大 韓 放 射 線 醫 學 會 誌 第25卷 第6號 pp. 988~992, 1989
Journal of Korean Radiological Society, 25(6) 988~992, 1989

Macrodystrophia Lipomatosa

Sun Wha Lee, M.D., and Jae Hoon Lim, M.D.

Department of Diagnostic Radiology, Kyung Hee University Hospital

〈國文抄錄〉

進行性 脂肪腫性 巨大症

慶熙大學校 醫科大學 放射線科學敎室

李 善 和·林 在 勳

進行性 脂肪腫性 巨大症은 불명료한 病因의 드문 先天性 畸形으로 손가락이나 발가락의 全間葉織 細胞들이 過度하게 發育하며 特히 纖維脂肪組織의 不均一한 增加가 特徵이다.

著者들은 4例의 進行性 脂肪腫性 巨大症을 經驗하여 他 形態의 巨指症과 구별되는 放射線學的 所見을 文獻考察과 함께 報告하는 바이다. 放射線學的으로 指部의 軟組織 및 指骨의 增大, 彎指, 軟組織內 放射線透通의 所見이 觀察되었다.

— Abstract —

Macrodystrophia lipomatosa is a unusual and bizarre congenital malformation of obscure etiology and pathogenesis, and characterized by progressive overgrowth of all the mesenchymal elements of a digit or digits, with a disproportionate increase in the fibroadipose tissues. Authors experienced four cases of macrodystrophia lipomatosa(three cases in the fingers and one case in the toe)and describe its radiologic and pathologic findings which differentiated this entity from other forms of macrodactyly. Radiologic findings include enlargement of the phalanges and soft tissue elements of the digit with a predominant involvement in distal part, clinodactyly, and radiolucencies in the soft tissue representing the dramatic overgrowth of fat.

Index Words: Hand, abnormalities
Gigantism

Introduction

Macrodystrophia lipomatosa is a rare form of localized giantism characterized by progressive overgrowth of all the mesenchymal elements of a digit or digits, with a disproportionate increase in the fibroadipose tissues. It is classified as a developmental anomaly and usually recognized at birth or in the neonatal period.

Authors have experienced 3 cases of macrodystrophia lipomatosa involving the fingers and 1 case in the toe, and wish to present the character-

이 논문은 1989년 9월 11일 접수하여 1989년 9월 20일에 채택되었음
Received September 11, Accepted September 20, 1989

istic radiolgical and pathological findings of this unusual entity, with emphasis on differentiation from other forms of localized giantism.

Materials and Methods

The hospital charts, radiologic examination(plain radiography in all cases and angiography in one case), and surgical specimens operated at the Kyung Hee University Hospital were reviewed. No surgery has been performed to date in one case.

On gross examination, affected part is increased in length and width, and the skin is thick with a large nail(Fig. 1–a). The involved digit had normal function except for one case(Case 2) Who had limited flexion at distal interphalangeal joint of the middle finger. Neither the patients nor their families had neurocutaneous manifestations of neurofibromatosis and other congenital abnormalities do not occur.

Results

Table 1 summarize the clinical and radiologic characteristics of four cases of macrodystrophia li-

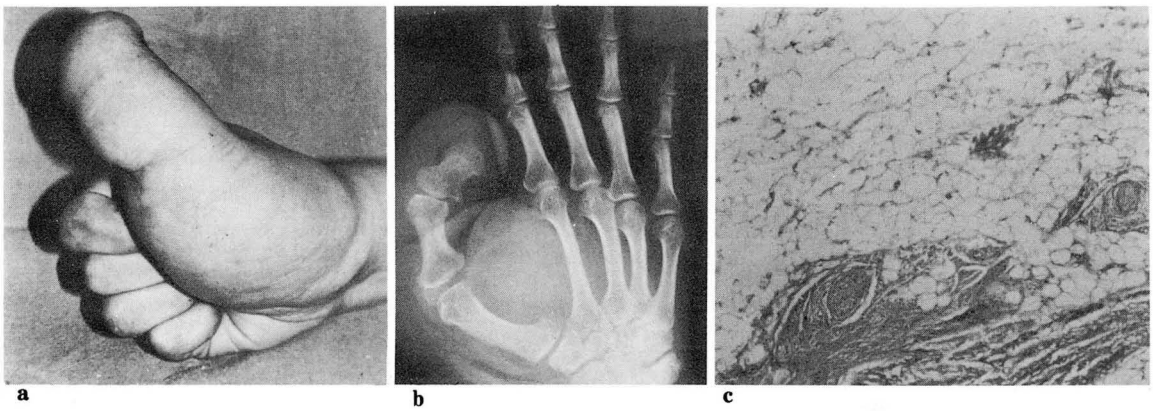

Fig. 1 Case 1. (a) Photograph shows markedly enlarged thumb. (b) Radiograph reveals long and broad phalanges of the thumb and first metacarpal bone with splayed distal end and clinodactyly. Marked enlargement of soft tissue with lucent areas are apparent. (c) Microscopic examination of amputated thumb demonstrates proliferation of the mature fatty tissues surrounding and splaying the nerves(H & E, X40).

