal tissue and limb formation. Therefore, a defect of AER is the most probable cause of aplasia of thumb.The extent of the deformity is related to the degree and extent of AER absence[3]. According to the rchipterygeal theory of Gegenbauer[4] the upper limb consists of a main stem and four accessory rays. The humerus, the ulna, the two carpal bones, the fifth metacarpal,and the

three phalanges of the fifth fmger make up the stem. The radius, the navicular and greater multi-angular carpal ones, the first metacarpal and the two phalanges of the thumb constitute the first accessory ray; the second, the third and the fourth accessory rays are constituted by the index finger, the middle fInger and the ring fmger with their respective metacarpals and bones. It is proposed that congenital absence of the thumb is due to the suppression of development of the first accessory ray. The cleavage plane between radial and ulnar paraxial hemimelia lies along the index finger; in defects of the ulna, the three ulnar fIngers with their respective metacarpals and carpals are often absent, whereas with defects of the radius, the thumb is often missing. Our case were unilateral absence of the thumb. Although the present case was diagnosed very late due to negligence by the parents.

Nowadays, with the availability of better medical facilities and awareness among parents, most ofaplasia of thumb with other hand anomalies are diagnosed during the first year of life with a better functional outcome after reconstructive surgery [5,6]Although aplasia of thumb can occur in isolation, or it is many times associated with other congenital anomalies like the cardiac,genitourinary, skeletal, and the haematopoietic system [7] .Our case didn't have any associated congenital anomaly. The cardiac, genitourinary, skeletal and haematopoietic system involvement requires clinical, radiographic, echocardiogram and laboratory evaluation as appropriate management.

Aplasia of thumb associated with genetic syndromes such as the Holt-Oram syndrome, Fanconi's anaemia and thrombocytopenia, sporadic syndromes such as the VATER (vertebral anomalies, analatresia, tracheo-oesophageal fistula, radial anomalies and cardiacdefects) and Nager's acrofacial dysostosis. Thalidomide ingestion results in a series of syndromes, depending on the time of ingestion. The trisomy-18 syndrome with low-set~ retroflexible thumb is a more frequent feature of radial dysplasia. Absence or hypoplasia of the thumb may occur in patients with deletion of the chromosome 13 long arm.

Our case had isolated aplasia of thumb and didn't fit into any of the above mentioned defined syndromes. Whenever aplasia of thumb is identified, it is imperative to conduct a thorough examination and a diagnostic evaluation of the new born to delineate the associated anomalies that may suggest a syndrome. Once the birth defects have been identified, a treatment plan needs to be developed for the infant, with the gastrointestinal, renal and cardiac anomalies usually equiring early surgical management. If the patient survives these surgeries, the prognosis is usually good. The orthopaedic abnormalities can be treated individually. Surgical management of aplasia of thumb include reconstruction by surgery that pollicinazation of index finger.[8]Specific contraindications for the operative treatment include severe associated Anomalies which are not compatible with long life, inadequate elbow flexion, better functional outcome has been documented with corrective surgery.

Conclusion: of this study whenever aplasia of thumb is identified, it is imperative to conduct a thorough examination and evaluation of the new born to delineate the associated anomalies that may suggest a syndrome, because an early diagnosis and appropriate treatment have a better outcome.

References:

[1] Poznanski Ak,GarnSM,Holt JF.The thumb in the congenital malformation syndromes.Pediatr Radio., 100;115-29, 1971.

[2] Ashley –MountagueFM .On the primate thumb J Phy Antrropol., 15:291-314, 1931.

[3] Kozin SH. Radial Club hand, April5, 2011.

[4] Gegenbauer, C.: Morph. Jahrb., 1876: 2, 396.

[5] S A, Rabah SM.. Congenital isolated right radial club hand.Libyan J. Med. 5: 5090, 2010.

[6] Farzan M., Mortazavi SMJ, et al. Congenital radial club hand: result of centralization in 10 cases. Acta Medica Iranica, 43(1): 63-67, 2005.

[7] Fraser, F. C. Disease of the newborn, 4th ed., p. 894. Philadelphia: W.B. Saunders, 1977.

[8] D Buck-Gramcko Pollicization of the index finger. Method and results in aplasia and hypoplasia of the thumb JBJSVolume 53-A, 1605-1617, 1971.