

# CASE REPORT

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## MACRODACTYLY WITH POLYSYNDACTYLY OF DISTAL PHALANX OF LEFT SECOND TOE – A RARE CASE REPORT AND ITS EMBRYOLOGICAL REVIEW'

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**ABSTRACT:** Polydactyly is one of the common congenital anomaly. It occurs in many forms, ranging from varying degrees of mere splitting to complete duplication. It belongs to the category of duplication. The extra digit can be a small piece of soft tissue that can be removed, but it can also contain bone without joints or it may be a complete, functioning digit. A 33 year old male patient presented with the complaint of pain in left second toe since last one year and uneasiness on wearing shoes. On physical examination, we found macrodactyly of the left second toe with duplication of the nail beds which was noticed by his parent since the birth of child. Radiological findings of the left foot showed the presence of duplicated distal phalanx of the second toe with fused proximal ends. The remaining toes and the right side structures were found to be normal. This is a rare case of macro-poly-syndactyly of distal phalanx of left second toe. Nevertheless orthopedic surgeons should be aware of this anomaly since these patients are often referred for corrective surgery on the feet.

**KEY WORDS:** polydactyly, syndactyly, macrodactyly

**INTRODUCTION:** Congenital anomalies (also referred as birth defects) affect approximately 1 in 33 infants and result in approximately 3.2 million birth defect-related disabilities every year including 0.3 to 1 congenital limb deficiencies per 1,000 live births [1]. Based on the embryonic failure causing the clinical presentation, congenital anomalies of the limb may be classified in seven categories as proposed by Frantz and O'Rahilly and modified by Swanson. These categories are failure of formation of parts, failure of differentiation, duplication, overgrowth, undergrowth, congenital constriction band syndrome, and generalized skeletal abnormalities [2,3]. Polydactyly belongs to the category of duplication [4]. The extra digit can be a small piece of soft tissue that can be removed, but it can also contain bone without joints or it may be a complete, functioning digit.

As there is an association between polydactyly and several syndromes (Majewski Polydactyly Syndrome), children with a congenital extremity deformity should be examined by a geneticist for other congenital anomalies [5]. Research has shown that the majority of congenital anomalies occur during the 4-week period of embryologic development of rapid limb [4]. Polydactyly has been associated with 39 genetic mutations [6]. More specific loci and genetic mechanisms responsible for disorders of duplications will be defined with time, as molecular research continues [4].

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Polydactyly occurs in many forms, ranging from varying degrees of mere splitting to complete duplication. Occasionally it consists of only fleshy nubbins on radial border. When duplication occurs alone, it is usually unilateral and sporadic. The duplication associated with triphalangeal finger or toe usually results from autosomal dominant inheritance. Here we report a very rare case of macrodactyly with polysyndactyly of distal phalanx of second toe.

**CASE REPORT:** A 33 year old male patient presented with the complaint of pain in left second toe since last one year and uneasiness on wearing shoes. On physical examination, we found macrodactyly of the left second toe which was noticed by his parent since the birth of child. There was no history of trauma or no family history of any congenital defects. There was no associated disease of CVS, Genitourinary system. This second left toe was seen to be having duplication of the nail beds [Figure 1A and 1B]. There is no exaggeration of pain on passive movements. Radiological findings of the left foot showed the presence of duplicated distal phalanx of the second toe with fused proximal ends [Figure 2A and 2B]. The remaining toes and the right side structures were found to be normal. All other investigations such as complete blood count, hemoglobin, random blood sugar, routine examination for urine, electrocardiogram and abdominal ultrasonography were normal.

**DISCUSSION:** The hand and foot malformations often require orthopedic assessment and treatment depending upon their embryological defects and clinical presentations. In the present case report, patient reported with macrodactyly. There was simulation of toe with Macrodystrophia lipomatosa which is a form of localized gigantism characterized primarily by proliferation of mesenchymal elements, particularly with a disproportionate increase in adipose tissue without duplication of nail bed [7, 8]. As in the present case, duplication of nail beds excluded Macrodystrophia lipomatosa. This localized mesenchymal proliferation was further ruled out radiologically which indicated duplication of distal phalanx.

The probable reasons of these developmental anomalies had been reviewed on the grounds of embryogenesis and gene regulations. Antero-posterior patterning of the limbs is controlled by the zone of polarizing activity (ZPA), a region of posterior limb bud mesoderm. In the ZPA, the key mediator is Sonic Hedgehog (Shh). Genetic studies in mice and chickens have demonstrated that Shh and GLI3, one of its downstream targets, are important in the development of the digits, more specifically in the regulation of digit number and digit identity. GLI3 gene is located on chromosome 7p14.3. Ectopic implantation of the ZPA or Shh in the anterior part of the limb bud of chick embryos induces mirror-image duplications [9]. GLI3, the homologue of the *Drosophila cubitus interruptus* gene, encodes a transcription factor that plays a crucial role in antero-posterior patterning of the limb bud. The extra toes (Xt) mouse mutation creates a GLI3 null allele, and Xt heterozygotes are characterized by preaxial digit alterations. Homozygote GLI3<sup>-/-</sup> mice have severe polydactyly [10]. Moreover, mice lacking GLI3 and SHH (SHH<sup>-/-</sup> GLI3<sup>-/-</sup>) have 6–11 syndactylous digits per limb [11]. GLI3 and SHH thus both act in normal patterning of the limb by inhibiting the development of more than five digits. The importance of GLI3 and SHH in digit formation is also reflected in human polydactyly syndromes. Mutations in a SHH enhancer have been implicated in isolated preaxial polydactyly [12]. Point mutations or deletions in the GLI3 gene cause Greig cephalo polysyndactyly (GCPS) [13]. GCPS is a rare autosomal dominant disorder characterized by abnormal craniofacial, hand and foot malformations with polydactyly.