Table 1. Summary of Four Cases of Macrodystrophia Lipomatosa

Case No.	Age(y)/Sex	Digit Involved	Radiologic Findings			Remarks
			Overgrowth of soft tissue/bone	Clino-dactyly	Soft tissue lucencies	
1	24/F	Rt. thumb	+	+	+	Amputation
2	22/M	Rt. index finger	+	+	−	Soft tissue
		Rt. middle finger	+	+	−	excision
3	8/M	Rt. second toe	+	+	+	DIP disarticulation & soft tissue resection
4	20/F	Lt. index finger	+	+	+	
		Lt. middle finger	+	+	+	No operation

DIP: distal interphalangeal joint

pomatosa.

In all cases, the plain radiographs of involved digit demonstrated various enlargement of both osseous and soft tissue structures with the most severely affected change in distal end of digit. The soft tissue hypertrophy was more obvious than osseous hypertrophy. Clinodactyly was present in all cases(Fig. 1–b, 2–a, b). Various degree of secondary degenerative change of interphalangeal joint with slanted articular surface and hypertrophic change was observed. Radiolucencies were apparent in the hypertrophied soft tissue of 3 cases(Fig. 1–a). The brachial angiograph performed in case 2 showed faint vascular pooling from right palmar metacarpal artery over second-third interosseous space and somewhat larger palmar digital arteries in index and middle finger than other digital arteries. There is no definite neovascularity or abnormal arteriovenous communications.

Histologic sections were available in 3 cases and confirmed the diagnostic infiltration of mesenchymal elements by fibroadipose tissues and enlarged nerves with fatty infiltration of the sheath(Fig. 1–c, 2–c).

Discussion

Macrodystrophia lipomatosa was first described by Feriz in 1925, and since then there have been 76 cases reported in the literature[1~4]. The causes of this anomaly reported here remains obscure and suggested theories include lipomatous degeneration, disturbed fetal circulation, an error in segmentation, the trophic influence of a tumefied nerve and the in utero disturbance of a growth-limiting factor[1,4,5].

It is almost invariably recognized at birth or in the neonatal period. The rate of accelerated growth of lesion varies among patients and even among affected digits. However growth of involved digit ceases with puberty. Involvement is usually unilateral and when more than one digit is involved the digits are invariably adjacent to one another. The predilection for involvement are second and third digits, corresponding to the territory of the median nerve in the upper extremity and the medial plantar nerve in the lower extremity, which is unexplained, and so "nerve territory oriented macrodactyly" has been suggested as a new synonym[6].

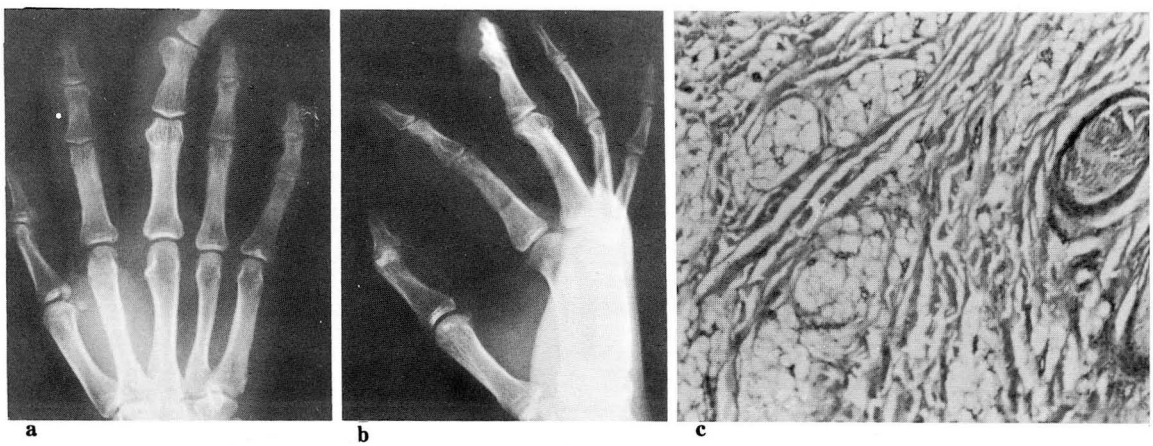

Fig. 2 Case 2. AP (a) and oblique (b) views of the hand show osseous and soft tissue enlargement of the index and middle fingers affecting predominantly the middle finger, degenerative change at DIP joint, and dorsal angulation of the involved digits produced by volar enlargement of soft tissues. (c) Histologic specimen reveals the fibroadipose tissues that surrounded and infiltrated the epineurium of the nerve bundles(H & E, X40).

