Congenital Pseudoainhum

In Joon Lee, M.D., Un Sun Choi, M.D., Soo Il Chun, M.D.

Department of Dermatology, Yonsei University College of Medicine
Seoul, Korea

Annular constrictions involving the lower end of both arms and legs were seen in a 10-month-old female infant. The annular constrictions of the extremities were present at birth and the general development of the child was below average. No radiologic abnormalities were found. Histopathologically, finger-like projections of dermal collagen bundles into the subcutaneous fat were seen. (Ann Dermatol 4 : (1) 57–61, 1992)

Key Words : Congenital pseudoainhum

Constricting annular band had been previously described under different headings by many authors. It is a rare form of abnormality involving mainly extremity with varying degree of involvement from shallow groove to amputation. Variable classifications of this phenomenon were offered by many authors. Wells and Robinson (1952) classified the disease into four general classes: 1. ainhum; 2. congenital band; 3. ainhum-like band associated with other disease; 4. band secondary to trauma. Neumann (1953) used the term pseudoainhum to distinguish the 3 latter classifications from ainhum. In 1972, Raque et al made a comprehensive classification as follows: 1. ainhum, 2. pseudoainhum: a. congenital constricting band, b. constricting band associated or secondary to specific disease, c. secondary to trauma.

The ainhum is characterized by a painful annular constriction of the fifth toe occurring predominantly in the adult Negro with eventual spontaneous amputation. In congenital pseudoainhum, constricting bands are present at birth which may vary from shallow depression to deep constriction extending down to the bone, and this deep variety may be associated with gross deformity or even amputation.

This report, which is the first one being reported in Korean literature, describes a case of congenital pseudoainhum in a 10-month-old female infant with annular constrictions around the lower end of upper and lower extremities of both side.

REPORT OF A CASE

A 10-month-old female infant patient was first seen in our clinic for delayed growth, weakness of lower extremities, and annular constricting bands at upper and lower extremities of both side.

The patient’s development was retarded compared to the same age group. The weight was in 10-25 percentile range and head circumference was in 5-10 percentile range. The patient was not able to sit firmly and was only able to crawl on abdomen backward. In addition, weakness of lower extremities was present. The
annular constrictions were seen just above the wrist of both hands and the ankle of both feet. The patient was a product of normal pregnancy of 36 weeks and uncomplicated normal spontaneous vaginal delivery with birth weight of 2.4kg. The mother was 22 years old and reported no medication, infection, or irradiation history prior or during pregnancy. She has had no particular occupation and reported no known family history of congenital disorders. The father was 40-year-old man who stated his occupation as a business man. He also denied any medication, infection, or irradiation history prior to conception, and also reported no known family history of congenital disorders.

Physical examination revealed constrictions just above both ankles and wrists. Infolded grooves involving medial side of both forearm were seen about 3cm above both wrists (Fig. 1) and also infolded grooves nearly encircling both lower leg were seen about 4cm above both ankles (Fig. 2). The constrictions were freely movable and were not fixed to the underlying bone. There were no associated anomalies of hand, finger, foot, toe, or any other part of the body. Radiologic survey of skull AP & lateral, chest PA, and long bone series revealed no abnormal findings except shadow of the grooves (Fig. 3). Skin biopsy was performed from the annular constriction of the leg and it showed dermal collagen fiber bundles extending into the subcutaneous fat as finger-like projections (Fig. 4, 5). Special stains such as Masson's trichrome and
Victoria Blue was done which showed relatively proliferated and thickened elastic bundles in the dermis as well as in the projection bundles. Collagen bundles were regarded as normal in amount and in size.

**DISCUSSION**

The word ainhum is derived from via Portuguese from ayun, or eyun, a word in Yoruba (language from eastern Guinea Coast, Africa) meaning “to saw”. True ainhum, which was first fully described in 1867 by Silva Lima, is a relatively common disease among Negroes in Africa with estimated reported incidence of 2.48 per 1,000 males and 1.08 per 1,000 females in Nigeria. It is a disease of unknown etiology affecting almost exclusively the fifth toe of the adult Negro. It usually start with painful constriction around the first interphalangeal joint with eventual distal degeneration and spontaneous amputation within three to ten years.

Raque et al² divided pseudoainhum into three types: 1. congenital form, familial and nonfamilial: 2. form associated with or secondary to specific disease: 3. form secondary to trauma. While pseudoainhum of the toe developing secondary to trauma or associated with other diseases may be confused with ainhum, congenital pseudoainhum present at birth is readily distinguished from other forms.

