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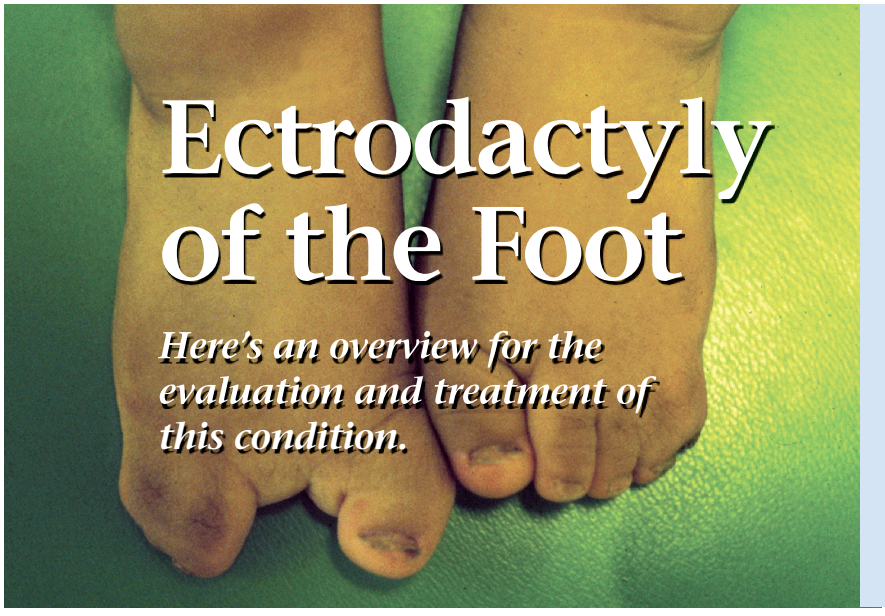

Ectrodactyly of the Foot

Here's an overview for the evaluation and treatment of this condition.

Goals and Objectives

At the completion of this article, the physician will be able to:

1) Define ectrodactyly of the foot.

2) List the various ectrodactyly associated syndromes and characteristics of each.

3) Discuss the common presenting clinical findings of ectrodactyly.

4) Understand the basic human embryology and genetics associated with the ectrodactyly defect.

5) Classify a pedal ectrodactyly deformity based on roentgenographic findings

6) Develop a treatment plan for a patient with pedal ectrodactyly.

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*Following this article, an answer sheet and full set of instructions are provided (p. 170).—**Editor***

By Mark A Caselli, DPM

Ectrodactyly, by strict definition, is the congenital absence of all or any part of a digit. The term ectrodactyly originates from the Greek ektroma (abortion) and daktylos (finger). The term is also applied to cleft foot, lobster foot, and crab-claw foot. In ectrodactyly, one or more toes and parts of their metatarsals are usually absent. The first and fifth rays are usually present (Figure 1). If a metatarsal is partially or completely absent, its respective toe is always absent. The incidence of cleft foot is 1:90,000 live births. Ectrodactyly was first documented in 1770 among a tribe of Guiana Indians.

Von Walther described the crab-claw deformity in 1829, and Cruveilhier first used the term "lobster-claw" in 1842. Pedal ectrodactyly may exist as a unilateral or bilateral deformity in many combinations with other deformities (Figure 2). It may be present as an isolated deformity or simultaneously with similar defects in the hand. It most frequently presents as part of a defect syndrome, including defects of the eye, palate, audition, and abnormalities of the kidneys.

Associated Syndromes

Several syndromes have been associated with ectrodactyly. One of the most common is the EEC syndrome consisting of ectodermal dysplasia, ectrodactyly, and cleft lip and palate. EEC was first described by Eckholdt and Martins in 1804. The clinical manifestations of ectodermal dysplasia include cutaneous alterations (dry, thin, hyperkeratotic skin), trichodysplasia (thin, sparse hair with little pigmentation), onychodysplasia (brittle and dysplastic nails), dental anomalies (small or absent teeth with excessive caries), and dyshidrosis (sweating dysfunction). The cleft lip may or may not be accompanied by a cleft palate.

