Acquired adermatoglyphia associated with sporadic inclusion body myositis

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Abstract

A 70-year-old male patient with inclusion body myositis presented to dermatology clinic for evaluation of loss of fingerprints. Eight years previously, he had fingerprints taken for a government permit. Inclusion body myositis (IBM) was diagnosed four years later after the patient developed muscle weakness in his left lower extremity and right upper extremity and was confirmed by muscle biopsy. The patient also reported loss of skin lines that gave his fingers a shiny appearance, most prominently on the left hand, in which he had weakness associated with inclusion body myositis. He was unable to renew the government permit due to an inability for the machine to read his fingerprints. Upon exam, the fingers of his hands, most prominently the left 3rd and 4th digits, had a smooth texture and glossy appearance circumferentially, associated with diminution of skin lines and palmar dermatoglyphs. Workup for connective tissue and other autoimmune diseases was negative. This case suggests asymmetric adermatoglyphia may distinguish a subset of patients with IBM.

Key Words: adermatoglyphia, loss of fingerprints, inclusion body myositis, dermatoglyphs

Introduction

Dermatoglyphs are volar skin lines that form complex and individually unique patterns. In contrast to other skin lines, which develop throughout life in association with muscle use and age-related loss of elasticity, fingerprint patterns are established in gestation. Detailed developmental studies have demonstrated that ectodermally derived epidermis forms dermatoglyphs through patterns of epithelial budding dependent on expression of EDAR and FGF20.[1,2] Unlike the hair placode, which requires the same developmental signals in the epidermis, WNT-dependent recruitment of mesenchymal cells does not occur. Subsequently, waves of WNT-driven proliferation of basilar epidermis originating at the apex of distal phalanges leads to the regularly spaced suprabasilar thickening that forms the primary ridges of fingerprints. Although no new primary ridges arise after 17 weeks gestation, secondary ridges arise between primary ridges and the ducts of sweat glands form pores on the surface of ridges.[2,3] A recent genome-wide association study uncovered variants in limb development genes associated with fingerprint type, likely because fingerprint patterns are correlated with hand and finger proportions.[4] The integrity of dermatoglyphs can be affected by acquired conditions such as dyshidrotic eczema, contact dermatitis, scabies, herpetic whitlow, trauma, micro-abrasions, psoriasis, or Steven Johnson Syndrome.[5]

Although rare, dermatoglyphs can also be affected by congenital conditions such as an inherited absence of epidermal ridges and ectodermal dysplasias.[6,7] Isolated autosomal dominant adermatoglyphia has been described in very few families and is also known as the "immigration delay disease" due to patients' difficulties obtaining government documents. A key finding common in these families is an irregular number of sweat gland openings.[6] Ectodermal dysplasias include a range of syndromes with multiple abnormalities of ectodermal structures including hair, nail, teeth, and sweat glands. Basan syndrome and autosomal dominant adermatoglyphia are ectodermal dysplasias caused by mutations in the skin specific isoform of the SMARCAD1 gene. These two syndromes are rare and have been grouped together into SMARCAD Syndrome. This acronym stands for SMARCAD1-associated congenital facial Milia, Adermatoglyphia, Reduced sweating, Contractures, Acral Bullae, and Dystrophy of nails.[7]

Inclusion body myositis (IBM) is the most common myopathy in patients aged 50 years or older.[8,9] IBM classically presents with progressive and asymmetric weakness. Finger flexors and quadricep muscles are predominantly affected.[10] IBM can be classified as either sporadic or hereditary, with the sporadic form being more common. Sporadic IBM has both inflammatory and degenerative features leading to ongoing debate over the primary driver of the pathogenesis.[9] Endomysial and perivascular immune cell infiltration, and circulating autoantibodies support involvement of the immune system, a lack of responsiveness to immunosuppression and immunomodulatory treatments supports degeneration rather than inflammation, and induction of protein aggregates by inflammatory cytokines supports inflammation rather than degeneration.[9,10,12] While other inflammatory myopathies, such as idiopathic inflammatory myopathy and dermatomyositis, have characteristic skin eruptions, IBM is not known to involve the skin.

