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PROFILE OF CONGENITAL LOWER LIMB DEFECTS IN A RURAL KENYAN HOSPITAL AND LITERATURE REVIEW

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SUMMARY

Knowledge of the profile of congenital limb defects in a population is important in informing their prevention, management and rehabilitation strategies. The profile displays geographical and ethnic variations. There are few reports from African countries, most of them being case reports. This study aimed at reporting congenital defects of lower limb observed in a rural hospital in Kenya. Records from PCEA Kikuyu Hospital were examined retrospectively for the period 2002-2011. Visible physical defects recorded, age and gender of the victims were analyzed. Images of some remarkable defects were examined. Literature review has been done regarding the defects. Ten varieties of congenital defects were identified. The most common of them were tibial pseudoarthrosis (6) followed by polydactyl, (5) and congenital talipes equinovarus (4). The mean age at presentation was 9 years and there were more males than females. Most of the cases occurred in isolation, with only a few occurring in combination. There is wider variety of lower limb defects in a Kenyan hospital than is usually reported in other studies. Tibial pseudoarthrosis is most common defect. Medical personnel should be aware of this wider scope of limb defects. In addition, greater emphasis should be given to systemic examination of the affected children and rehabilitative management.

Key words: Diplopodia, cleft foot, macrodactyly, pseudoarthrosis, polydactyly, syndactyly.

Introduction

Profile and frequency of lower limb malformations, relative to other congenital defects, varies between populations. Details of such variations are important in planning healthcare measures for prevention, rehabilitation (Abdi-Rad et al, 2008) and management of birth defects in general. There are, however, few reports from Africa on the profile of lower limb defects. In Kenya, these defects constitute a significant cause of lower limb amputation (Ogeng'o et al, 2009). A recent study, however, made a reference only to polydactyly (Muga et al, 2009). This article reports the profile of birth limb defects recorded in Kikuyu Mission Hospital, Kenya and includes literature review on the observed cases.

Materials and methods

This was a retrospective study at Presbyterian Church of East Africa (PCEA) Kikuyu Hospital, a church based 400 bed capacity health facility with an orthopedic centre, in rural Kenya 17 Kilometers to the West of Nairobi. This Hospital has two resident orthopedic surgeons and two consultant general surgeons who visit on need. It caters for natives mainly from the Kikuyu community of central Kenya. Ethical approval for the study was granted by the Hospital Management Board. Records of patients who came to the orthopaedic centre of the hospital with diagnosis of congenital defects of lower limb between January 2002 and December 2011 were retrieved. The

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type of defect, age and gender were analyzed. Only those cases where diagnosis was confirmed, age and gender were specified were included. Macrographs of the cases were retrieved from the image gallery maintained at the hospital. They were categorized into two year age groups starting with one month. The mean ages were calculated for each category of defects. Data were analyzed using the statistical package for social sciences for windows version 11.50 Chicago Illinois, 2002. General descriptive statistics were applied to determine means, frequencies and ranges.

Results

Altogether 29 cases of congenital lower limb defects were retrieved (Table 1). The most common defects were tibial pseudoarthrosis (6), polydactyly (5); macrodactyly (4); Talipes equinovarus (4) (Fig 1 & 2) [Table 1].

Table 1: Types of congenital defects

Type of defect	Frequency	%
Tibial pseudoarthrosis	6	20.7
Polydactyly	5	17.2
Macrodactyly	4	13.8
Talipes equinovarus	4	13.8
Syndactyly	3	10.3
Congenital hip dislocation	2	6.9
Brachydactyly	1	3.4
Cleft foot with macrodactyly	1	3.4
Amputation due to amniotic band syndrome	1	3.4
Diplopodia	1	3.4
Unclassified	1	3.4
Total	29	100

Two cases of congenital talipes equinovarus were each associated with Spina bifida, The case of amniotic band syndrome involved all four limbs while that of diplopodia was associated with arachnodactyly of the other foot.

