CAMPTODACTYLY : A PHENOTYPE OF DUPUYTREN'S CONTRACTURE

Kamlender Singh and Vijay R Battu

A 15-year-old boy had permanent flexion contractures at proximal interphalangeal joints of the little and ring fingers of both hands, along wit' slight hemiatrophy of face and bilateral mild ptosis. The hand deformity superficially resembled Dupuytren's contracture. This combination of features does not fit with the syndromes described so far, whose one of the components is camptodactyly.

Key words : Camptodactyly, Hemiatrophy of face, Ptosis.

Camptodactyly is an inherited anomaly characterised by permanent flexioncont ractures at the proximal interphalangeal joints. The little finger is affected in practically all instances, but the ring, middle and index fingers may also be involved. Thumb and the other interphalangeal joints are rarely affected. There is no limitation to further flexion of the fingers, but complete extension is not possible. This disorder is usually bilateral and is present at birth, though it may go undetected till early childhood.[1] Possible etiological explanations include, congenitally contracted flexor sublimis, abnormal expansion on the back of first interphalangeal joint, volar tilt of the distal portion of the proximal phalanx, and finally, contraction of anterior capsule of the joint. Of these, contraction of both slips of flexor sublimis have been observed during surgery, though it is a moot point whether it is consequential or causative.[2] Herein is reported a case which hitherto had been considered to be a case of Dupuytren's contracture with which it has a marked superficial resemblance.

Case Report

A 15-year-old male high school student, born to non-consanguinous parents, noticed a flexion deformity of the little and ring fingers of both the hands, of 5 years duration. This had developed gradually over the first 3-4 years and was now almost stationary. The flexion deformity initially affected the little fingers and later the ring fingers. He had occasional, transient episodes of tingling and burning along the ulnar border of his hands and forearms which would last a few minutes. During heavy manual work, he would experience pain in the affected fingers, though otherwise, the disability did not interfere with his less strenuous work. He had no other symptoms. There was no history of preceding trauma. He had sustained fractures of right radius and

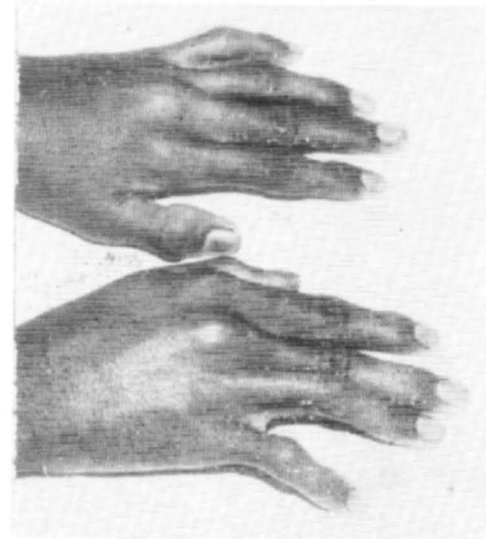

Fig. 1. Flexion contracture of little and ring fingers at the proximal interphalangeal joint of both hands.

From Department of Dermatology and Venereology, All India Institute of Medical Sciences, New Delhi-110 029, India.

Address correspondence to : Dr. Kamlender Singh, A/2 Swasthya Vihar, Vikas Marg, New Delhi-110092, India.

right clavicle 4 and 3 years ago respectively and these had healed uneventfully. There was no history of a similar disorder in his blood relatives.

The little and ring fingers of both hands were equally deformed, slender and had tight skin; the former were much more affected than the latter. Flexion at the proximal interphalangeal joints of the little fingers was almost 90 degrees. Metacarpophalangeal and distal interphalangeal joints were unaffected (Fig. 1). Fingers could be flexed further at the affected joints, but forceful extension was not possible and it produced a taut web (Fig. 2). Except for a very