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The Wassel classification system is a useful method for classifying duplicated thumbs. It describes 7 classes of thumb deformities with the duplication in each class occurring at different levels along the bones of the thumb. Wassel type II consists of a completely duplicated distal phalanx down to the interphalangeal joint [14]. Under the light of present literature reviewed, it is seen that till date no one has focused on duplicated distal phalanx of second toe with syndactyly. In the present case report, we found duplication of distal phalanx of second toe like Wassel type II thumb.

Nevertheless orthopedic surgeons should be aware of this anomaly since these patients are often referred for corrective surgery on the feet. Since orthopedic surgeons are sometimes the first to see such patients, they should know that the hand and foot malformations can occasionally be associated with other organ anomalies. Further investigations and genetic counselling are therefore mandatory both in sporadic and familial cases, especially since inter- and intrafamilial variability of the phenotype has been described [15].

The primary deformity in such duplications is that the useful toe is significantly deviated at the interphalangeal joint (anything more than 25° of deviation affects the cosmetic appearance). The favored approach to this deformity includes fixation of the deviated toe in an anatomically correct position. The extensor digitorum longus tendon necessitating its release and reattachment to the periosteum of the dorsal base of the midportion of the correct phalanx. Postoperatively, the primary concerns in toe duplications are pulp atrophy, scar hypertrophy, joint instability/deformity, web space contracture, and joint stiffness. Most functional impairment comes from a persistent deviation of the remaining phalanx[16].

**CONCLUSION:** The present case report indicates emphasis on examination of children with congenital deformity by the geneticist.

## REFERENCES:

1. WHO2012. Congenital anomalies. Fact sheets. Available at <http://www.who.int/mediacentre/factsheets/fs370/en/index.html> cited on 6<sup>th</sup> March 2013.
2. Frantz CH, O'Rahilly R (1961). Congenital skeletal limb deficiencies. *J Bone Joint Surg Am* 43: 1202-24.
3. Swanson AB (1976). A classification for congenital limb malformations. *J Hand Surg Am* 1: 8-22.
4. Watt AJ, Chung KC (2009). Duplication. *Hand Clin* 25 (2): 215-227.
5. Van Nieuwenhoven C, Boehmer A, Hovius S (2009). Polydactyly. *Praktische pediatrie* 2.
6. Biesecker LG (2002). Polydactyly: how many disorders and how many genes? *Am J Med Genet* 112 (3): 279-283.
7. Barsky AJ. Macroductyly. *J Bone Joint Surg Am.* 1967;49:1255-66.
8. Mahmood A, Mahmood NF. Macroductyly lipomatosa: A troubled second big toe. *Radiology Case Reports.* 2008;3:92
9. Pearse RV 2nd, Tabin CJ (1998) The molecular ZPA. *J Exp Zool* 282:677-690.
10. Buscher D, Bosse B, Heymer J, Ruther U (1997) Evidence for genetic control of Sonic hedgehog by Gli3 in mouse limb development. *Mech Dev.* 62:175-182.
11. Litington Y, Dahn RD, Li Y, Fallon JF, Chiang C (2002) Shh and Gli3 are dispensable for limb skeleton formation but regulate digit number and identity. *Nature* 418(6901):979-983.

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12. Lettice LA, Heaney SJ, Purdie LA, Li L, Debeer P, Oostra BA, Goode D, Elgar G, Hill RE, de Graaff E (2003) A long-range Shh enhancer regulates expression in the developing limb and fin and is associated with preaxial polydactyly. *Hum Mol Genet* 12:1725–1735.
13. Debeer P, Devriendt K, De Smet L, deRavel T, Gonzalez-Meneses AG, Grzeschik KH, Fryns JP. The spectrum of hand and foot malformations in patients with Greig cephalopolysyndactyly. *J Child Orthop* (2007) 1:143–150.
14. Wassel HD. The result of surgery for polydactyly of the thumb: A review. *Clin Orthop* 1969; 64: 175.
15. Debeer P, Peeters H, Driess S, De Smet L, Freese K, Matthijs G, Bornholdt D, Devriendt K, Grzeschik KH, Fryns JP, Kalff-Suske M (2003) Variable phenotype in Greig cephalopolysyndactyly syndrome: clinical and radiological findings in 4 independent families and 3 sporadic cases with identified GLI3 mutations. *Am J Med Genet A* 120:49–58.
16. Ogino T, Ishii S, Takahata S, Kato H. Long-term results of surgical treatment of thumb polydactyly. *J Hand Surg [Am]*. 1996;21(3):478–86.



**Figure 1:**A.Superior view of left foot showing all five toes. B. Closure view of left second toe showing macrodactyly and duplicated nail.



**Figure 2:** Radiological finding of: A. Showing all five toes B. Closure view of left second toe showing presence of duplicated distal phalanx of the second toe with fused proximal ends.