Defects in other locations may also be associated with the classical EEC triad, such as the eyes, including defects in the nasolacrimal duct, entropion, telecanthus, trichiasis, absent lashes,and meibomian gland disfunction, manifest-

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ing as blephoritis, corneal scarring, recurrent erosions, blepharospasm, dry eye, and photophobia. Genitourinary system defects include hypospadias, hydronephrosis, renal agenesis, and renal duplicity. Both deafness and mental retardation are also associated with EEC syndrome. The clinical expression of the defects associated with EEC is very diverse, ectodermal dysplasia occurring 100%, lachrymal duct alterations 70-96.5%, ectrodactyly 78-88%, cleft lip/palate 58-88%, urinary alterations 15-55%, deafness 8-28%, and mental retardation 1-16%. The incidence of EEC is 1:190,000 live births.

Other Syndromes

Other syndromes associated with ectrodactyly include Carpenter's syndrome (acrocephalopolysyndactyly type II), DeLange syndrome (multiple congenital anomaly/mental retardation (MCA/MR) disorder), Goltz syndrome (focal dermal hypoplasia (FDH) disorder), Jarcho Levine syndrome (spondylothoracic dysplasia (STD) and spondylocostal dysostosis (SCD) disorder), and Miller syndrome (post-axial acrofacial dysostosis). Raas-Rothschild, et al. described a syndrome of ectrodactyly, short stature, mental retardation, sensorineural deafness and abnormal faces.

Limb-reducing defects that can be associated with ectrodactyly include aplasia and hypoplasia of the tibia or fibula (i.e., Volkmann syndrome). This distal extremity reduction may occur concurrently with normal formation of the proximal limb segment. Ectrodactyly has also been found in patients with aplasia cutis congenital and epidermolysis bullosa. Pedal ectrodactyly, without similar upper extremity deficits, occurs in 10% to 50% of cases of trisomy.(18)

Syndromes involving both polydactyly and ectrodactyly have been described with the same digit usually involved (absence or duplication); the thumb in Fanconi anemia, Thalidimide syndrome and Nager acrofacial dysostosis; the fourth digit in congenital hypothalamic hamartoblastoma polydactyly syndrome. Triphalangeal thumbs have been associated with ectrodactyly in a rare autosomal dominant syndrome. The combination of polydactyly and oligodactyly has been observed with both genetic and teratogen induced malformations in experimental animals.

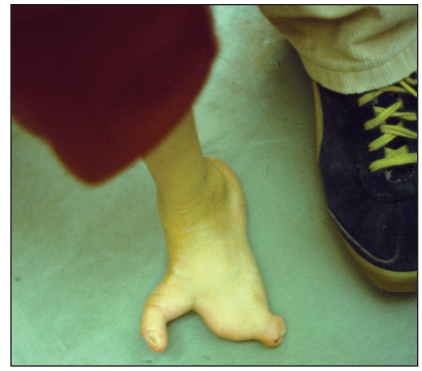

Figure 1: A classic presentation of pedal ectrodactyly.

In Thalidomide embryopathy, the polydactyly always occurs on the hands with a reductional abnormality involving the feet. Autosomal recessive syndromes involving ectrodactyly have been described in a closed island population in Japan and in Middle Eastern cultures where marriage between first cousins is encouraged.

Embryology and Genetics

Ectrodactyly in Africa

In 1770, Jan Jacob Hartnick, a director of the Dutch East India Company, made a reference to a touvinga or two-fingered tribe from Dutch Guyana in South America. They were slaves who exhibited ectrodactyly, brought over from Central Africa. In the 1960's, newspapers published interviews with unimpeachable witnesses who saw the phenomenon in Africa. Gelfland, et al., in 1974 investigated a Wadoma tribesman from the Zambezi valley of Zimbabwe. He was diagnosed as having "lobster claw" deformity of both feet. Two of his cousins were affected and the condition was thought to be autosomal dominant.

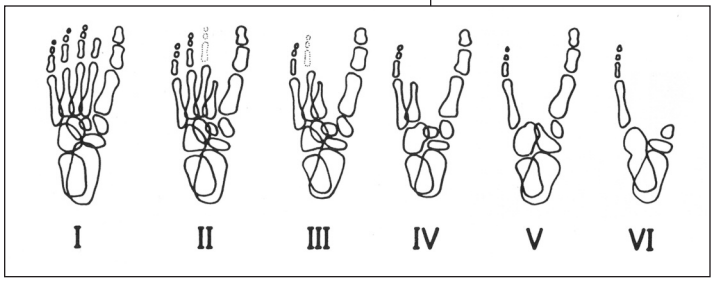

Figure 3: Blauth and Borisch radiological classification of pedal ectrodactyly.

