Case Report / Olgu Sunumu

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Familial Peripheric Polyneuropathy Plus Camptodactyly; Three Sisters

Ailesel Kamptodaktili ve Periferik Nöropatili Üç Kız Kardeş

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Summary

Camptodactyly, the flexion contracture of the proximal interphalangeal joint usually involves the small finger and may be a component of some autosomal dominant diseases, such as Marfan's syndrome, cranio-carpo-tarsal dystrophy and oculo-dento-digital dysplasia. Camptodactyly may also coexist with anomalies, such as high arched palate, anomalies of scapula, scoliosis, ptosis, hemi-hypertrophy and taurinuria. Hereditary autonomic and sensory neuropathies (HSAN) are a clinically and genetically heterogeneous group of inherited peripheral neuropathies, which primarily affect the peripheral sensory and autonomic nerves. Patients usually have prominent distal sensory loss with complaints of insensitivity to pain. Prominent distal sensory loss may cause chronic ulcerations in the feet and hands, and less frequently, severe complications as extensive soft tissue infections, osteomyelitis and amputations.

Herein, we report three sisters with hereditary peripheral polyneuropathy associated with concomitant camptodactyly. *Turk J Phys Med Rehab 2012;58:72-4.*

Key Words: Hereditary neuropathy, camptodactyly

Özet

Kamptodaktili proksimal interfalangeal eklemlerin fleksiyon kontraktürü olup genelde küçük parmağı tutar ve Marfan sendromu, kranio-karpo-tarsal distrofi, okülo-dento-dijital displazi gibi bazı otozomal dominant sendromların parçası olabilir. Kamptodaktili yine yüksek damak, skapula anomalileri, skolyoz, ptozis, hemi hipertrofi ve taurinüriye de eşlik edebilir. Herediter otonomik ve sensoriyal nöropati (HSAN) klinik ve genetik olarak heterojen, genetik geçişli bir grup periferik nöropati olup primer olarak periferik sensoriyal ve otonomik sinirleri etkiler. Hastaların genelde belirgin distal sensoryal his kaybı ve ağrı algısında bozulma şikayetleri vardır. Distal his kaybı, el ve ayaklarda kronik ülserlere ve daha nadir olarak yaygın yumşak doku enfeksiyonu, osteomyelit ve amputasyon gibi komplikasyonlara neden olabilir.

Biz bu yazıda, herediter periferik polinöropatiye komptodaktilinin eşlik ettiği üç kız kardeşi rapor ediyoruz. *Türk Fiz Tıp Rehab Derg 2012;58:72-4.*

Anahtar Kelimeler: Herediter nöropati, kamptodaktili

Introduction

Camptodactyly, the flexion contracture of the proximal interphalangeal joint, usually involves the small finger, and is related to a multiple etiologies; tightness of the skin or the underlying fascia or the tendon of the flexor digitorum superficialis. Most of the patients report little functional impairment, and usually a mild contracture. Therefore, if there are mild functional limitations, nonsurgical care is necessary and it is the first-line treatment, including serial casting and splinting. Others with moderate to severe contracture (60° to 90°) or those unresponsive to splinting may benefit from surgical interventions (1,2). The outcomes of surgery are not clear, therefore, surgery as a treatment option must be decided individually.

Foucher et al. (2) have established a treatment and a classification algorithm after evaluation of 155 patients. He

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classified most of the patients as early and stiff (<5 years of contracture and without passive extension); or according to time interval (early or late) and mobility (stiff or mobile). The small finger was mostly involved, and abnormalities related to bone of the proximal interphalangeal joint was noted in 29% of patients and in 58% of those with joint stiffness. Camptodactyly is also a part of some autosomal dominant diseases, such as Marfan's syndrome, cranio-carpo-tarsal dystrophy and oculo-dento-digital dysplasia. It may also coexist with anomalies such as high arched palate, anomalies of the scapula, scoliosis, ptosis, hemi-hypertrophy and taurinuria (3,4).

Hereditary autonomic and sensory neuropathies (HSAN) are a clinically and genetically heterogeneous group of inherited peripheral neuropathies, and primarily affect the peripheral sensory and autonomic nerves. Patients usually have prominent distal sensory loss with complaints of insensitivity to pain. Prominent distal sensory loss may cause chronic ulcerations in the feet and hands, and less frequently severe complications as extensive soft tissue infections, osteomyelitis and amputations (5). Electrophysiologically, axonal nerve damage of the sensory neurons is frequently shown, but additionally, demyelination may be present (6). HSAN can be transmitted as an autosomal dominant (AD) or autosomal recessive (AR) trait. The AD types of HSAN frequently present in the second or third decade of life with significant sensorial involvement and minimal autonomic and variable motor involvement (7). Dyck classified the hereditary sensory neuropathies into types as HSAN I-V based on the age at onset of disease, inheritance pattern and additional features (5). The motor involvement in the AD form of HSAN (type-1) vary a lot, which makes the distinction between hereditary motor and sensory neuropathies (HMSN) or Charcot-Marie-Tooth disease (CMT) difficult. Only type-1 HSAN is inherited autosomal dominantly, with an onset after the second decade of life. The genetic loci is on 9q22 and the gene is SPTLC1 for type-1 HSAN (5-7).