Case Presentation

A 70-year-old male with a past medical history of inclusion body myositis, coronary artery disease, type II diabetes mellitus, hypertension, gastroesophageal reflux disease, obstructive sleep apnea, and a 60 total pack year smoking history presented to dermatology clinic for evaluation of loss of fingerprints. Although he had previously normal fingerprints (Fig. 1), the patient had recently been unable to renew a permit due to inability to verify identity because the fingerprinting machine could not detect his fingerprints. The patient was aware of the loss of skin lines on his hands, specifically his fingers because his fingers had developed a shiny appearance. The changes were most apparent on the right second, third, and fourth fingers, where his muscle weakness was most severe. Consequently, he attributed these changes to his diagnosis of inclusion body myositis.

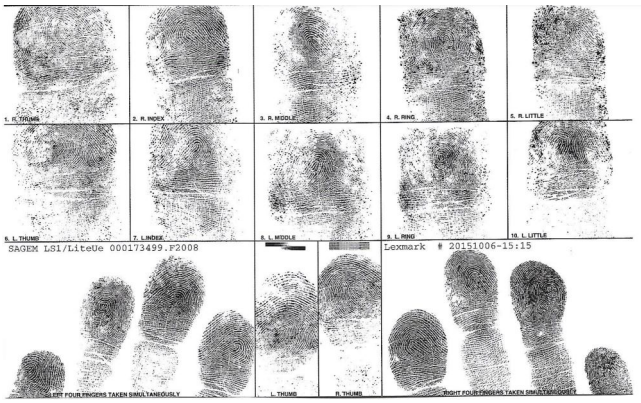


Figure 1. Fingerprints. Patient's normal fingerprints taken three years prior to diagnosis.

At the age of 65 years, the patient presented to his primary care physician with balance difficulty, some weakness of the upper extremities, and occasional trouble swallowing. The patient was referred to neurology, where he was noted to have atrophy of his right arm and forearm with decreased finger flexion on the right side 4/5; decreased dorsiflexion and eversion of the right foot 4+/5; and decreased dorsiflexion, eversion, and inversion of the left foot 4/5. Normal muscle tone and bulk was noted in the left upper extremity and bilateral lower extremities. A modified barium swallow study showed decreased anterior hyoid excursion and decreased laryngeal closure, but swallow was within functional limits. EMG and needle study was performed on the right upper and lower extremities. Myopathic findings were found in the left psoas and right flexor digitorum profundus, with some active denervation potentials seen in the right flexor digitorum profundus, right flexor pollicis longus, right xtensor hallucis longus, right flexor digitorum longus, and left tibialis anterior. MRI of the cervical and lumbosacral spine showed degenerative disc disease but not severe enough to cause his degree of weakness. MRI of the pelvis and left femur showed patchy abnormal signal and enhancement within the rectus femoris, vastus lateralis, and tensor fascia lata muscles. This pattern was most consistent with a myopathy. Based on the asymmetric findings, IBM was suspected. A muscle biopsy of the right rectus femoris muscle using local anesthetic showed endomysial inflammation and rimmed vacuoles, suggestive of IBM. The diagnosis of IBM was confirmed with NT5CIA antibody. A creatine supplement and over the counter taurorsodeoxycholic acid (TUDCA) were started, as well as physical, occupational, and speech therapy for long-term support.

Subsequently, weakness and atrophy in the right upper extremity progressed to the patient being unable to completely close his fist. The patient started having shoulder and hip dislocations due to muscle weakness and first noticed the tips of his fingers were becoming smooth and glossy. He experienced no related pain, sensory issues, redness, discoloration, or Raynaud-like phenomenon. The smoothness and loss of skin lines on his hands increased, and he was referred to the dermatology clinic for evaluation.