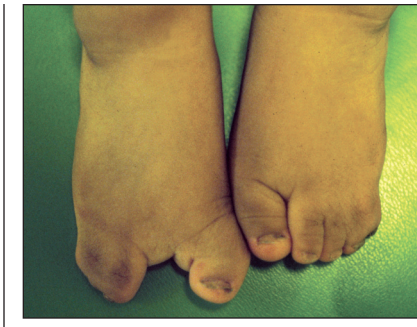

Figure 2: Patient exhibiting ectrodactyly deformity of the right foot and multiple syndactilizations of the left foot.

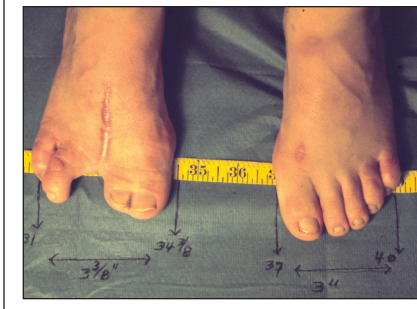

Figure 4: Postsurgical correction of ectrodactyly. Presurgically, the corrected foot was too wide to fit into shoes.

The anomaly was also seen in the Talaunda tribe from eastern Zimbabwe and Botswana. The Talaunda migrated from eastern Zimbabwe and may have been of the same origins as the Wadoma. In 1979, a magazine reported a group of at least 100 "ostrich footed" men. Recent studies have found three families of affected individuals among these tribes and concluded that the previous tales of hundreds of ostrich-footed persons belong in the realm of fantasy.

Embryology

Human limbs develop from interactions between limb ectoderm and mesoderm. An important embryonic somite-somatopleural relationship precedes the emergence of the limb bud during the fourth week of gestation. The lower limb buds develop as lateral thickenings of the somatopleure. At the fifth week, vertebrate limb buds develop a thickened epithelium at their distal tips. This apical ectodermal ridge (AER) functions as a principle inducer of axial limb elongation. The AER is required for progressive specification of mesodermal limb parts and promotion of axial growth through influencing cell division. The ridge also protects subridge mesodermal cells from necrosis during the active period of limb development.

The deep midline cleft, character-

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istic of ectrodactyly, is caused by the absence of one or more rays and toes. The cleft formation begins at the 2nd or 3rd ray and proceeds from the distal to the proximal aspects and transversely from the tibial to the fibular side of the foot. The defects are always greater distally than proximally. There may be associated syndactyly of the remaining digits. A defect in the AER is suspected to be the initiating factor in this pattern of development.

Genetics

Ectodactyly is frequently bilateral and can be autosomal dominant or genetically heterogenous. The gene responsible for the inheritance of this condition has been mapped to the long arm of chromosome 7 at q21.3-q22.1, an area implicated as important in normal human limb development.

Ectrodactyly is an unusual disorder in that it displays several genetic phenomena including variable expressivity, reduced penetrance, and segregation distortion. Variable expressivity is the phenomenon in which expression of a disease may differ considerably among individuals who carry the same gene mutation. One individual may have involvement of both of the hands and feet, while another member of the same family may have only one affected limb. An isolated family report studied six generations and found progressive increase in the severity and extent of involvement in the later generations.

Penetrance is the probability that a gene will have any phenotypic expression at all. Reduced penetrance is the case in which an individual carries the defective gene but fails to express any phenotypic abnormalities. Ectrodactyly of the hand and foot is reported to be approximately 70% penetrant. The third genetic phenomenon is genetic distortion. Alleles, or alternate forms of a gene (all organisms have two alleles for each trait), typically segregate randomly and therefore, there should be no excess of either sex becoming afflicted on transmission of a gene.

However, in the many cases of ectrodactyly studied, there was an obvious excess of afflicted male offspring through paternal transmission. In one study, a Pakistani kindred comprising seven generations and 36 members with the split-hand/split-foot anomaly, the full expression of the trait, monodactylous or split hand and split foot, mainly of the lobster-claw type, was present in 33 males and 3 females. Other females showed a distinctly milder expression of the trait, usually in the form of partial syndactyly, metacarpal and phalangeal hypoplasia and malformation.

