Symphalangism in a Nigerian family: a case report and literature review

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Abstracts

Background: Symphalangism is a rare congenital abnormality characterized by partial or total absence of interphalangeal joints of both fingers and toes. It is inherited in an autosomal dominance pattern. Both proximal and distal interphlangeal joints can be affected, but proximal interphalangeal joint involvement is much commoner. The condition may be associated with skeletal or non-skeletal abnormalities. Compensatory hyperflexibility of joints proximal and distal to the fused joints is common.

Methodology/Results: We present a case of a 42 year-old lady who presented with retrosternal goitre for surgery who on physical examination was found to have proximal symphalangism in both hands and feet. She however gave a history of similar condition in her father who has associated absence of finger nails. She does not complain of any functional impairment in her usual physical activities and hence did not seek for any medical assistance. On further clinical evaluation she was found to have associated hearing impairment.

Conclusion: The authors are not aware of any previously reported cases of symphalangism in Nigeria. This case is presented to alert clinicians of its existence and possible association with other abnormalities.

Key Words: Symphalangism, Nigerian family, 42 year-old lady

Introduction

Symphalangism is a congenital anomaly characterized by fusion of interphalangial joints of the hands and feet. Harvey Cushing in 1916 coined the term "symphalangism" after investigating a family members who could not flex proximal interphalangial joints.[1] Since then much has been reported about the condition and its association with other congenital anomalies.[2] Symphalangism is a rare condition with very few reports in Africa describing its existance and mode of presention. First reported familial symphalangism in Africa was documented by Moumouni in 1991, where he noticed the transmission through five generation in an African family.[2] Borah and colleagues in another findings in 2006 reported familial symphalangism among two brothers in an Indian family.[3] The authors of this report have not yet come accross any previous report of this anomaly in Nigeria. The aim of this paper is to report the presence of symphalangism in a Nigerian family and alert

clinicians on the mode of presentation and possible association with other abnormalities.

Methodology/Results - Case Report

A 42 year old lady presented for surgical treatment of retrosternal goiter with nil other complaint. On physical examination she was unable to flex all her proximal interphalangial joints of her fingers and toes, in addition to being unable to make a fist. Further examination revealed lack of joint creese on the dorsal aspect of proximal interphalangeal joint of both hands (Fig. 1, 2&3). She has positive family history of similar condition in her father with associated absence of finger nails. She does not complain of any functional impairment in her usual physical activities and hence did not seek for any medical assistance to regain movement of these phalanges. Further clinical evaluation revealed associated hearing impairment. However, we did not conduct a complete audiometry in her.

Discussion/Review

Symphalangism is a very rare congenital anomaly of fingers and toes. It is inherited in an autosomal dominance pattern. The condition is characterized by ankylosis of the interphalangeal joint. It can occur in any finger or toe but commoner in little finger followed by ring, middle and then index finger.[3]

Figure 1: Note inability to flex proximal interphalangeal joint

Figure 2: Note absence of proximal interphalangeal joint creese

Figure 3: Note the inability to make a fist

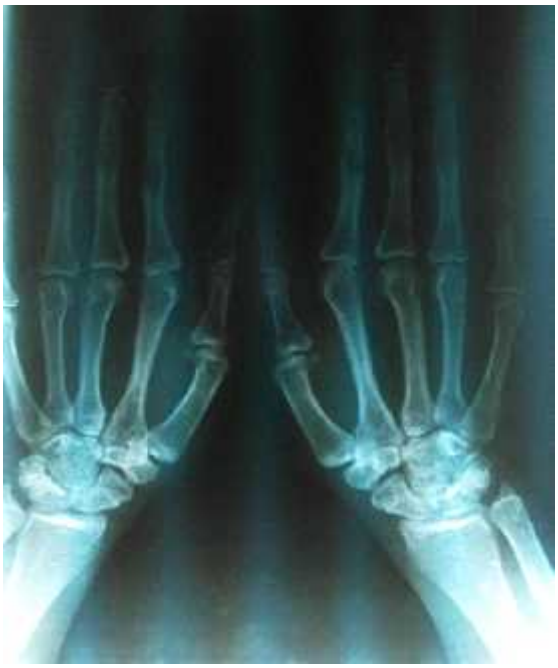

Figure 4: Plain radiograph of the hand confirmed bilateral proximal symphalangism (Note absence of proximal interphalangeal joints and fusion of some carpal bones)

Fusion can either involve proximal or distal interphlangeal joints and hence termed proximal or distal symphalangism respectively. However, proximal symphalangism is more common than the distal type.[3-6] This happened to be the finding in our reported index patient. In addition to having proximal symphalangism was complete ankylosis of the joint with no evidence of a joint capsule. Furthermore, she also has associated fusion of the carpal bones (Fig. 4). Although, involvement of the thumb and great toe has been reported by a researcher in United Kingdom,[7] but this was not the finding in the index patient. Anatomically, there is no joint capsule in cases of symphalangism. A radiograph representation is the appearance of a clear joint space, although complete ankylosis is usually seen after adolescence.[6]

There can be functional impairment in the grip and pinch actions of the affected hand, as well as unsightness of the affected finger, however insome individuals there is no limitation in performing normal physical activities, hence they do not usually seek medical attention to restore movement. Although our patient did not complain of any limitation in perfoming her usual physical activities however she is unable to make a fist and thus hand grip may be difficult for her. Hyperflexibility of the proximal and distal joint as seen in this case usually present as fused joint. And this is a commonly seen phenomenon.[3,8]

Symphalangism is often associated with skeletal and non-skeletal abnormalities, and can be syndromic in some patients. The most common associated skeletal abnormalities is synostosis of the tarsal and carpal bones. Others include brachydactaly, syndactaly, polydactaly clinodactaly, craniosynostosis, hip dysplasia and pes planus.[3,5]

Conductive deafness is often an association as seen in our patient. This results from restricted movement of ear ossicles following fusion.[3,9] Some of the syndromes associated with symphalangism include Poland syndrome, Apert syndrome, Herrmann's syndrome to mention a few.[3]

Different surgical treatment modalities have been described to restore functional ability of the affected joint, although most of these yielded unsatisfactory results. Some of the surgical options include arthorodesis, arthroplasty and free vascularized joint transfer.[6,8]

Conclusion

Symphalangism, first described in 1916, is a rare ankylosis of interphalangeal joints. It is inherited as an autosomal dominance and can be associated with other congenital abnormalities and syndromes. There are few reported cases in the literature about its existence among family members in Africa. This to the best of our knowing is the first report of symphalangism in a Nigerian Family.

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