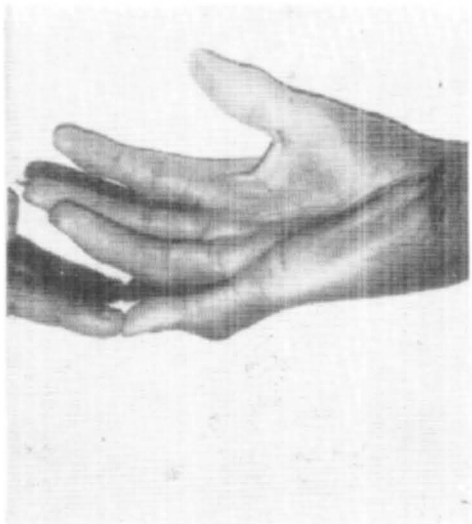

Fig. 2. A taut web between the fingers.

mild thickening near the bases of the little and ring fingers, the palmar skin was of normal texture and thickness, with no puckering or hard nodulation. There was no contractural band beneath the volar skin of the affected joints, but its transvere creases were less. There was only one major horizontal palmar crease corresponding to the simian crease. Intrinsic muscles of the hands were normal except for a mild wasting of the hypothenar muscles. There was no sensori-motor deficit, nor any thickened and/or tender nerve or any hypopigmented lesion on the skin. The face was slightly

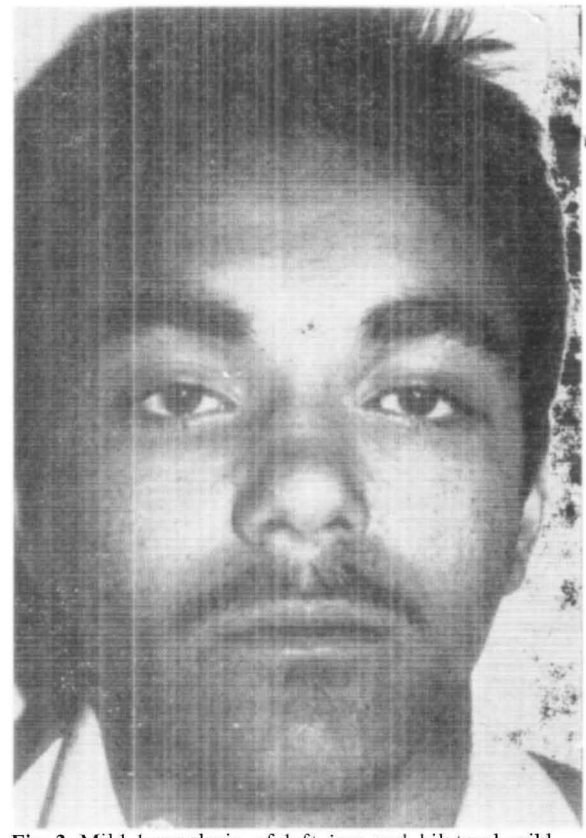

Fig. 3. Mild hypoplasia of left jaw and bilateral mild ptosis.

asymmetrical due to hypoplasia of the left jaw (Fig. 3). There was no sensori-neural deafness. General physical and systemic examination including the ophthalmological examination did not reveal anything abnormal except for bilateral mild ptosis.

Hemogram, routine and microscopic urine and stools analysis, urine aminoacidogram, blood sugar, serum electrolytes, blood urea, serum proteins, electromyogram, and motor nerve conduction velocity were within normal limits. X-ray of the skull revealed mild hypoplasia of left mandible. X-ray of the hands and chest were within normal limits.

Comments

Diagnosis of camptodactyly was made on the basis of characteristic clinical features,

namely flexion deformity at the proximal interphalangeal joints, essentially normal texture of palmar skin, loss of transverse skin creases, earlier age of onset and slight progression. On the other hand, its phenotype, Dupuytren's contracture is characterised by greater involvement of the ring finger, contracting bands, loss of subcutaneous fat, palpable hard nodules, induration in the skin, transverse skin folds, crescentic puckerings, onset between 40-60 years and continued progression[1] Other associated findings in Dupuytren's contracture include, similar changes in the foot in 5% of the cases, Peyronie's disease in 3% of cases and epilepsy.[3]

Camptodactyly can occur as an isolated anomaly. This variety is believed to be autosomal dominant with a variable penetrance and expressivity[4] though holandric (Y-linked) inheritance[5] and X-linked dominant inheritance[6] have also been proposed for some cases.