The etiology of the principle malformations of EEC has been established as an autosomal dominant alteration in the long arm of chromosome 7 at q21.2-q21.3 with variable penetrance. Four other chromosomes have been associated with this syndrome including Xq26, 10q24, 3q27 (TP63), and 2q31.

Ectrodactyly has also been associated with trisomy 18.

Classification

Blauth and Borisch introduced a classification of six groups of ectrodactyly of the foot based on the roentgenographic number of retained metatarsals (Figure 3).

Grade I: Five completely normal metatarsals. Total or partial aplasia of toes 2 to 5, but usually with the involvement of toes 2 and 3. Occasionally cross-bone present.

Grade II: There are five metatarsals, but they are partially hypoplastic or synostotic with other metatarsals or phalanges. The second or third ray is always affected. At least one toe is absent.

Grade III: There are only four metatarsals. Number 2 or 3 is always missing and the other is hypoplastic.

Grade IV: There are only three metatarsals. Numbers 2 and 3, or 3 and 4 are absent. Toes 2-4 are absent.

Grade V: The "Lobster-claw" foot. Complete absence of second, third, and fourth rays.

Grade VI: Monodactylous cleft foot with only the fifth metatarsal and toe.

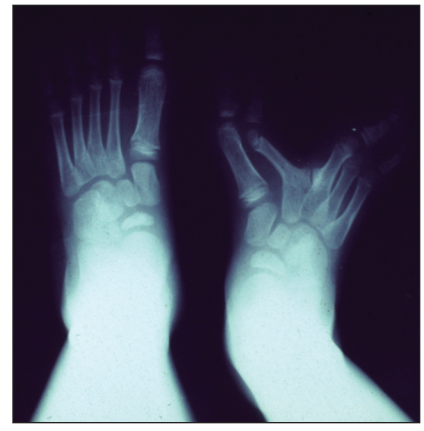

Figure 5: Presurgical roentgenogram of foot in figure 4.

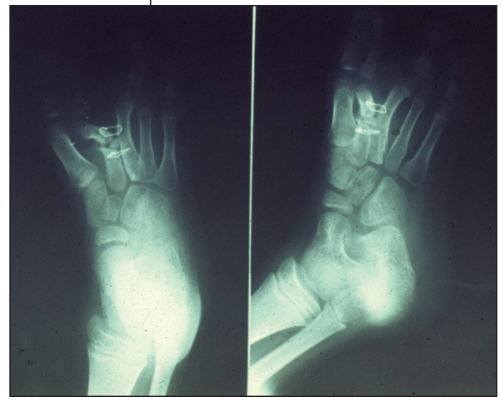

Figure 6: Postsurgical roentgenogram of foot in figure 4. Note the use of metatarsal osteotomies to narrow the foot.

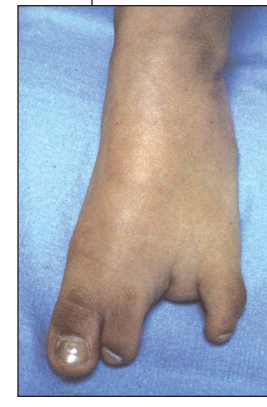

Figure 7: Ectrodactyly deformity of the left foot in an 11-year-old male.

Biomechanics

In an anatomic study of a case of ectrodactyly, congenitally short limb, and club foot, the absence of lumbrical and extensor digitorum brevis muscles was noted. The function of the extensor digitorum brevis during midstance is to stabilize the toes at the metatarsophalangeal joint and resist the weak plantarflexory force of the flexor digitorum longus during midstance. The lumbricals extend the proximal interphalangeal joint and distal interphalangeal joint during propulsion. During midstance, as the flexor digitorum longus contracts to help decelerate the tibia and subtalar joint, the extensor digitorum brevis prevents flexion at the metatarsophalangeal joint while the lumbricales extend the proximal interphalangeal joint and distal interphalangeal joint. The absence of these muscles could lead to clawing of the toes and forefoot instability.

Whenever toes are missing, whether it is from amputation, toe transplant surgery to replace missing fingers, or ectrodactyly, a filler or buttress should be provided. Without buttressing, hallux valgus and subluxations of lesser digits may occur. If the second toe is missing, a bunion may progress rapidly

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because stage 2 in the development of a bunion is bypassed, leading to a stage 3 or 4 bunion deformity.