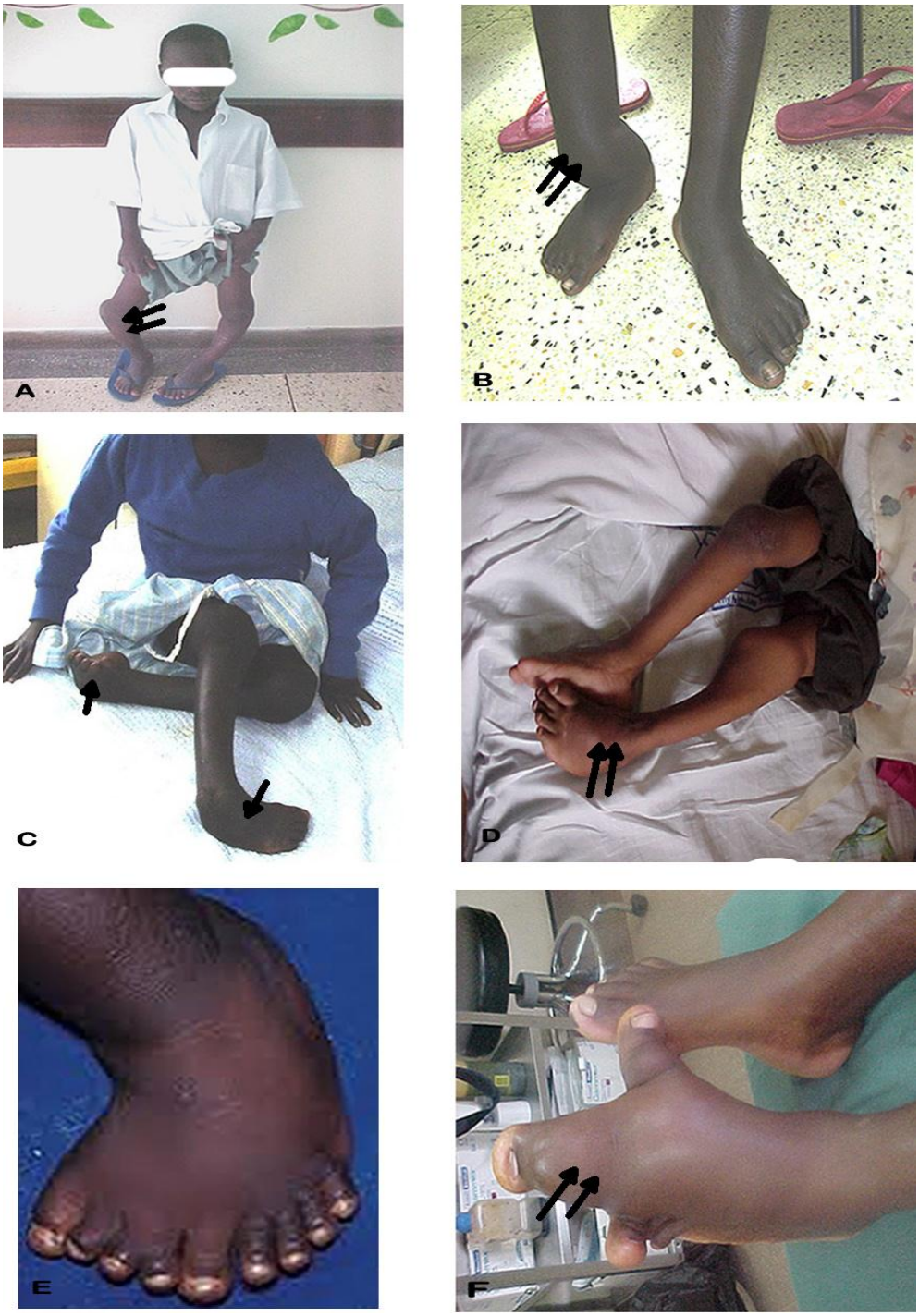
The overall mean age at presentation was 9 years (range 0-20years) varying with type of defect. The most commonly involved age groups were those below 36 months; and above 120 months (Table 2).

