Secondary Pseudoainhum in a Patient With Turner Syndrome

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Secondary pseudoainhum is an autoamputation that develops in individuals aged approximately 20 to 50 years and is caused by diseases such as keratodermas, trauma, or congenital factors. The authors report a novel case of secondary pseudoainhum in a patient with Turner syndrome (45,X) who presented with bandlike constrictions in the toes bilaterally. To the authors’ knowledge, secondary pseudoainhum has not been reported to be associated with Turner syndrome. However, physicians should be aware of this potentially deforming disease in patients with Turner syndrome.

Pseudoainhum comprises rare manifestations of bandlike constrictions around the trunk, limbs, or digits, and it may result in autoamputation of the affected part. While ainhum is limited to the toes and is most often found in patients of African descent, pseudoainhum strikes a population defined by broader demographic characteristics.1

Known causes of pseudoainhum include keratodermas, such as leprosy, psoriasis, Vohwinkel syndrome, Mal de Meleda disease, Papillon-Lefèvre syndrome, Clouston syndrome, and congenital ainhum, as well as trauma.2,3 Usually these patients are between the ages of 20 and 50 years at presentation and the lesions arise on the digits of the hands or feet.1 Few reports exist on the best treatment method. If left untreated, the affected digit may autoamputate in 5 to 10 years.1

We present the case of a 44-year-old woman with Turner syndrome and secondary pseudoainhum discovered incidentally during a routine examination.

Report of Case

A 44-year-old white woman with Turner syndrome (45,X) presented for a routine rheumatologic examination for episodic oligoarthritis in her right ankle. A complete medical history and physical examination revealed bilateral lower extremity toe lesions. Her left second and third, and her right second, third, and fourth toes demonstrated linear bandlike constrictions between the interphalangeal joints (Figure) consistent with grade I pseudoainhum. The toes were warm, non-tender, and had good capillary refill. There was no clinical evidence to suggest peripheral vascular disease. The patient could not recall how long the painless, asymptomatic lesions had been present. She reported no trauma to the toes or feet.

Notably, the patient did not have a family history of constricting bands, ainhum, or pseudoainhum. She had diffuse xerotic eczema. Medications included daily Protandim, multivitamin, 1000 mg of L-arginine, and 100 mg of celecoxib, as well as approximately 500 mg of acetaminophen per week. She did not smoke cigarettes. Pertinent examination findings included episodic oligoarthritis without any objective evidence of a systemic inflammatory disorder or arthropathy and right first metacarpophalangeal subluxation and short digits. No synovitis, clubbing, nail pitting, or onycholysis were observed, and she was able to clench her fists.

Complete blood cell count, complete metabolic profile, rheumatoid factor, anti–cyclic citrullinated peptide antibodies, antinuclear antibody, and uric acid laboratory test results were within normal limits. Because the incidental toe findings were asymptomatic, an approach to resolution was not considered. However, her chief complaint of oligoarthritis in the right ankle was addressed and recommendations made. The differential diagnosis included systemic inflammatory arthropathy, reactive arthritis, ostearthritis, and crystal disease. She was advised to continue taking celecoxib and was prescribed orthopedic shoes. Follow-up care was recommended on an as-needed basis.
Discussion

The terminology for ainhum has been varied in the past, but efforts have been made to categorize the different types. For example, primary pseudoainhum is a congenital autoamputation that is not restricted to the digits, and secondary pseudoainhum has been defined as autoamputation that develops in adults aged 20 to 50 years and is caused by keratodermas or trauma or can be congenital.\(^1\)

Often, ainhum is termed *dactylolysis spontanea* and is limited to the fifth toe. Most common among persons of African descent, ainhum is likely triggered by trauma, which induces the formation of a constricting band that grossly limits circulation, resulting in autoamputation. Histologically, ainhum resembles vegetative foreign body granulomas.\(^5\) We suspect that pseudoainhum manifests the same granulomatous histologic findings.

Our patient had grade I pseudoainhum, which indicates the appearance of a groove of constricting soft tissue but without symptoms. Grade II is ulceration of the floor of the groove; grade III, bony erosion, and grade IV, spontaneous amputation.\(^1\)

To our knowledge, pseudoainhum has not been reported in patients with Turner syndrome. A common genetic disorder in women (1 in 2500),\(^6\) Turner syndrome is caused by X-chromosome monosomy, mosaicism for X-chromosome monosomy, or structurally abnormal second X chromosomes, and it affects many organ systems.\(^7\) Patients with Turner syndrome classically develop a triad of short stature, impaired sexual development, and infertility.\(^8\) They also manifest osteoporosis (linked to decreased estrogen), cardiovascular disease (usually seen as left-sided heart anomalies), sensorineural hearing loss, and a variety of autoimmune disorders such as hypothyroidism, early-onset insulin resistance, and inflammatory bowel disease.

Some dermatologic disorders have been linked to Turner syndrome. Most dermatologic symptoms are sequelae of autoimmunity, including vitiligo, alopecia areata, and psoriatic arthritis.\(^9\) For unknown reasons, patients with Turner syndrome have a greater number of benign melanocytic nevi, with a prevalence of up to 70%.\(^11\) An increased number of melanocytic nevi is the strongest risk factor for melanoma.\(^12\) Other dermatologic manifestations include premature aging of facial skin, decreased prevalence of acne vulgaris, and an increased number of café au lait macules.\(^13,14\)

A few nonautoimmune manifestations exist. Patients with Turner syndrome are exceptionally prone to lymphedema and pterygium coli (webbing of the neck), theorized to stem from lymphatic hypoplasia in the subcutis resulting from developmental failure of connections between the venous and lymphatic system.\(^10\)

Perhaps the link between Turner syndrome and secondary pseudoainhum is the increased likelihood of keloids and hypertrophic scarring.\(^15\) Trauma to the area could lead to a hypertrophic scar and thus the banding and constricting phenomena seen in secondary pseudoainhum. It is possible that the patient described in the current report incurred an unknown trauma to the toes.

Another hypothetical link between Turner syndrome and secondary pseudoainhum lies in the intrinsic autoim-
Community seen in Turner syndrome. One study linked autoimmunity to the formation of keloids and hypertrophic scars, stating that an increased number of immunoglobulins in the affected tissue suggest an abnormal immune reaction. It is possible that mechanical stretch and tension in the toes overstimulated collagen production—a result of abnormal epithelial-mesenchymal interactions.

The best treatment for patients with pseudoainhum has been debated. There are a few common therapeutic techniques, including skin grafting and Z-plasty. Most commonly, patients undergo a procedure to release the constricting band using a Z-plasty technique. This surgical technique is associated with relief of pseudoainhum after 18 months.

Although Z-plasty remains an option for these patients, managing secondary pseudoainhum as a keloid could yield promising results. Treatment methods range from classic (e.g., intraslesional corticosteroids) to novel techniques, including topical imiquimod, topical retinoids, and intraslesional fluorouracil.

Conclusion
Secondary pseudoainhum is a rare disease previously unassociated with Turner syndrome. Physicians should be aware of this potentially deforming disease in patients with Turner syndrome or the conditions listed previously. The nature, frequency, and strength of this newly reported association needs to be further explored.

References