On exam, skin atrophy with diminution of flexion creases and dermatoglyphs was noted, with the most profound changes in the distribution of the right flexor digitorum profundus (Fig. 2). Dermoscopy of affected fingers revealed effacement of dermatoglyphs but no other specific features. ANA profile and serologies for autoimmune conditions including autoimmune myositis were ordered. ANA titer was positive to 1:160 with speckled pattern, which was concluded to be nonspecific. Antibodies against SSB, RNP, Sm, SSA Ro52, SSA Ro60, Scl-70, Jo 1, and dsDNA were negative. Additional testing for a panel of antibodies against PL-7, PL-12, EJ, OJ, SRP, Mo-2, TIF-1-gamma, MDA-5, NXP-2, Ku, Scl-100, U1 RNP, U2 RNP, and U3 RNP was also negative.

Eccrine dysfunction is a feature common to genetic adermatoglyphia syndromes. Therefore, eccrine function was assayed by applying iodine to the patient's fingers and pressing the finger pads, once dry, onto plain white paper. This modification of the starch-iodine sweat test technique permits visualization of eccrine secretion to the resolution of individual glands.[13] Results demonstrated fewer secreting eccrine glands from the finger pads, most noticeably on the right hand where the patient's weakness and fingerprint effacement are also the greatest (Figs. 3A-3B).

Discussion

Evidence of muscle involvement in the development or maintenance of dermatoglyphs has not been reported. Congenital absence of fingerprints is a feature of Naegeli-Franceschetti-Jadassohn syndrome and dermatopathia pigmentosa reticularis, which are associated with mutations in keratin 14 and support fingerprints as primarily epithelial structures.[14] A loss of fingerprints from pathological skin thickening is a feature of scleroderma, an autoimmune condition characterized by fibrotic infiltration of the