Table 2: Mean age at presentation and gender distribution

Defect	Mean age	Gender		Total
		M	F	
Tibial pseudoarthiosis	10.16	3	3	6
Polydactyly	6.0	3	2	5
Macrodactyly	8.25	3	1	4
Congenital talipes equinovarus	4.33	2	2	4
Syndactyly	1.5	2	1	3
Others	10.0	4	3	7
Total		17	12	29

The male: female ratio was 3:2 with males predominantly in most categories

Figure 1: Showing various lower limb defects



Legend: Figure 1: A: Ten years old with bilateral tibial pseudoarthrosis. Notice the severe deformities on both tibia. B: Eight years old with right distal tibial pseudoarthsosis. C: Seven years old girl with bilateral talipes equinovarus. D: Eight years old boy with unilateral talipes equinovarus. E: Four years old with polydactyly *(Photo courtesy of Science Photo library).* F: A child with macrodactyly of the second digit

Discussion and Review of literature of observed defects

There are only a few reports of profiles of limb defects (Tayel et al., 2005; Agha et al 2006).The current series is one of the few reports with such diverse profile of defects from a single rural hospital. This suggests that the population studied is rich in lower limb defects and resources should be expended in establishment of corrective and rehabilitation facilities.

Congenital pseudoarthrosis: This condition results from a basic defect in the mesoderm with failure of normal bone growth and callus formation (Levin et al., 2003). It considered to result from of a primary defect in the embryonic tibial cartilage occurring at the 5^{th} week of intrauterine life. It is a variant of fracture non- union in which there is formation of a false joint cavity with a synovial like capsule and even synovial fluid (Al-Hadidy et al., 2007). A passive constriction phenomenon such as occurs in constriction band syndrome and vascular malformation, has been implicated in its development (Magee et al., 2007; Al Hadidy et al., 2007; Agashe et al., 2011).

The tibia is the most common site but other long bones such as fibula, ulna, radius, femur and clavicle may be affected. All the cases observed in this study were those affecting the tibia only. It is associated with fibrous dysplasia, neurofibromatosis, osteogenesis imperfecta and various other malformations. It is commonly sporadic but hereditary examples have been described (Al Hadidy et al., 2007)

Polydactyly: This is a fairly common congenital condition characterized by presence of supernumerary toes. It is generally classified into three major groups; medial ray (preaxial) central ray and lateral ray (post axial). The duplication may appear at the distal and middle phalanges or at the whole digit and metatarsal. The complexity of the deformity ranges from a simple soft tissue problem to a completely developed accessory ray (Galois et al., 2002). Of the three types, postaxial polydactyly occurs most frequently. Indeed majority of cases reported in African population are postaxial (Watanabe et al., 1992). Polydactyly is further divided into fifth or sixth ray duplication and various spectra of these defects have been described (Belthur et al., 2011).

Macrodactyly: Also called partial acromegaly, dactylomegaly, limited gigantism; and macrodystrophia lipomatosa. This is a rare congenital non hereditary malformation presenting as an increased size of one or several toes. It is characterized by an increase in all mesenchymal elements particularly fibro fatty tissue (Yuksel et al., 2009). The final pathway in causation is either local deficiency of growth inhibiting factor or excessive local expression of basic intrisinc factor, causing excessive growth of all elements of the digit (Syed et al., 2005). There are two varieties namely static and progressive. In static, type which is more common, increase in size is proportionate with normal growth. In the progressive type, there is

disproportionate growth of the affected digits and increases in size faster than could be attributed to the normal growth spectrum (Sharma et al., 2006). Other conditions that may mimic macrodactyly include neurofibromatosis, harmatomas, primary lymphatic disorders, and vascular malformations. There is a slight male predominance, and the 2nd and 3rd digits are involved most frequently (Sharma et al., 2006; Khan et al, 2010).