Approach to Management

The feet of patients with ectrodactlyly are usually functional, and ambulation is not a problem. The inability to fit into normal footwear is the most common problem. Bursitis and painful callosities may develop as a result of the bony deformities and associated abnormal distribution of weight. The more severe forms of ectrodactyly can cause gait problems such as instability during the propulsive phase. Ideal treatment would bring the size of the foot into the normal range, fill the cleft, correct the secondary deformities, and maintain good function.

Surgical criteria for ectrodactyly can include the inability to fit into shoes and the elimination of discomfort. Surgical intervention, if deemed necessary, should be offered to the patient at a very early age to avoid further pathological changes that can occur over time. There is no clearly defined surgical technique, though the surgical options that are described generally agree on resection of the rudimentary elements, osteotomies at different levels, syndactylization, desyndactylization, hammertoe, and hallux valgus surgery (Figures 4, 5, 6).

Biomechanical management, ideally, should consist of a foot orthosis, the elements of which should include: a posterior and central stabilizing component to control the movements of the subtalar joint; an anterior component with a balancing effect to facilitate propulsion, stabilize the front of the foot, and cushion the first and fifth metatarsal heads.

A filler for the cleft to impede further forefoot structural deformation must also be included. A toe buttress manufactured in the office from a silicone putty compound may be used as a temporary spacer, or if deemed appropriate, as sole management for the least severe cases. It should be anchored on a flanged toe buttress running under the existing toes. Latex prostheses may be constructed to plaster casts. Other alternatives include the use of a shoe insert constructed of Plastizote or lamb's wool, or a custom-molded shoe with a spacer.

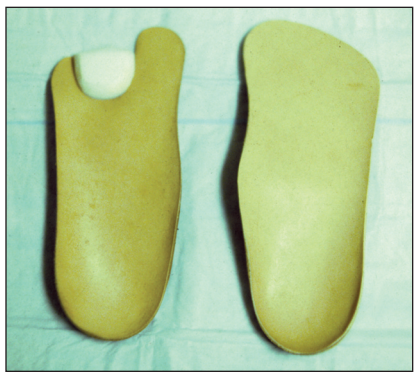

Figure 9: Custom orthoses fabricated for patient with filler to accommodate for ectrodactyly deformity.

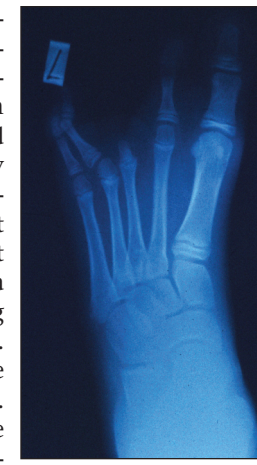

Figure 8: Roengtgenogram of patient's left foot with absent third toe and syndactyly of the fourth and fifth toes.

Case Presentation

An 11-year-old white male presented with a chief complaint of absent third and fourth toes, and a wide fifth toe of his left foot (Figure 7). His right foot was normal. The patient was currently having no problem fitting into conventional shoe gear and was in no discomfort. His mother was concerned about the curling of his most lateral toe, which she claimed had been getting worse over the past year. She was also concerned about possible future problems with the deformity.

The patient's past medical history was unremarkable with no known allergies and no medications. There was no family history of a similar problem. The birth history was uneventful. Vascular and neurological findings were within normal limits.

Roentgenograms revealed the absence of the distal, middle, and most of the proximal phalanx of the third toe, with syndactylization of the fourth and fifth toes of his left foot (Figure 8). The treatment plan was to fabricate a pair of leather laminate orthoses from a neutral impression cast of his feet. An accommodative spacer was fitted on the left orthosis to accommodate for the space created by the deformity, and to act as a buttress for the lateral digit, reducing its tendency for adductovarus curling (Figure 9). The patient was fitted with the orthoses and tolerated them well. ■

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EXAMINATION

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See answer sheet on page 171.