Patients with macrodystrophia lipomatosa most frequently seek surgical correction for cosmetic purpose even though there has been reported cases with mechanical interference of joint function, vascular supply and innervation. Mechanical problems are not encountered until later in childhood or adolescence, when secondary degenerative changes reduced joint function, and large osteophytes result in compression of the neurovascular structures[1,4,5].

The most distinctive pathologic finding is the increase in adipose tissue interspersed in a fine mesh of fibrous tissue, which involves the bone marrow, periosteum, muscles, nerve sheaths, and subcutaneous tissues. Neural enlargement and irregularity may be prominent, which is caused by infiltration of the sheath by fibroadipose tissue, not by an increase in the number of axons. The phalanges are enlarged by both endosteal and periosteal deposition of the bone. The periosteum is studded with 1mm nodules consisting of chondroblasts, osteoblasts and osteoclasts which become larger and more numerous toward the distal ends of the phalanges[1,3,4,7].

The radiologic findings of macrodystrophia lipomatosa are both soft tissue and bony overgrowth[2~6]. The soft tissue overgrowth is most marked at the distal end of the digit and along its volar aspect which causes dorsal deviation of the affected parts. The radiolucencies within soft tissue lesion reflecting dramatic overgrowth of fat characteristic of this anomaly is usually observed. The involved phalanges are long, broad, and often splayed at their distal ends. The articular surfaces may be slanted, and in the late childhood or adolescence secondary degenerative joint supervenes. The osteophytes and reactive new bone formation are disproportionately large in relation to the joint space narrowing, which may be the result of the periosteal nodules associated with overgrowth[4]. The clinodactyly is almost invariably present, which is caused by the side-to-side variation in the accelerated rate of growth[2,4]. There is a high incidence of associated local anomalies, including syndactyly, symphalangism and polydactyly[1,2,3] but absent in authors cases.

The radiologic differential diagnosis of localized giantism includes both congenital and acquired disorders, and listed in the table 2[4,6]. On the basis of the history the acquired causes can be eliminated. The majority of congenital causes can also be excluded. Hemangiomatosis and lymphangiomatosis produce soft tissue hypertrophy and symmetric overgrowth of the bone, which related to hyperemia secondary to tumorous overgrowth. The possibility of Ollier's disease and Klippel-Trenaunay-Trenaunay-Weber syndrome can be excluded by absence of enchondromas and obvious cutaneous manifestations, respectively. The most difficult differential diagnosis, both radiologically and pathologically, is neurofibromatosis. Macrodactyly in Recklinghausen's disease attributes to flexiform neurofibromas(with hemangiomatous and lymphangiomatous elements), combined with a mesodermal dysplasia. Neurofibromatosis can be differentiated form macrodystrophia lipomatosa by several

Table 2. Classification of Macrodactyly

Congenital Disorders	Acquired Disorders
Increase in all mesenchymal elements: macrodystrophia lipomatosa hyperostotic macrodactyly	Dactylitis producing hypervascularity: infection infarction trauma Still's disease
Tumorous overgrowth of a single elements: hemangioma lymphangioma Ollier's disease plexiform neurofibroma Klippel-Trenaunay-weber syndrome arteriovenous malformation	Hypervascular tumor: osteoid osteoma Melhorrheostosis

Note. Adapted from reference 4 and 6.

radiological findings. First, involved digits may be bilateral and not invariably contiguous. The distal phalangeal involvement is not always the most severe. Second, premature fusion of the growth plate can be observed, which may be related to hemangiomatous elements of the flexiform neurofibroma. Third, the enlarged osseous structures in neurofibromatosis may have a wavy cortex and an elongated sinuous appearance. Last, the lucencies can not be observed within soft tissue hypertrophy[2,4,6,8].

Summary

Four cases of macrodystrophia lipomatosa are reported with brief review of literature.

REFERENCES

1. Barsky AJ: *Macrodactyly. J Bone Joint Surg(Am) 49:1255-1266, 1967*
2. Thijn CJP: *Radiology of the hand lst ed. Berlin, Springer-Verlag, 1986*
3. Moran B, Butler F, Colville J: *X-ray diagnosis of macrodystrophia lipomatosa. Br J Radiology 57:523-526, 1984*
4. Goldman AB, Kaye JJ: *Macrodystrophia lipomatosa: radiographic diagnosis. AJR 128:101-105, 1977*
5. Kleikian H: *Macrodactyly in congenital deformities of the hand and forearm. Philadelphia, WB Saunders, 1974*
6. Resnick D, Niwayama G: *Diagnosis of bone and joint disorders, 2nd ed. Philadelphia, WB Saunders, 1988*
7. Minkowitz S, Minkowitz F: *A morphological study of macrodactylism a case report. J Pathol Bacteriol 90:323-325, 1965*
8. Pitt MJ, Mosher JF, Edeiken J: *Abnormal periosteum and bone in neurofibromatosis. Radiology 103:143-146, 1972*