Congenital talipes equinovarus: This is a relatively common malformation thought to show familial tendency. The anatomical abnormalities include malposition of tarsal bones, atrophy of calf muscle /tendon, shortness of tendons/ligaments. In about 20% of cases, CTEV is associated with other congenital abnormalities. In the observed series shortness of the tendons was the main cause. The cause is unknown but proposed theories include vascular, local, genetic, anatomical, following a compartment syndrome, environmental factors and effect of position in utero. Other factors linked with the condition include amniocentesis, thyroid disorders, small pox vaccination in the first trimester, use of salicylate preparation and prenatal exposure to barbiturates, male gender, maternal anaemia and maternal hypermesis (Byron –Scott et al., 2005). A comprehensive examination of the infant is necessary to detect physical signs which suggest that the condition is not idiopathic. Associated malformation include absence of toes, tibial dysplasia, finger stiffness, arthrogryposis, spina bifida, cerebral palsy and various neuromuscular defects (Siapkara and Duncan, 2007).

Diplopodia: Refers to duplication of the foot. As an independent entity, it is rare when compared to diplocheiria or duplication of the hand. The diplopodia is usually manifested by partial duplication of the foot, and extra part being on the tibial side. Various bones of the foot, tarsal and metatarsals are usually duplicated to various extents. For example, there could be eight toes, ten metatarsals heads, five cuneiforms, a navicular, a cuboid, a bifid talus and a calcaneous (Mysorekar and Lohokare, 1970) or two calcanei, one talus, one navicular, two cuboids and four cuneiforms and nine metatarsals. Muscles may also be duplicated (Narang et al 1982). The condition may be associated with hypoplastic or aplastic tibia and double fibula (Jones et al 1978).