CMT is an inherited disorder of the nerves that has different forms. It is characterized by loss of muscle tissue and touch sensation. This disease is one of the most common inherited neurological disorders (36 /100.000) (8). CMT have remarkably similar symptoms, such as a high arched foot, structural foot deformities, progressive muscle wasting leading to problems with walking, running, and balance, ankle weakness and sprains and foot drop (8,9).

Herein, we report 3 sisters with peripheral polyneuropathy associated with concomitant camptodactyly.

Case

A 69-year-old female patient presented to our outpatient unit with diffuse joint pain. She had arthritis on the proximal interphalangeal, metocarpophalangeal, wrist and metotarsophalangeal joints. The complaints first begun 12 years ago and she was was diagnosed in another hospital as having rheumatoid arthritis. She described morning stiffness lasting 2 hours, and was also hypertensive. On the physical examination, bilateral camptodactyly on the second through fifth fingers was detected (Figure 1). She also had back pain, and lower extremity weakness going on for the last 25 years. Laboratory examination revealed sedimentation rate: 72 mm/hour, rheumatoid factor: 69.9 IU/ml (0-20) and C-reactive protein: 8.4 mg/dl (0-0.8). The blood biochemistry including the transaminases, the serum glucose level, and the renal function tests was all normal. The X-ray images of the hands demonstrated periarticular osteoporosis, narrowing of the metocarpophalangeal joints and the wrists. By the combination of chronic, symmetric polyarthritis of the hands, radiological and laboratory data, rheumatoid arthritis was diagnosed. An electromyelography (EMG) was ordered for the lower extremities to search the etiology of the weakness; and the report of the EMG was as chronic sensorial and motor polyneuropathy also affecting the axons. A disease modifying anti-rheumatic drug (DMARD) combination was prescribed for the patient and it was learned from

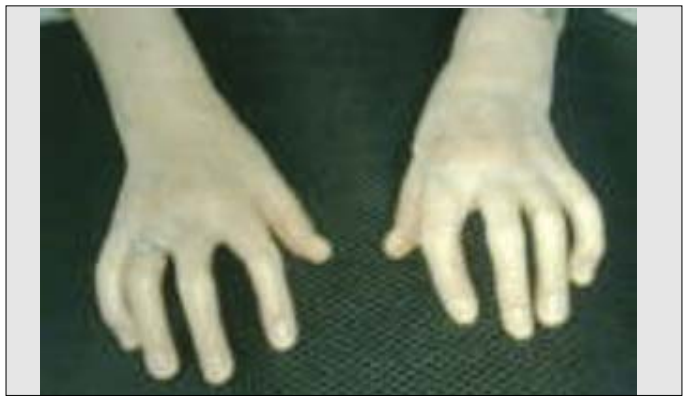

Figure 1. Case 1; the first patient with rheumatoid arthritis.

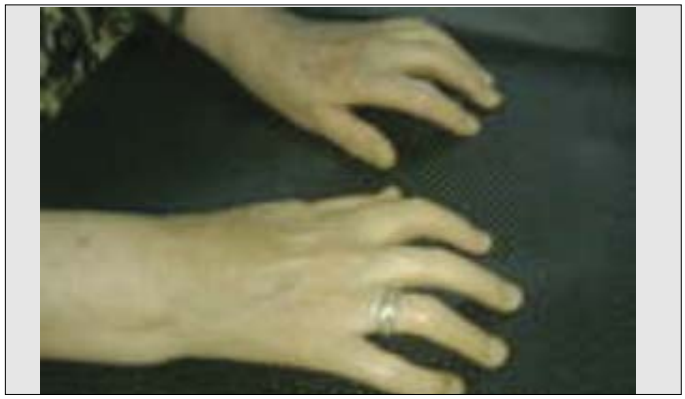

Figure 2. Case 2; the second patient.