Camptodactyly can also occur as a component of other syndromes as in autosomal dominant whistling face syndrome or cranio-carpo-tarsal dysplasia;[7,8] probably autosomal dominant syndrome of camptodactyly, dwarfism, hypogonadism, pectus carinatum and ptosis;[9] autosomal recessive Goodman camptodactyly syndrome A;[10] autosomal dominant Gorden syndrome of camptodactyly, cleft palate and club foot;[11] autosomal recessive Christian adducted thumb syndrome;[1] autosomal dominant camptodactyly and sensori-neural hearing loss;[12] autosomal recessive camptodactyly V, ectodermal dysplasia and sensori-neural hearing loss;[13] Lichtenstein syndrome of unknown inheritance;[14] probably autosomal recessive syndrome of camptodactyly, multiple ankylosis, facial anomalies and pulmonary hypoplasia;[15,16] autosomal dominant camptodactyly and congenital absence of finger prints;[17] autosomal recessive cerebral hepato-renal syndrome;[18,19] Nielson syndrome which is probably x-linked;[1] Golden-Lakim syndrome which is probably autosomal dominant;[20] autosomal dominant pterygium multiplex universalis[1] and the 10 trisomy syndrome due to chromosomal aberration.[21]

The mode of inheritance in our case is a matter of speculation as there was no history of similar disorder in the blood relatives of the patient. Camptodactyly in our case was associated with mild hemiatrohy of the face and mild ptosis. Asymmetry of face and ptosis have been observed in cranio-carpo-tarsal dysplasia.[7] However, there were no other malformations of cranio-carpo-tarsal syndrome which include a high skull compressed fronto-dorsally and broadened in the frontal plane, arching of frontal bone in the midline, deeply set eyes occasionally associated with squint, blepharophimosis and epicanthosis or ptosis, relatively narrow nose with narrow nostrils and bent alae, microstomia with protrding lips as if whistling and reduced electromyographic activity. In the upper limbs, smaller muscle mass, restricted mobility of shoulders, limited pronation and supination of forearms is characteristic. In the lower limbs bilateral talipes equinovarus and tightly contracted toes are features. In the absence of all these features, our case can at best be called a 'forme fruste' case of cranio-carpo-tarsal dysplasia, or alternatively, it is a new syndrome as it does not correspond with any syndrome described so far.

Incidence of camptodactyly as an isolated anomaly is unknown for our country. In the West, it has been observed in 7 out of 800 school children by one group and in 31 out of 6000 school children by another group.[1] Incidence of the associated ones is naturally very low. Though most authors mention the isolated variety to be more frequent in girls, a survey by Welch and Temtamy[4] of 303 reported cases suggested that the condition may be equally distributed between the sexes.

Treatment is rarely sought as it is usually a mild digital malformation interfering little with the function of hand. This is perhaps an im-

portant reason why camptodactyly especially the one occurring as an isolated anomaly is rarely seen and is thus frequently found by chance among patients presenting for other probably unrelated conditions. During childhood, treatment can be of benefit. The deformity can be corrected by reducing the tension in the superficialis tendon by flexing the wrists. In severe progressive forms, lengthening of the rogue tendon at its musculo-tendinous junction may ameliorate the contition.[22] In the adults the contractures become fixed and a completely normal digit cannot be restored[22] whence arthodesis at the first interphalangeal joint placing the finger in a servicible position, can correct the deformity.[2,22] Other useful procedures include Z-plasty of the palmar skin supplemented with anterior capsulotomy of the first interphalangeal joint in moderate cases. In severe cases subcapital osteotomy or resection of distal end of proximal phalanx can be ameliorative.[2]

