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POLYSYNDACTYLY OF UNILATERAL THUMB OF THE LEFT HAND- A CADAVERIC CASE REPORT.

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ABSTRACT

Synpolydactyly also called Polysyndactyly is a birth defect that occurs in the fingers or toes to cause the formation of an extra digit or extra toe that are web-like or fused to the other digit.

We are making an unusual presentation of a polysyndactyly of left thumb of a cadaver case that was found during the process of dissection. It is a genetically acquired anomaly that occurs in the form of polysyndactyly mainly an autosomal dominant one. The exact mutations that cause the development of polysyndactyly differ between the type of the condition and in different families. But most of them are caused genetically by a shift in the genetic factors concerning the signaling molecule belonging to the Sonic Hedgehog (SHH). Mostly, mutations are present in the zone of polarizing activity regulatory sequence or ZRS which regulates expression of SHH in the developing limbs. Numerous instances of polysyndactyly are findings of duplications involving the ZRS or its neighbouring pre-ZRS zone.

In this case report of a Weird and rare case of unilateral thumb synpolydacty in an adult male cadaver is presented. These extreme incidences emphasize the necessity of intensive anatomical studies on developing our understanding of congenital limb anomalies and provide informative facts on its scope.

Synpolydactyly which is also known as **Polysyndactyly** is a congenital anomaly which affect the fingers or toe resulting into a formation of an extra finger or toe that are webbed or fused together with the adjacent digit.

We present a rare case of a polysyndactyly on left thumb of a cadaver discovered during dissection. Polysyndactyly is mostly an inherited autosomal dominant anomaly. The specific mutations leading to polysyndactyly are varied between types of the condition and different families. However, the etiology of most of the cases are due to changes in genetic elements affecting the signaling molecule Sonic Hedgehog (SHH). Primarily, mutations are found in the zone of polarizing activity regulatory sequence, or ZRS, that controls the expression of SHH in developing limbs. Many cases of polysyndactyly are the result of duplications of the ZRS or the nearby pre-ZRS region.

An unusual instance of unilateral thumb synpolydactyly in an adult male cadaver is highlighted in this case report. These exceptional cases highlight the need of thorough anatomical research in expanding our knowledge of congenital limb abnormalities and offer insightful information on their range.

INTRODUCTION

Synpolydactyly (Polysyndactyly)
is a congenital anomaly, combining polydactyly and syndactyly, in which affected individuals have an extra finger or toe that is connected, via fusing or webbing, to an adjacent digit.
The extra digit is most commonly postaxial(Ko K.Ret al., 2021), on the same side as the pinky or little toe(Stevenson R.E.,2015). Preaxial polysyndactyly, in which the duplicated digit is on the side of the thumb or big toe, is less common(Stevenson R.E.,2015). Crossed polysyndactyly, in which polysyndactyly is present on the hand and foot, and is preaxial on one and postaxial on the other, is extremely rare and often occurs with other genetic disorders(Dewan P,et al., 2010).

CASE REPORT

Our case was found during a routine gross anatomy dissection for medical students, polysyndactyly of the right thumb was seen. These anomaly was found in a male adult cadaver of

about 40 years at the Kenya Methodist University gross anatomy laboratory on the 14th of February 2024. The individual has an extra thumb(preaxial polysyndactyly) composed of a double metarcapals and phalanges connected via fusing (figure 1). Other than these findings there were no other malformations or conditions noted after intensive investigation.

FIGURE I

DISCUSSION

Polysyndactyly is typically an inherited autosomal dominant anomaly(Holmes L.B., 2011, Dewan P,et al., 2010, Saygin D.1985). The specific mutations leading to polysyndactyly are varied between among types of the condition and different families. However, the etiology of most of the cases are due to changes in genetic elements affecting the signaling molecule Sonic Hedgehog (SHH). Primarily, mutations are found in the zone of polarizing activity regulatory sequence, or ZRS, that controls the expression of SHH in developing limbs. Many cases of polysyndactyly are the result of duplications of the ZRS(Dai et al., 2013, Lohan et al., 2014, Xu et al., 2020) or the nearby pre-ZRS region(Potuijt et al., 2018 and Potuijt et al., 2022). The mechanisms bringing about the resulting SPD presentations via these changes are yet to be fully understood.

In this case the unilateral expression of SPD in thumb is not typical. Most of the reported cases involve bilateral and symmetrical involvement of digits. This instance is further distinguished by the lack of other deformities, which may indicate a novel expression of a known mutation or a possibly unique mutation.

Polysyndactyly can be associated with the presence of other genetic disorders. It is a hallmark of Carpenter's syndrome, an autosomal recessive disorder that is also associated with craniosynostosis, obesity, short stature, and other malformations(Temtamy,1966).Patients with other syndromes, including Pallister-Hall syndrome(Consales, et al., 2022) and Greig cephalopolysyndactyly syndrome(Biesecker,2008) may also display polysyndactyly of varying severity.

Preaxial synpolydactyl of the thumb is rare according to 3, hence a good comprehension of this anomaly of the limb will be of great advantage towards a surgical procedure for corrections or repair of synpolydactyl.

CONCLUSION

In case of polysyndactyly, other genetic disorder cannot be overruled and should be kept in mind most especially for future investigation in prenatal diagnosis. A corrective surgery of the anomaly should be carried out as soon as possible.

An unusual instance of unilateral thumb synpolydactyly in an adult male cadaver is highlighted in this case report. These exceptional cases highlight the need of thorough anatomical research in expanding our knowledge of congenital limb abnormalities and offer insightful information on their range.

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