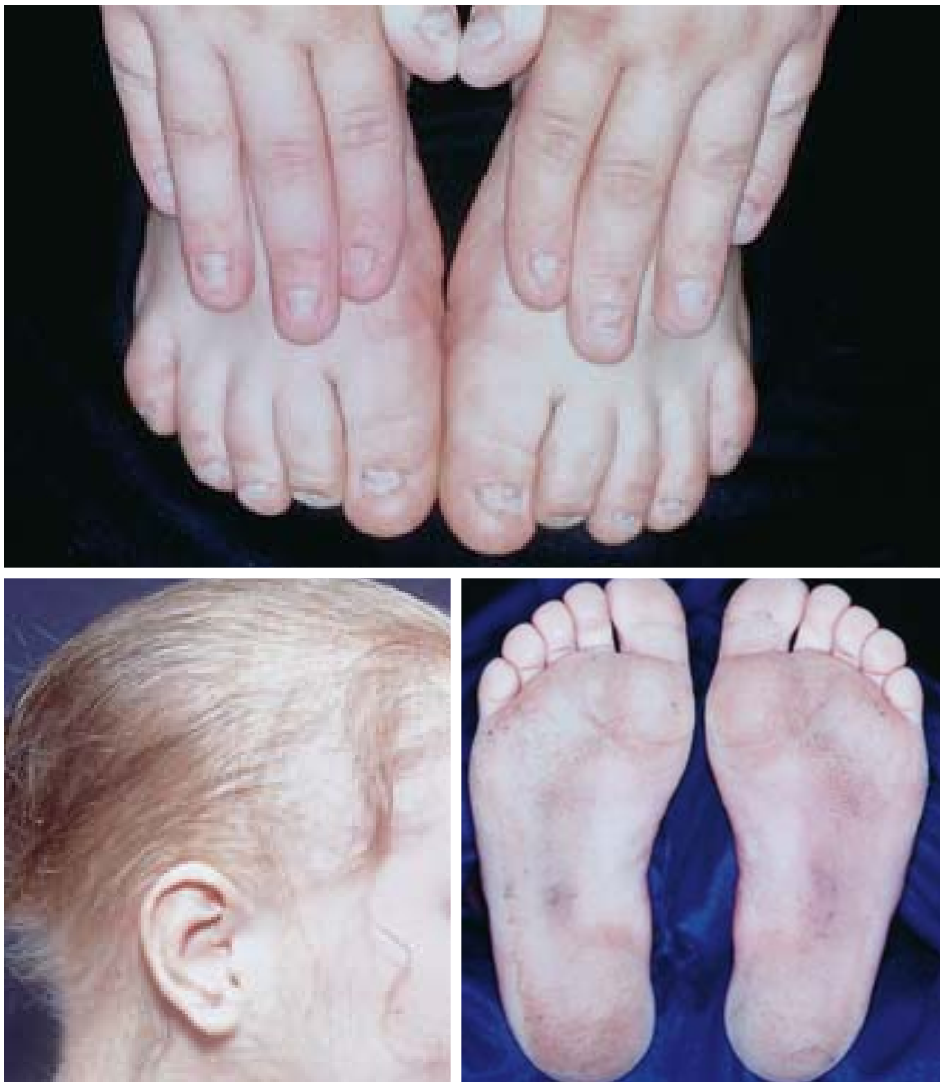
What Is Your Diagnosis?