References

1. Temtamy SA and McKusick VA : Contractural deformities, in : The Genetics of Hand Malformations, Birth Defects : Original Article Series, The National Foundation– March of Dimes, Alan R Liss Inc, New York, 1978; XIV (3) : 441-479.
2. Kelikian H : Anomalies of digital posture, in : Congenital Deformities of the Hand and Forearm, Editor, Kelikian H : WB Saunders Company, Philadelphia, 1974; p 555-603.
3. Early PE : Population studies in Dupuytren's contracture, J Bone Joint Surg, 1962; 44B : 602-613.
4. Welch JP and Temtamy SA : Hereditary contractures of fingers (Camptodactyly), J Med Genet, 1966; 3 : 104-113.
5. Stern C : The problem of complete y-linkage in man, Amer J Hum Genet, 1957; 9 : 147-150.
6. Parish JG, Horn DB and Thompson M : Familial streblodactyly with aminoaciduria, Brit Med J, 1963; 2 : 1247-1250.
7. Burian F : The 'whistling face' characteristic in a compound craniofacio-corporal syndrome, Brit J Plast Surg, 1963; 16 : 140-145.
8. Fraser FC, Pashayan H, and Kadish ME : Craniocarpotarsal dysplasia, Report of a case in father and son, JAMA, 1970; 211 : 1374-1376.
9. Hanley WB, McKusick VA and Barranco FT : Osteochondritis dissecans and associated malformations in two brothers, A review of familial aspects, J Bone Joint Surg, 1967; 49A : 925-937.
10. Goodman RM, Miriam B, Katznelson M et al : Camptodactyly– occurrence in two new genetic syndromes and its relationship to other syndromes, J Med Genet, 1972; 9 : 203-212.
11. Gorden H, Davies D and Berman M : Camptodactyly, cleft palate and club foot : A syndrome showing the autosomal-dominant pattern of inheritance, J Med Genet, 1969; 6 : 266-274.
12. Stewart JM and Bergstrom L : Familial hand abnormalities and sensorineural deafness, A new syndrome, J Pediat, 1971; 78 : 102-110.
13. Michaelian DO, Kaloustian VMD, Shahin NA et al : Congenital ectodermal dysplasia with hearing loss, Arch Otolaryngol, 1970, 92 : 85-89.
14. Lichtenstein JR: A "new" syndrome with neutropenia, immunoglobulin deficiency, peculiar facies and bony anomalies, in : Blood Birth Defects : Original Articles Series, White Plains, The National Foundation, 1972; VIII (3), p 178.
15. Pena SDJ and Shokeir MHK : Syndrome of camptodactyly, multiple ankylosis, facial anomalies and pulmonary hypoplasia : a lethal condition, J Pediat, 1974; 85 : 373-375.
16. Punnett HH, Kistenmacher ML, Valdes-Dapena M et al : Syndrome of ankylosis, facial anomalies and pulmonary hypoplasia, J Pediat, 1974; 85 : 375-377.
17. Baird HW : Kindred showing congenital absence of the dermal ridges (finger prints) and associated anomalies, J Pediat, 1964; 64 : 621-631.
18. Smith DW, Opitz JM and Inhorn SL : A syndrome of multiple developmental defects including polycystic kidneys and intrahepatic biliary dysgenesis in two siblings, J Pediat, 1965; 67 : 617-624.
19. Passarge R and McAdams AJ : Cerebro-hepato-renal syndrome : a newly recognised hereditary disorder of multiple congenital defects, including sudanophilic leukodystrophy, cirrhosis of the liver and polycystic kidneys, J Pediat, 1967; 71 : 691-702.
20. Golden RL and Lakim H : The forme fruste in Marfan's syndrome, N Eng J Med, 1959; 260 : 796-801.
21. Yunis JJ and Sanchez O : A new syndrome resulting from partial trisomy for the distal third of the long arm of chromosome 10, J Pediat, 1974; 84 : 567-570.
22. Fixen JA and Watson M : Some other developmental abnormalities in the extremities, The upper limb, in : Postgraduate Textbook of Clinical Orthopedics, Editor, Harris NH : John Wright & Sons Ltd, Bristol, 1983; p 169-183.