

UNILATERAL CONGENITAL THENAR HYPOPLASIA: A CASE REPORT

Tan Chen Liang* and Surinder Singh

International Medical University, Kuala Lumpur, Malaysia.

*Corresponding Author: Tan Chen Liang

International Medical University, Kuala Lumpur, Malaysia.

Article Received on 12/07/2020

Article Revised on 02/08/2020

Article Accepted on 23/08/2020

ABSTRACT

We present a case of a 15 years old girl with features of Cavanagh syndrome with hearing loss, visual defects, congenital cardiac problem and developmental defect of her right hand. It is a rare presentation worldwide with a higher incidence among females. This case was misdiagnosed twice and this presentation is to increase awareness of the condition.

KEYWORDS: Cavanagh Syndrome, clinical features, misdiagnose, awareness.

INTRODUCTION

The existence of congenital thenar hypoplasia is a rare anomaly and was first described by Muller and later modified by Blauth. It may be unilateral or bilateral and may occur in isolation or associated with some other developmental disorder. The syndrome of significance in this patient is the Cavanagh Syndrome which was first described in 1979 by N.P.C.Cavanagh^[2] It has also been described in relation to Holt-Oram Syndrome and Okihiro Syndrome.

CASE REPORT

The patient presented to our clinic at 15 years of age with a complaint of mild dysfunction of her hand especially the thumb. Although she was aware of the deformity from young she was unconcerned by it. There was no history of trauma to her right hand. She had a history of a congenital cardiac problem and she was on follow-up. She was of small stature, using corrective lenses for her sight and a hearing aid for her right ear. Her thumb was small and low based with wasting of the thenar muscles. The grip was poor and there was weakness of abduction and opposition. There was no sensory loss and the radial pulse could not be felt; Allens test was positive. The ulnar and brachial pulses were both felt. There was no deformities of the spine or anomalies of the rest of the right upper limb. X-rays done showed smaller than normal first metatarsal and a CT angiogram showed absence of the radial artery 6cm from the wrist joint. The ulnar artery was intact and showed good collaterals in the hand. Based on the findings it was concluded that the patient had a hypoplastic right thumb with absent radial artery in the terminal 6 cm, wasting of her thenar eminence, with weakness of the opponens and abductor

pollicis brevis but with no sensory loss. This led us to the diagnosis of Cavanagh Syndrome.

In the syndrome described by Cavanagh in 5 patients with thenar hypoplasia and absence of the terminal radial artery the patient had neurophysiological studies showing a decreased response in the abductor pollicis brevis muscle as isolated lesions. Unfortunately our patient defaulted her appointments to have a nerve conduction study; despite that we were able to demonstrate weakness of the abductor pollicis brevis. Our patient also displayed a hearing and visual defect with a congenital cardiac condition.

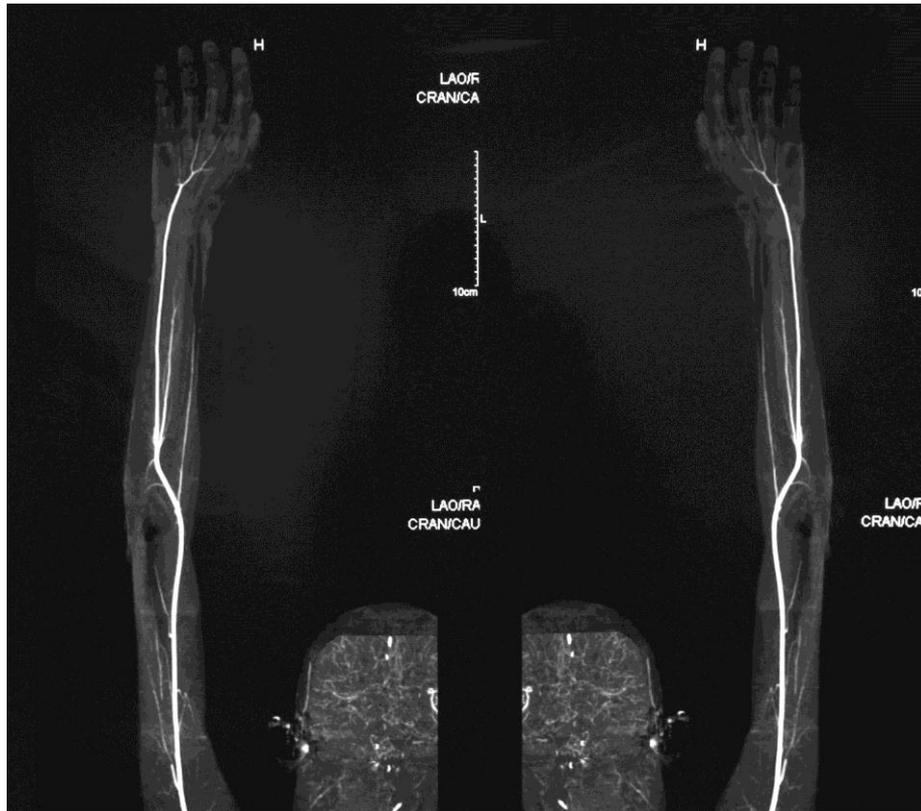


Fig. 1: The CT Angiogram of the patient showing the radial artery cut-off 6 cm from the wrist.