1) Which one of the following terms is not a synonym for pedal ectrodactyly?
- A) Cleft foot
- B) Skew foot
- C) Lobster foot
- D) Crab-claw foot

2) What is the incidence of cleft foot?
- A) 1:500 live births
- B) 1:10,000 live births
- C) 1:90,000 live births
- D) 1:190,000 live births

3) Which one of the following is not part of the EEC syndrome?
- A) Ectodermal dysplasia
- B) Chondromalasia
- C) Ectrodactyly
- D) Cleft lip

4) The clinical expression of ectrodactyly in EEC syndrome presents in _____% of cases.
- A) 8-28
- B) 15-55
- C) 78-88
- D) 100

5) Ectrodactyly is an associated finding in which one of the following trisomy defects?
- A) Trisomy 8
- B) Trisomy 13
- C) Trisomy 18
- D) Trisomy 21

6) Embryologically, what is the function of the apical ectodermal ridge (AER)?
- A) Induce axial limb elongation.
- B) Retard digital differentiation.
- C) Responsible for nail growth.
- D) None of the above.

7) A defect in the AER can result in:
- A) Kidney abnormalities
- B) Cleft foot
- C) Deafness
- D) None of the above

8) The gene that is responsible for the inheritance of ectrodactyly has been mapped to chromosome _____.
- A) 5
- B) 7
- C) 12
- D) 18

9) Genetically, variable expressivity is a phenomenon in which the expression of a disease:
- A) Appears the same in all who have the same gene mutation.
- B) May be radically different in all who have the same gene mutation.
- C) Only occurs in female family members.
- D) Only occurs in male family members.

10) Which one of the following is most correct concerning the inheritance of ectrodactyly?
- A) It tends to skip generations.
- B) There is an excess of afflicted males.
- C) There is an excess of afflicted females.
- D) It becomes less severe in later generations.

11) Which one of the following is the key characteristic of Blauth and Borisch Grade I ectrodactyly?
- A) Five completely normal metatarsals
- B) Partially hypoplastic metatarsals
- C) At least one absent toe
- D) Classic "lobster-claw" foot appearance

12) The main characteristic of Blauth and Borisch Grade III ectrodactyly is:
- A) At least one absent toe.
- B) Only four metatarsals.
- C) Monodactylous cleft foot.
- D) Only three metatarsals.

13) What Blauth and Borisch group represents the classic "lobster-claw" foot?
- A) Grade I
- B) Grade III
- C) Grade V
- D) Grade VI

14) A foot with an ectrodactyly defect was found to have absent extensor digitorum brevis and lumbrical muscles. Which one of the following is most likely to result from the absence of these muscles?
- A) Weak push-off phase of gait
- B) Hallux valgus
- C) Clawing of toes
- D) Equinus gait

15) Which one of the following has been found to be the main problem of pa-

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EXAMINATION

(cont'd)

tients with ectrodactyly?
A) Plantar callosities
B) Inability to fit in shoes
C) Foot pain
D) Inability to ambulate

16) Which one of the following is the most common gait problem in severe forms of ectrodactyly?
A) Instability during propulsion
B) Inversion sprains at heel contact
C) Midstance pronation
D) Drop foot

17) Absence of the second toe promotes which one of the following deformities?
A) Hammertoes
B) Hallux valgus
C) Metatarsus adductus
D) None of the above

18) Which of the following represents ideal treatment for pedal ectrodactyly?
A) Normalize size of foot
B) Fill the cleft
C) Maintain good function
D) All of the above

19) Which of the following is a consideration for surgical intervention in the treatment of pedal ectrodactyly?
A) Inability to fit into shoes
B) Elimination of discomfort
C) Age of patient
D) All of the above

20) Which one of the following orthotic modifications is needed to prevent forefoot structural deformities such as hallux valgus in Grade II or above type ectrodactyly?
A) Rearfoot varus posting
B) Forefoot varus posting
C) Heel lift
D) Toe filler

See answer sheet on page 171.

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(1) Each participant achieving a passing grade of 70% or higher on any examination will receive an official computer form stating the number of CE credits earned. This form should be safeguarded and may be used as documentation of credits earned.

(2) Participants receiving a failing grade on any exam will be notified and permitted to take one re-examination at no extra cost.

(3) All answers should be recorded on the answer form below. For each question, decide which choice is the best answer, and circle the letter representing your choice.

(4) Complete all other information on the front and back of this page.

(5) Choose one out of the 3 options for testgrading: mail-in, fax, or phone. To select the type of service that best suits your needs, please read the following section, "Test Grading Options".