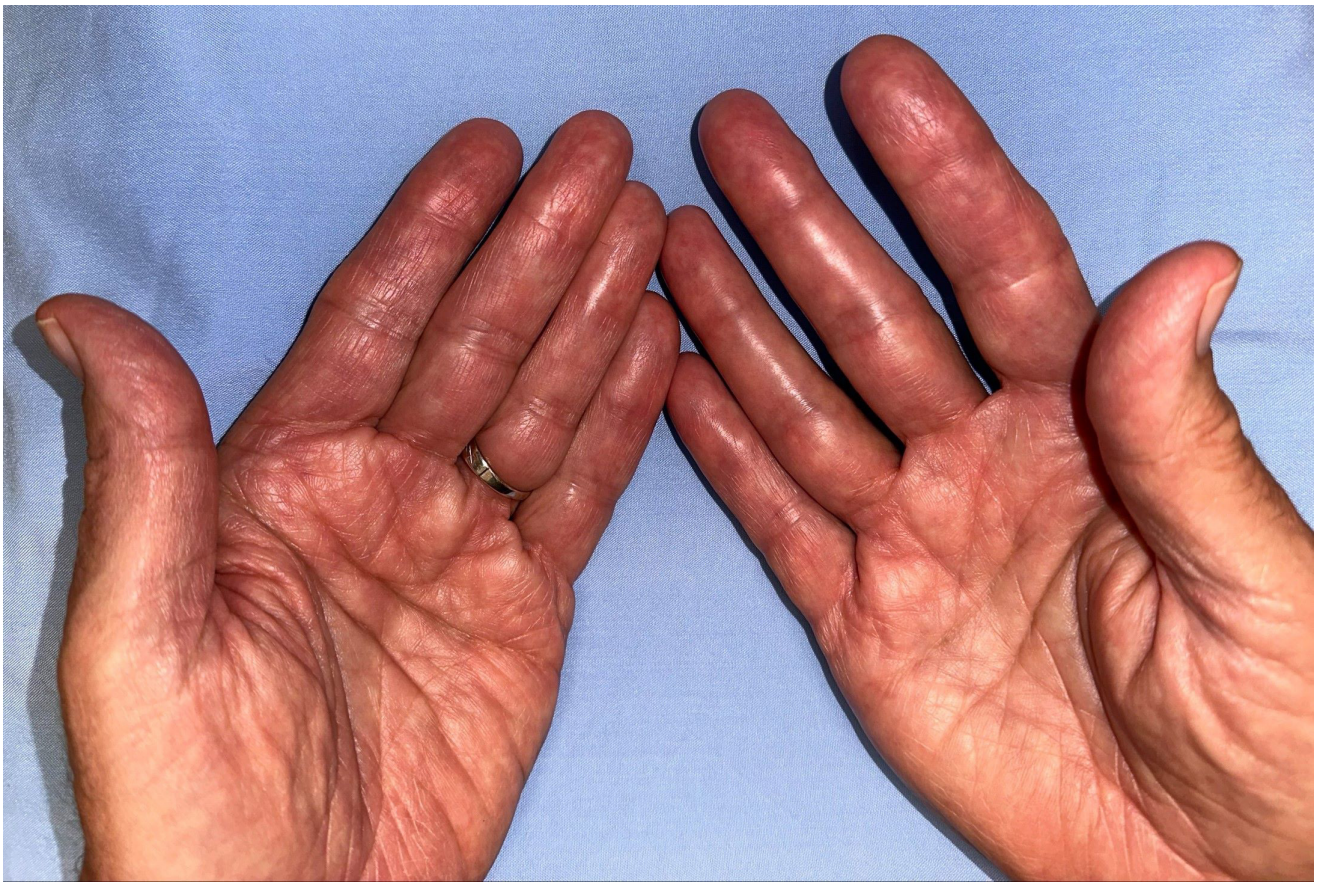


Figure 2. Bilateral palmar hands. Smooth texture and glossy appearance of the fingers of bilateral hands, most prominently the left 3rd and 4th digits. There is absence of skin lines and diminution of palmar dermatoglyphs.

dermis and other organs.[15,16] Overlap between inclusion body myositis and scleroderma has been rarely reported.[17,18] However, fibrosis is not the underlying mechanism for the loss of fingerprints in our patient, whose skin was thin and atrophic rather than fibrotic, and there were no other symptoms nor serological evidence of scleroderma. Similarly, there were no symptoms of dermatitis, accidental trauma, burns, or infection. The patient's history did not support a drug-related cause or other dermatologic condition known to impact fingerprints.

Diminished skin wrinkling of the dorsal fingers has been reported in a series of three patients with IBM who had a loss of wrinkling of the dorsal distal interphalangeal joints in association with flexor weakness of these fingers.[19] Fingerprints were not discussed. In the study of fingerprint analysis, these flexion creases are called white lines and are known to become exaggerated with age, to the point of obscuring dermatoglyphs in some individuals.[20] The authors of the case series suggest flexion creases are maintained by regular skeletal muscle contractions, as in facial rhytides, which can be temporarily relieved by botulinum toxin chemo-denervation of facial musculature.[19] However, chemo-denervation of the palms and soles is not reported to efface dermatoglyphs in patients treated with botulinum toxin for palmar hyperhidrosis. Dermatoglyph effacement is also not described in stroke or other degenerative processes of the central or peripheral nervous system. Therefore, asymmetric absence of fingerprints may be a cardinal feature of some patients with IBM. The associated eccrine dysfunction also matches the patient's distribution of IBM symptoms. Whether this a cause or consequence of acquired adermatoglyphia is uncertain.

Acknowledgment

We are grateful to the patient for his perspicacity, curiosity and patience during this study.