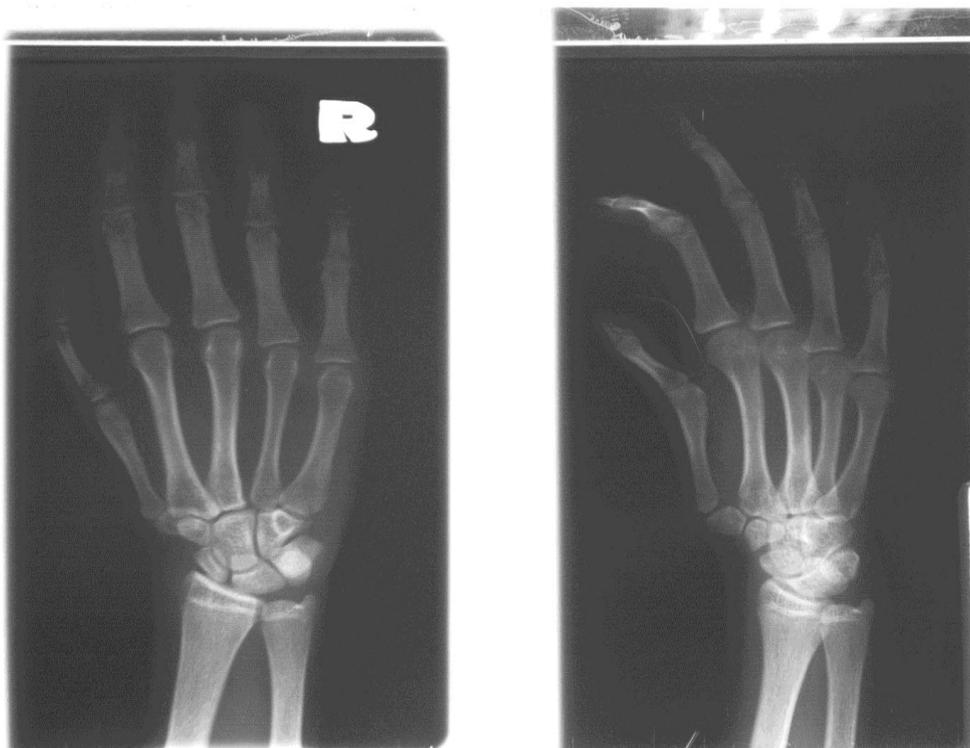


Fig. 2: A radiograph of the patient's right hand showing a slender first metatarsal.

DISCUSSION

Hypoplasia of the thumb was first described by Muller in 1937 and later expanded by Bauth in 1967, it can be in isolation or as part of another syndrome. The incidence is about 1:100,000 with males being more predisposed to

the condition to females. Blauth's Classification describes 5 types of deficiencies of the thumb starting from minor dysplasia to the complete absence of the thumb. Our patient was classified as type II that is all osseous structures are intact with thenar hypoplasia. The

patient was not so severely impaired as to have required surgical intervention.

There may also be defects of the branchial arches which give rise to the 3rd aortic artery and lead to some defects of the arteries of the aortic arch including PDA.^[1] Vascular development precedes bone development in the development of the limb buds.^[1] A number of syndromes are associated with hypoplasia of the thenar muscles, namely Cavanagh Syndrome Holt-Oram Syndrome (HOS gene), Okihiro Syndrome, Kippel Fiel Syndrome, Fanconi's pancytopenia with radial anomalies or as part of VACTERL and trisomy 18. All these syndromes have a genetic link. These syndromes are mentioned to show the complexities involved in association with anomalies of the thumb.

As was established, this patient presented with an isolated lesion but on close questioning and examination it is possibly part of Okihiro's Syndrome (hearing and visual defect with thenar malformation). Cavanagh Syndrome is a description of a hypoplastic thenar eminence which can present in isolation or as part of a syndrome.

Okihiro syndrome, also known as Duane Radial Ray Syndrome (DRRS), is an autosomal dominant condition characterized by radial ray defects, Duane anomaly and bilateral deafness.^[1] Duane syndrome (DS) is a congenital eye movement disorder characterized by limitation of abduction and narrowing of the eyelid fissure with retraction of the globe on adduction.^[3]

In the majority of affected patients with Okihiro syndrome, mutations in the SALL4 gene of the chromosome 4 20q13, 13-13.2, have been identified^[4] The SALL4 gene product is a zinc finger protein that is thought to act as a transcription factor. Therefore, it has been hypothesized that SALL4 mutations may interfere with the differentiation of stem cells during embryonic development.^[5]

REFERENCES

1. Zhao F, Bosseroff A-K, Buettner R, Moser M A Heart-Hand Syndrome Gene: *Tfap2b* Plays a Critical Role in the Development and Remodelling of Mouse Ductus Arteriosus and Limb Patterning. *PloS ONE*, 2011; 6(7): E22908. Doi:10.1371/journal.pone.0022908 accessed on 1 Oct 2016 8pm.
2. Cavanagh N.P.C, Yates D.A.H, Sutcliffe J: Thenar Hypoplasia with Associated Radiological Abnormalities. *Muscle & Nerve*, 1979; 2: 431-436.
3. Walll, Lindley B, Piper Samantha L, Habenicht R., Oishi Scott N, Ezaki Marybeth, Goldfarb Charles A. Scientific Article: Defining Features of the Upper Extremity in Holt-Oram Syndrome *Journal of Hand Surgery* 2015 Sept 40(9):1u764-1768.
4. Tay, Shian-Chao MD; Moran, Steven L. MD; Shin, Alexander Y. MD; Cooney, William P. III MD *Journal of the American Academy of Orthopaedic Surgeons*, 14(6): 354-36.
5. Thumb Hypoplasia: Mark Karadshe Orthobullets accessed on, 12 Oct Ocular manifestations in Okihiro Syndrome Elena Garcia-Martin, PhD,1 Isabel Pinilla, Prof,2 Carmen Almarcegui, PhD,3 Javier Fernandez, PhD,1 Elizabeth C. Engle, Prof,4 and Feliciano J. Ramos, Prof, 2016; 5.