TEST GRADING OPTIONS

Mail-In Grading

To receive your CME certificate, complete all information and mail with your credit card information to:

Podiatry Management
P.O. Box 490, East Islip, NY 11730

There is **no charge** for the mail-in service if you have already enrolled in the annual exam CPME program, and we receive this exam during your current enrollment period. If you are not enrolled, please send $20.00 per exam, or $139 to cover all 10 exams (thus saving $61* over the cost of 10 individual exam fees).

Facsimile Grading

To receive your CPME certificate, complete all information and fax 24 hours a day to 1-631-563-1907. Your CPME certificate will be dated and mailed within 48 hours. This service is available for $2.50 per exam if you are currently enrolled in the annual 10-exam CPME program (and this exam falls within your enrollment period), and can be charged to your Visa, MasterCard, or American Express.

If you are *not* enrolled in the annual 10-exam CPME program, the fee is $20 per exam.

Phone-In Grading

You may also complete your exam by using the toll-free service. Call 1-800-232-4422 from 10 a.m. to 5 p.m. EST, Monday through Friday. Your CPME certificate will be dated the same day you call and mailed within 48 hours. There is a $2.50 charge for this service if you are currently enrolled in the annual 10-exam CPME program (and this exam falls within your enrollment period), and this fee can be charged to your Visa, Mastercard, American Express, or Discover. If you are not currently enrolled, the fee is $20 per exam. When you call, please have ready:

1. Program number (Month and Year)
2. The answers to the test
3. Your social security number
4. Credit card information

In the event you require additional CPME information, please contact PMS, Inc., at **1-631-563-1604**.

ENROLLMENT FORM & ANSWER SHEET

Please print clearly...Certificate will be issued from information below.

Name _____ Soc. Sec. #_____
Please Print: FIRST MI LAST

Address_____

City_____State_____Zip_____

Charge to: _____Visa _____ MasterCard _____ American Express

Card #_____Exp. Date_____

Note: Credit card is the only method of payment. Checks are no longer accepted.

Signature_____Soc. Sec.#_____Daytime Phone_____

State License(s)_____Is this a new address? Yes_____ No_____

Check one: _____ I am currently enrolled. (If faxing or phoning in your answer form please note that $2.50 will be charged to your credit card.)

_____ I am not enrolled. Enclosed is my credit card information. Please charge my credit card $20.00 for each exam submitted. (plus $2.50 for each exam if submitting by fax or phone).

_____ I am not enrolled and I wish to enroll for 10 courses at $139.00 (thus saving me $61 over the cost of 10 individual exam fees). I understand there will be an additional fee of $2.50 for any exam I wish to submit via fax or phone.

Over, please

ENROLLMENT FORM & ANSWER SHEET (cont'd)

EXAM #1/09
Ectrodactyly of the Foot (Caselli)

Circle:

1.	**A**	**B**	**C**	**D**	**11.**	**A**	**B**	**C**	**D**
2.	**A**	**B**	**C**	**D**	**12.**	**A**	**B**	**C**	**D**
3.	**A**	**B**	**C**	**D**	**13.**	**A**	**B**	**C**	**D**
4.	**A**	**B**	**C**	**D**	**14.**	**A**	**B**	**C**	**D**
5.	**A**	**B**	**C**	**D**	**15.**	**A**	**B**	**C**	**D**
6.	**A**	**B**	**C**	**D**	**16.**	**A**	**B**	**C**	**D**
7.	**A**	**B**	**C**	**D**	**17.**	**A**	**B**	**C**	**D**
8.	**A**	**B**	**C**	**D**	**18.**	**A**	**B**	**C**	**D**
9.	**A**	**B**	**C**	**D**	**19.**	**A**	**B**	**C**	**D**
10.	**A**	**B**	**C**	**D**	**20.**	**A**	**B**	**C**	**D**

LESSON EVALUATION

Please indicate the date you completed this exam

_____

How much time did it take you to complete the lesson?

_____ hours _____minutes

How well did this lesson achieve its educational objectives?

_____Very well _____Well

_____Somewhat _____Not at all

What overall grade would you assign this lesson?

A B C D

Degree_____

Additional comments and suggestions for future exams:

_____

_____

_____

_____

_____

_____