In congenital pseudoainhum the distal parts of the extremity such as finger, toe, forearm, and leg are the most affected sites. The proximal part of the affected limb is normal but distal part of the constriction band can be associated with anomalies such as syndactyly, microdactyly, and club foot. The defects usually range in severity from superficial annular grooves of the skin to, rarely, amputation of the part of affected limb. It may be deep enough to affect the limb in utero resulting in baby with amputated limb and terminal scar formation at the end of the stump. Pillay and Hesketh³ illustrated range of severity of the cases in their report. One case was deep annular constriction resulting in gangrenous change requiring amputation after birth, second case was deep grooves causing gross swelling at

---

**Fig. 4.** Collagen bundles projecting into the subcutaneous tissue in a fingerlike projection (H & E, ×40).

**Fig. 5.** Higher magnification of the Fig 4 (H & E, ×100).
birth which was relieved by surgery, and third case was chronic elephantiasis of the foot and ankle developed from constriction band. In our case, the degree of constriction was superficial enough to be freely movable from underlying structure with no accompanying secondary changes. Also, no anomaly of distal part was present.

Peterka and Karon\textsuperscript{5} in 1964 reported histopathological findings of congenital pseudoainhum for the first time. The findings are summarized as follows. The constriction band showed the hyperkeratotic, slightly acanthotic epidermis with fibrous tissue staining as dense collagen fibers resembling scar tissue in the dermis. The deeper portion of dermis was formed into broad finger-like projections of collagenous and coarse elastic bundles penetrating deeply into the subcutaneous fat. This findings suggest a form of collagen dysplasia. Our biopsy showed mainly collagen bundles extending into the subcutaneous tissues in a finger-like projection accompanied by relative proliferations of elastic fibers.

No satisfactory explanations concerning etiology or pathogenesis of constricting band exists. However, many speculations are abound. Streeter\textsuperscript{6} in 1930 discarded the intrauterine amniotic adhesion theory which stated that macerated sheets of hyalinized fibrous tissue, which were remains of localized defective tissue, causing mechanical obstruction which in turn restricts normal tissue development. He speculated that a focal degenerative process in the extremity itself was the primary cause and was hereditarily transmitted. Kohler\textsuperscript{7} and Streeter\textsuperscript{6} reported familial cases of pseudoainhum. Animal experiment done by Wooley and Cole\textsuperscript{8} reported development of spontaneous grooves surrounding the tails of Norway rats and subsequent loss at the point of constriction. This finding was found in certain strains with a change in diet. Bagg\textsuperscript{9} exposed roentgen rays to fetal mice and noted defects which included congenital amputations. Duraiswami\textsuperscript{10} in 1952 injected insulin and other substances into yolk sac of developing chick embryo which produced congenital defects. Injection of riboflavin and niacinamide, however, prevented these defects. Pillay and Hesketh\textsuperscript{1} in their reports stated that the annular constricting defects in Singapore was 1.15 per 10,000 in Malays compared to 0.0005 per 10,000 in the Chinese. The Malays are poorer group compared to Chinese, and more prone to be exposed to harsher environments and malnutritions. These clinical and experimental findings suggest that both genetic predisposition and enviromental factors are involved in the pathogenesis. No known facts concerning any genetic or environmental predispositions are suggested in our case. One can only impute imperfect development to factors residing in the genes being influenced by unknown factors affecting this particular generation.

There are two views concerning treatment of this disease. Neumann\textsuperscript{11} and Peterka and Karon\textsuperscript{5} were of opinion that treatment was usually not indicated because the defects usually do not progress and the annular bands grow with the underlying structures. However, multiple incisions can be made to lessen the depth of the band. Wells and Robinson\textsuperscript{12} reported success with such methods. In the orthopedic community, recommendation of staged Z-plasty as the treatment of choice is the normally accepted practice\textsuperscript{1,4,13,14,16,17}. Its purpose is to improve the cosmetic appearance and to prevent eventual development to obstruction of lymphatics or vessels. There are also report of successful therapy with oral etretinate (1mg/kg per day) in aborting impending amputation in Vohwinkel’s syndrome\textsuperscript{15}.

In conclusion, the authors report a case of nonfamilial congenital pseudoainhum in a 10-month-old female infant whose lesion was presented both in upper and lower extremities. In our opinion, the child is to be observed further
and perform multiple stage Z-plasty at later times if there is any sign of strangulations.

REFERENCES