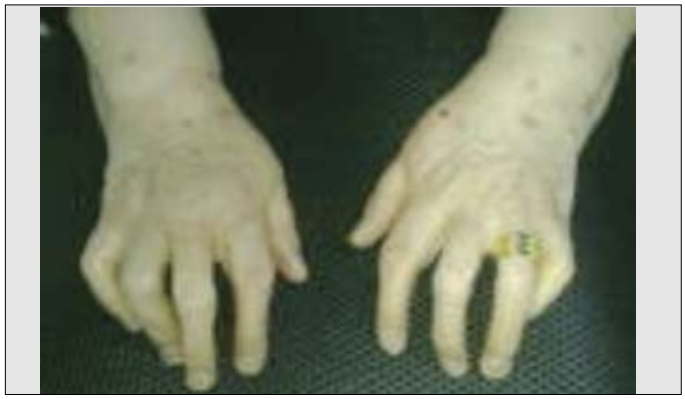

Figure 3. Case 3; the third patient.

the patient that she had 2 sisters with camptodactyly of the hands and also lower extremity weakness, as she had. They were also invited to our outpatient unit for a medical evaluation. The sisters were 65 and 62 years old. Both had the complaints of lower extremity weakness for the last 20 years, besides, they had bilateral camptodactyly on the second to fifth finger as their sister had (Figure 2 and 3). Both were examined by EMG and the results were same with the first case; chronic sensorial and motor polyneuropathy also affecting the axons. None of these patients were diabetic or had any other disease that can cause peripheral polyneuropathy. The blood biochemistry analysis of the sisters was all normal and the serologic tests for hepatitis A, B and C were negative. None of them were alcoholic or were using narcotics. Only the first case had rheumatoid arthritis but the others did not. Vitamin B 12 tests were not ordered because there were no megaloblastic anemia, nutritional deficiency, amnesia or other clinical situation to doubt about vitamin B 12 deficiency. Therefore, the family was diagnosed to have camptodactyly and hereditary sensory and autonomic neuropathy combination with a probably autosomal dominant trait.

Discussion

Herein, we report 3 sisters with camptodactyly associated with hereditary sensory and autonomic neuropathy.

Type 1 is the most common type of HSAN and is transmitted autosomal dominantly. It is characterized by sensory deficit in the distal part of the lower extremities, chronic ulcers of the feet and sometimes progressive destruction of the bones beneath the ulcers. Sweating abnormalities may occur but other autonomic features are not frequent in this type. Symptoms appear in late childhood or in early adolescence, mostly after the second decade of life with trophic ulcers as a result of decrease in the pain sensation. Some patients may have accompanying nerve deafness and atrophy of the peroneal muscles. Histopathologically, a marked reduction in the number of unmyelinated fibers may be seen. The motor nerve conduction velocities are generally normal, but the sensory nerve action potentials are usually absent. The Aα, Aδ and C axons are mostly affected. The genetic loci is on 9q22 and the gene that is defined as SPTLC-1. On the other hand, HSAN type2-5 have autosomal recessive trait and the neurologic findings are congenital (5).

HSAN type-1 was the most probable neurologic diagnosis for this family. The patients reported that their weakness begun after 40 years of age and because all had polyneuropathy, it was more probable that the syndrome is autosomal dominantly inherited. But the coexistence of HSAN type-1 and camptodactyly was not reported before. Camptodactyly may be a part of some autosomal dominant diseases, such as Marfan's syndrome, cranio-carpo-tarsal dystrophy and oculo-dento-digital dysplasia. It may also coexist with anomalies such as high arched palate, scapula anomalies, scoliosis, ptosis, hemi-hypertrophy and taurinuria (3,4). None of these anomalies were present in our family.

CMT, with other words Charcot-Marie-Tooth neuropathy, or peroneal muscular atrophy, is an inherited disorder of nerves that takes different forms. It is characterized by loss of muscle tissue and touch sensation, usually in the feet and legs but also in the hands and arms at advanced stages. This disease is one of the most common inherited neurological disorders (36/100.000) (8).

Although there are many different genetic causes of CMT, all types tend to have remarkably similar symptoms; a high arched foot, structural foot deformities, (high-arched foot and hammertoes), progressive muscle wasting leading to problems with walking, running, and balance, ankle weakness and sprains, foot drop, "steppage" gait, progressive atrophy of the thenar muscles, loss of the opposable pinch, tingling and burning sensations in the hands and feet, severe neuropathic pain, diminished sense of touch, dry skin and hair loss, deafness, "stocking-glove" pattern of atrophy, loss of insulating muscle mass, cold hands and feet, swelling (edema) of the feet and ankles, loss of deep-tendon reflexes, weakness of the respiratory muscles, scoliosis, gait abnormalities, and hip dysplasia. One of the major characteristics of CMT is that the clinical findings may vary a lot also in the same family (8,9). The severity of the neurological symptoms may be very mild or very severe. Therefore, this family may also fit the neurological features of CMT but than camptodactyly is not covered by CMT.

In conclusion, we defined a family with polyneuropathy and camptodactyly which may fit HSAN type-1 or a mild form of CMT for the neurologic symptoms, but to our knowledge, this is the first family in the literature with the coexistence of camptodactyly and polyneuropathy.

Conflict of Interest:

Authors reported no conflicts of interest.

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