Legend: Figure 2

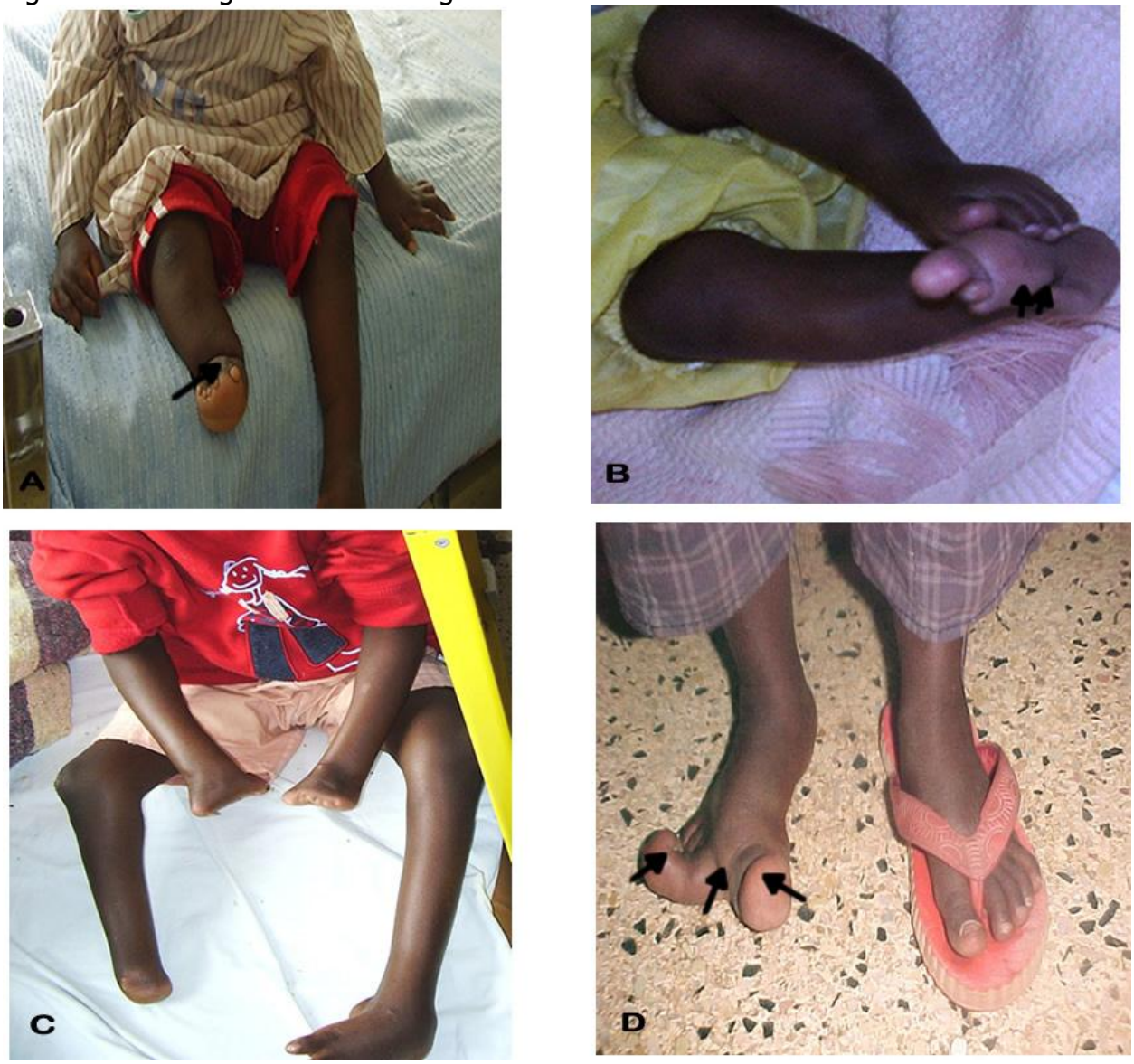
A: Four years old with brachydactyly

B: Three years old diagnosed with left diplopodia

C: Nine years old with congenital amputation as a result of amniotic band syndrome affecting all the limbs

D: Six years old boy with cleft foot with macrodactyly of the 1st and 2nd digits

Figure 2: Showing defects affecting lower limbs



Congenital constriction band syndrome: Also called Amniotic band syndrome (ABS). This group of defects is considered to be caused by rapture of the amnion with secondary effects on the fetus producing malformation due to interruption of normal morphogenesis, deformation due to distortion of established structures and disruption of structures already formed. It is characterized by a wide range of congenital anomalies including annular constrictions of the extremities, acrosyndactly, talipes equinovarus, cleft lip/palate and haemangiomas (Goldfarb et al., 2009). It may be associated with distal lymphedema, localized gigantism, congenital tibial pseudoarthrosis, localized gigantism and neurosensory deficits. Other phenotypic features of

this syndrome include digital imputation, cranial/ CNS defects, limb deficiency/amputation, body wall defects and short cord, craniofacial clefting, thoracoabdominoschisis, limb defect adhesion, placental adhesions and internal malformations (Halder, 2010; Agashe et al., 2011).

Syndactyly: This is the most common congenital malformation of the limbs, with an incidence of 1 in 2000 -3000 live births. It is caused by deficient apoptosis during digit sculptation in the 6^{th} and 8^{th} weeks of embryonic development. It can be classified as simple when it involves soft tissues only and complex when it involves bone or nail of adjacent fingers. Complex forms can include bony abnormalities such as extra, missing or duplicated phalanges. Abnormally shaped bones such as delta phalanges, abnormalities in the musculotendinous and neuro vascular structures may also be present. Syndactyly can be an isolated finding, or be found in association with other abnormalities such as polydactyly, cleft hands/feet, ring constrictions, or craniofacial syndromes as in Apert syndrome. Another syndrome is Poland syndrome in which pectoralis major muscle abnormality is found in association with syndactyly and /or other anomalies of the ipsilateral upper extremity . Constriction band syndrome may also be associated with syndactyly (Upton, 1991).