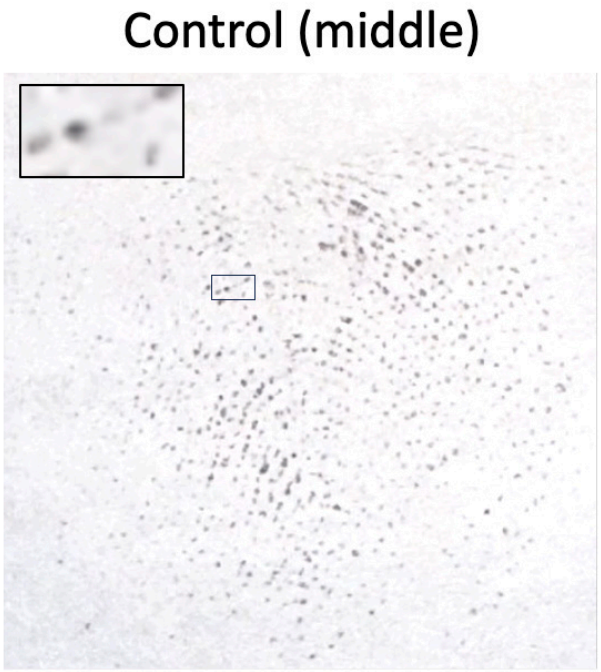
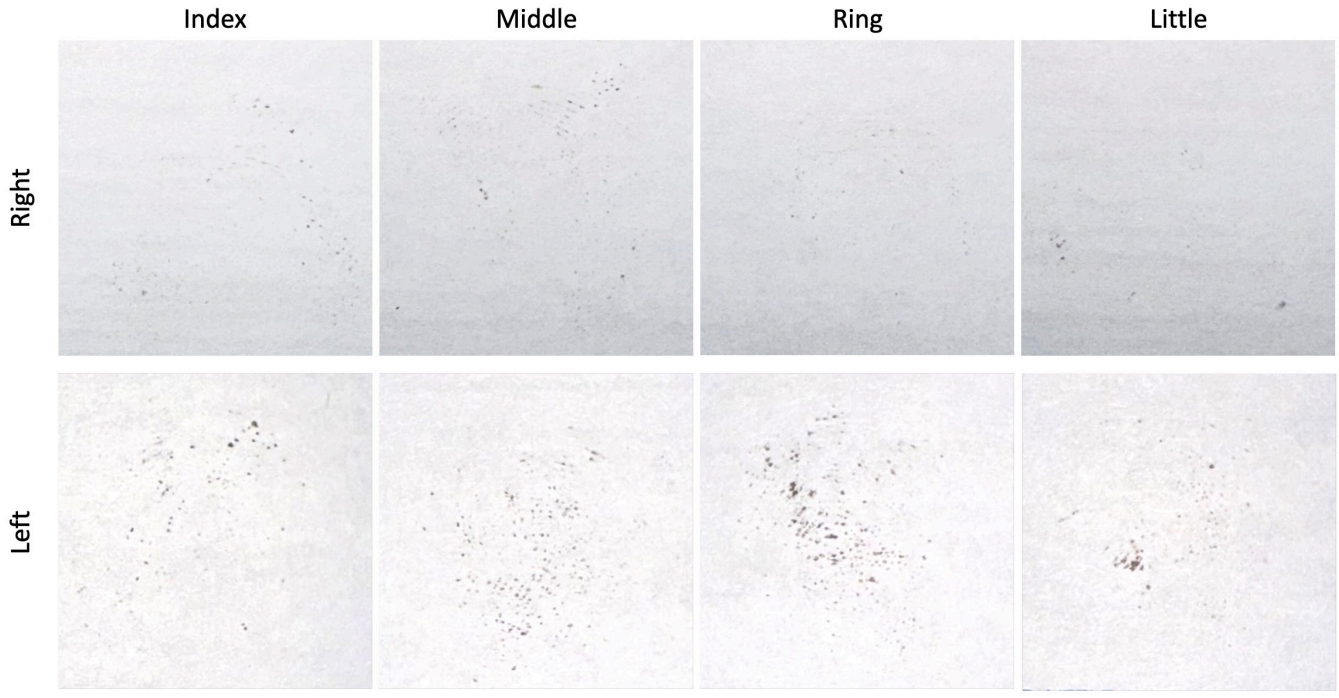


Figure 3A (left). Control. Third finger pad of a control subject. Each dot (magnified in inset) represents the secretory output of an individual eccrine gland.

Figure 3B (below). Patient. Patient finger pads two through four from the right and left hand.



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