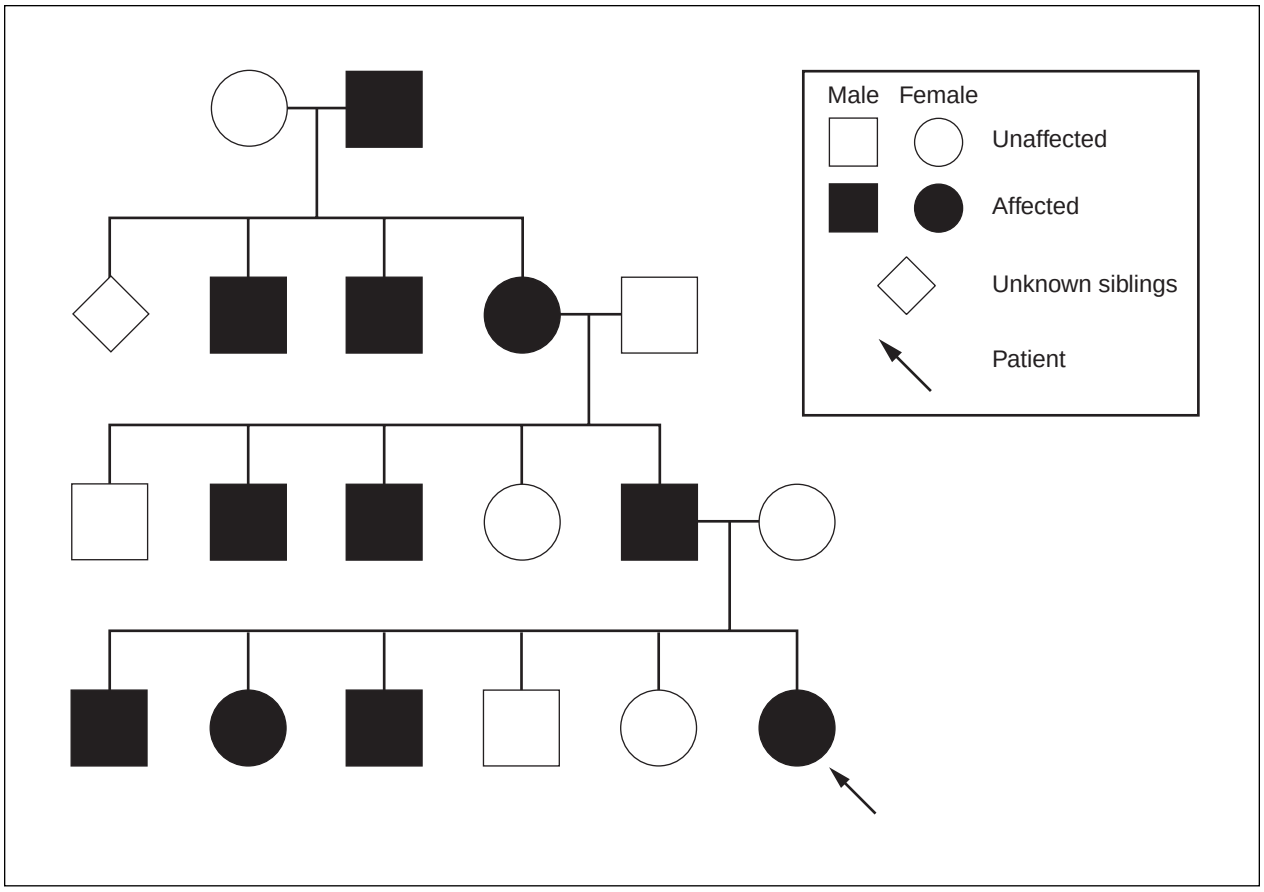


A 20-year-old pregnant woman of French-Canadian ancestry presents with nail dystrophy, sparse hair, and keratoderma since birth. She also reports that many members of her family members have similar features.

PLEASE TURN TO PAGE 224 FOR DISCUSSION

CPT Sidney B. Smith, MC, USA, and CPT John W. Ervin, MC, USA, Department of Dermatology, Wilford Hall Medical Center, Lackland Air Force Base, Texas.

The Diagnosis: Hidrotic Ectodermal Dysplasia



Pedigree shows the autosomal dominant inheritance pattern of hidrotic ectodermal dysplasia.

Our patient of French-Canadian descent presented with the classic triad of hidrotic ectodermal dysplasia, also known as Clouston syndrome. She was born after a full-term, noncomplicated pregnancy to nonconsanguineous parents. Her pedigree shows the autosomal dominant inheritance pattern of this condition (Figure). This condition demonstrates complete penetrance and variable expressivity clinically,[1] which correlates to the patient's description of some of her relatives. Neither the patient nor her family are aware of how many generations in which this condition has been present beyond what is represented in the patient's family pedigree.

In 1929, Clouston[2] originally described hidrotic ectodermal dysplasia in 119 French Canadians. The condition was subsequently given the eponym Clouston syndrome. This rare genodermatosis is characterized by a triad of dystrophic nails, alopecia, and palmoplantar keratoderma. Hidrotic ectodermal dysplasia is predominately associated with French-Canadian families. However, the condition also has been reported in other ethnic groups (Indian, Scottish/Irish, Chinese, French, Japanese, Chinese/Malay, British, African American, and Malaysian).[1-6]

In 1996, Kibar et al[1] mapped the mutation in French-Canadian families to the pericentromeric region of chromosome 13q. Further research has

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located the defect to chromosome 13q11–q12.1. These results also have been confirmed in other ethnic groups, but different haplotypes have been noted among the varying ethnic family lines.[3,4] With the discovery of the chromosomal mutation, prenatal diagnosis may become possible.

In hidrotic ectodermal dysplasia, the nails present with micronychia, partial anonychia, slowed growth, thickening, and brittleness. Frequent paronychial infections can occur, as well as periungual thickening. Scalp hair can be normal early in life but may become sparse and or absent after puberty. As with scalp hair, generalized hypotrichosis can occur in the eyebrows, eyelashes, axillae, genital region, and extremities. Palmoplantar keratoderma is the third aspect of the triad.[1-6]

Other isolated anomalies have been reported in addition to the standard triad, including tufted terminal phalanges, ocular abnormalities, hearing loss, and impaired mental development.[1,5] Squamous cell carcinomas have been reported in the nail plate and on palmar tissue.[6] Patients with hidrotic ectodermal dysplasia experience normal growth and development and have a normal life span. Management of the patient's keratoderma can include treatment with keratolytics or systemic retinoids.[5] Referral to a geneticist for prenatal counseling is advised.

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