Cleft foot: This involves the central rays of the distal division of the foot, and presents with syndactyly, median clefts, aplasia and or hypoplasia of the phalanges and metatarsals. The fundamental defects is a failure of the apical ectodermal ridge to direct the outgrowth and patterning of the developing limb. It can occur in isolation, but frequently occurs as part of ectrodatyly- ectodermal dysplasia-cleft syndrome characterized by ectrodactyly, syndactyly, atypical anhydroric ectodermal dysplasia; cleft lip with or without cleft plate; lacrimal duct abnormalities, urogenital abnoramalities Omphalocele and anal atresia (De Smet and Fryns 1995) and a wide variety of other limb defects (Majewski et al., 1985; Roelfsema and Cobben, 1996; Wong et al 1998). It has a genetic basis and specific loci have been identified (Buss, 1994).

Age and gender distribution: The mean age of 9 years observed in the present study compares favorably with 9.9 years reported in literature (Yigister et al., 2005) and is expected considering that these conditions are congenital. In the case of tibial pseudoarthrosis, although the disease becomes evident within a child's first year of life, it may be undetected up to the age of 12 years (Roach et al., 1993).

The gender distribution observed reveals that it depends on the defect, which implies a genetic predisposition. The association of both tibial pseudoarthritis and macrodactyl with Neurofibromatosis suggests that they are heritable conditions (Stevenson et al., 1999). In our finding, the mean age for tibial pseudoarthrosis was 10.2 years

Conclusion:

There is wider variety of lower limb defects in Kenyan hospitals than usually reported in other studies. Tibial pseudoarthrosis is most common defect. Medical personnel should be aware of this wider scope of limb defects. In addition, greater emphasis should be given to systemic examination of the affected children and rehabilitative management.

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Conflict of interest: None

References:

1. Abdi –Rad I , Khoshkalam M, Farrokh-Islamlou HR. 2008. The prevalence at birth of overt congenital anomalies in Urmia North western Iran Arch Iran Med 11: 148 -151
2. Agashe M, Rathod CM, Dhamele JA. 2011. Congenital Pseudoarthrosis of the tibia with localized gigantism in a case of congenital constriction band syndrome. Ind J plast surg 44: 139 -141
3. Agha MM, William JI, Marret L To TI, Dodds L. 2006. Determinants of survival in children with severe congenital anomalies. A long term population based cohort study. Birth defects Res A in Mol Teratol 76: 46-54
4. Al-Hadidy A, Haroun A, Al-Ryalat N, Hamamy H, Al-Hadidi S. 2007. Congenital Pseudoarthrosis associated with venous malformation. Skeletal radiol 36: 515-518
5. Belthur MV. Linton JL, Barnes DA. 2011. The spectrum of pre axial polydactyly of the foot . J pediatric orthop 31 : 435-447
6. Byron - Scott R, Sharpe P, Hasler C. 2005. South Australian population based study of congenital talipes equinovarus. Paediatr perinat epidemiol 19: 227-237.
7. Buss PW. 1994. Cleft hand/ foot; Clinical and developmental aspects. J. Med Genet 31:726 – 730.
8. De Smet L, Fryne JP. 1995. Anal Atresia and abdominal wall defect as unusual symptoms of EEC syndrome . Genet counsel 6: 127-130
9. Galois L, Mainard D, Delagoutte JP. 2002. Polydactyly of the foot. Literature review and case presentations. Acta orthop Belg 68: 376-380.
10. Goldfarb CA, Sathienkijkanchai A, Robin NH. 2009. Amniotic constriction band: A multidisciplinary assessment of aetiology and clinical presentation. J Bone joint Surg (AM) 68-75
11. Halder A. 2010. Amniotic band Syndrome and /or limb body wall complex: split or lump the Appl chin Genet 3: 7-15.
12. Jones D, Barnes J, Lloyd-Robers GC. 1978. Congenital aplasia and dysplasia of the tibia with intact fibula. J Bone Joint Surg (Br) 60-B: 31-39

