

## Patient with “Lobster Claw” Hands and Feet: Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome

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### To the Editor:

Ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome (MIM 129900) is the most common of a heterogeneous group of hereditary diseases in which ectodermal dysplasia and facial clefting coexist. Ectodermal dysplasia is defined as the abnormality of 2 or more organs of ectodermal origin, including hair, teeth, nails, sweat glands, external ear, cornea, conjunctiva, tear ducts and glands, and central nervous system. The 3 most important syndromes with associated ectodermal dysplasia and facial clefting are EEC syndrome, Rapp-Hodgkin syndrome—ectodermal dysplasia, cleft palate or lip, high forehead, midfacial hypoplasia, and small mouth (MIM 129400)—and Hay Wells or ankyloblepharon-ectodermal dysplasia-clefting (AEC) syndrome (MIM 106260).<sup>1,2</sup> The apparent origin of this type of disease lies in mutations of the region of chromosome 3q27 that codes for protein p63—a homolog of the tumor suppressor p53—that regulates ectodermal development.<sup>3,4</sup>

We present the case of a 36 year-old man who was seen for a 3-month history of lesions at the corners of the mouth. Examination of the skin revealed some slightly excrescent, erythematous lesions associated with fissures at both oral commissures (Figure 1). Above the lips, superficial scars ran up on each side to the nasal vestibules.

General physical examination revealed malformations of both hands and feet, with absence of some of the central digits, a midline cleft, and fusion of some of the remaining digits, giving the appearance of “lobster claw” hands and feet (Figures 2 and 3). In addition, there was severe xerosis of the trunk and extremities and the hair was fine and blond. Intense bilateral conjunctival injection was also evident.

When the patient was asked about the lesions he reported no relevant dermatologic family history, only congenital malformations of the extremities. He had also undergone surgical intervention as a child for cleft lip and palate present at birth. In recent years he had been followed up in various

ophthalmology departments for blepharoconjunctivitis, recurrent eye infections, trichiasis, and entropion, for which he had received treatment with artificial tears, corticosteroids, and ophthalmic antibiotics on many occasions.

The lesions at the oral commissures were diagnosed and treated as angular cheilitis in the context of a patient with EEC syndrome.

EEC syndrome is a rare congenital disease of autosomal dominant transmission, first described by Eckholdt and Martens in 1804.<sup>5</sup> In 1970, Rüdiger introduced the acronym EEC (Ectrodactyly-ectodermal dysplasia-clefting), the current name for the syndrome. Patients affected by EEC syndrome characteristically present ectrodactyly (84% of cases)—also known as “lobster claw” hands and feet—which is caused by abnormal development of the hands and feet with an alteration of the central axis of the digits, absence of digits, a deep midline cleft, and fusion of some of the remaining digits.<sup>6</sup> There may also be



**Figure 1.** Lesions compatible with angular stomatitis at the oral commissures.



**Figure 2.** “Lobster claw” malformation of the hands.



**Figure 3.** “Lobster claw” malformation of the feet.

abnormalities of the teeth and hair: hypoplastic or absent teeth, or early loss of the permanent teeth due to caries secondary to hypoplasia of the enamel (77% of patients), cleft lip or with or without cleft palate (68% of cases), and abnormalities of the lachrymal system (59% of cases). All of these were present in our patient. Abnormalities of the lachrymal system consist mainly of atresia of the tear duct and aplasia of the Meibomian glands, with defects in the tear film leading to epiphora, dacryocystitis, blepharitis, blepharoconjunctivitis, recurrent infections, corneal scarring, and deteriorating vision.<sup>6,7</sup> Other less common features include abnormalities of the genitourinary system (hydronephrosis, chronic pyelonephritis, vesicoureteric obstruction, renal duplication or duplication of the collecting system, cryptorchidism, and hypospadias), conductive deafness, facial dysmorphism (broad nasal bridge, mandibular hypoplasia, and pointed chin), strabismus, recurrent respiratory infections, choanal atresia, short stature, and mental retardation.<sup>1,3,8,9</sup>

From a dermatological point of view, patients with EEC syndrome have fine, dry, blond hair, with the occasional presence of *pili torti* (twisted hair) and uncombable hair syndrome. Patients with ectodermal dysplasia and cleft palate can also present dermatitis and folliculitis of the scalp.<sup>1</sup> Other dermatological abnormalities include dry skin, dermatitis at various sites, hypopigmentation, increased numbers of nevocellular nevi, hyperkeratosis of the palms and soles, and nipple abnormalities.<sup>1,3,8,9</sup> We also observed angular cheilitis in this patient—a

finding previously described by other authors.<sup>3</sup> These lesions could be caused by the excess of saliva in the area as a result of anatomical changes secondary to the corrective surgery for the cleft left lip and palate, though bacterial or fungal agents could also be involved.

Although many published reports of the syndrome are of sporadic cases with no family history, prenatal detection of specific chromosomal abnormalities of this type of syndrome marks an important step forward for those patients wishing to have children. Prenatal detection of abnormalities of the p63 gene of chromosome 3 has already been used successfully, and healthy children have been born to parents with EEC syndrome.<sup>4,10</sup>

Management of these patients must be multidisciplinary. This should start with the surgical correction of defects that could cause a functional deficit, such as problems of phonation and hearing due to cleft palate, difficulties walking or handling objects due to ectrodactyly, and abnormalities of the genitourinary system. Odontologic follow-up is also essential, with the correction of poor dental occlusion and caries, and the insertion of implants where necessary. Artificial tears and topical antibiotics may be required to avoid ocular problems, although the onset of ocular complications is sometimes insidious and may require other techniques in order to avoid problems such as entropion or trichiasis.

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