13. Khan F, Shah SA, Khan NU, Ullah I, Ullah F. 2010. Our experience with macrodactyly: a rare congenital anomaly. Pak J Surg 26 : 242-245
14. Levin SM, Lambiase, RE, Petchpara CH. 2003. Cortical lesions of the tibia: characteristics appearance at conventional radiography. Radiographics 23: 157-177
15. Magee T, Mackey Dr, Segal LS. 2007. Congenital constriction band with Pseudoarthrosis of the tibia: a case report and literature review. Acta orthop Belg 73: 275-278
16. Majewski F, Kuster W, Ter haar B, Goecke T. 1985. Aplasia of tibia with split hand/split foot deformity, report of six families with 35 cases and considerations about variability and pentrance. Hum Genet 70: 136-147
17. Malik S, Afzal M, G and S, Wahab A, AAhmad M. 2010. Autosomal dominant syndrome of comptodactyly, clinodactyly, syndactyly and bifid toes. Am J med Genet A 152 A : 2313-7
18. Muga Ro, Mumah SCJ, Juma PA. 2009. Congenital malformations among new borns in Kenya. Afr J food Agric Nutric Dev 19: 819-829.
19. Mysorekar VR, Lohokare Sk. 1970. A case of eight toed foot. Br J Radiol NS 43: 740-2
20. Narang IC, Mysorekar, Marthur BP. 1982. Diplopodia with double fibula and agenesis of tibia. J Bone joint surg 64-B: 206-209.
21. Ogeng'o JA, Obimbo MM, King'ori J. 2009. Pattern of limb amputation in Kenyan Rural Hospital. Int Orthop 33:1449 – 1453.
22. Roach JW, Shindell R, Green NE. 1993. Late onset pseudoarthrosis of the dysplastic tibia. J. Bone J Surg Am 75 – A: 1593 – 1601.
23. Roelfema NM, Cobben JM. 1996. The EEC Syndrome: A literature study . Chin Dysmorphol 5: 115-127.
24. Sharma S, Vyas S, Sood RG, Auupam KN. 2006. Macrodactyly: report of 3 cases. Ind J Radiol Imag 16: 583-584.
25. Siapkara A, Duncan R. Congenital talipes equinovarus. 2007. A review of current management. J Bone Joint Sugr (Br) 89B: 995 – 1000.
26. Stevenson DA, Birch PH, Friendman JM, Viskochil DH, Balestrazzl P, Boni S, Buske A, Korf BR. 1999. Descriptive analysis of tibial pseudoarthrosis in patients with Neurofibromatosis 1. Am J Med Gen 84: 413 – 419.
27. Syed A, Sherwani R, Azam Q, Haque F, Akhter K. 2005. Congenital macrodactyly: A clinical study . Acta orthop Belg 71: 399-404.
28. Tayel SM, Fawzia MM, Niran AA, Said G, Al Awadi SA, Naguib KK. 2005. A morpho-etiological descritption of congenital limb anomalies. Ann Saudi Med 25: 219-227.
29. Upton J. 1991. Apert syndrome classification and pathologic anatomy of limb anomalies Clin plast surg 18: 321-355.
30. Watanabe H, Fujita S, Oka I. 1992. Polydactyly of the foot; an analysis of 265 cases and a morphological classification. Plast reconstruct surg 89: 856-877

31. Wong Sc, Cobben JM, Hiemstria S, Robinson PH, Heeg M,. Karsch. 1998. Neugebaur syndrome in two sibs with unaffected parents, Am J Med genet 75: 207-210.
32. Yigister K, Ulger O, Sener G, Akdogass S, Erbahceci F, Bayar K. Demography and function of children with limb loss. Prosthet orthot Inte 2005; 29:131-138.
33. Yuksel A, Yagnut H, Kural BS. Prenatal diagnosis of isolated macrodactyly. Ultrasound obstet gynaecol 2009; 33: 360-362